Testing Baby: Parents' Perspectives on Genetic Diagnosis

By

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Abstract

TESTING BABY: PARENTS' PERSPECTIVES ON GENETIC DIAGNOSIS

by

Rachel N. Grob

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This dissertation examines the impact of diagnostic processes on parents, with particular focus on how newborn screening is changing the way they perceive and experience their children's genetic disorders. What is the effect of knowing one's child has such a disorder at birth, and how is this experience different from that of knowing later, after symptoms of the disease emerge? How is it different from knowing earlier, before birth? How is newborn screening changing the relationship, for affected families, between medically-defined "disease" and socially-defined "illness"? In qualitative interviews with parents who learned about their child's genetic disorder in various ways -- i.e., prenatally, through newborn screening, or after the emergence of symptoms -- I have asked how the diagnostic process unfolded for them, and how their lives were changed as a result. I have also asked more specifically about whether and how the diagnostic process influenced their perception of their child; their parenting practices; and their relationships with health-care professionals. My analysis draws heavily on sociology of families, sociology of science, and sociology of health and illness.

Recently, technology has emerged that makes it easy and inexpensive to screen infants' heel blood for large numbers of disorders. Eagerly, and with unprecedented

speed, policy-makers have seized the opportunity to add more and more new conditions to state newborn-screening panels. The manifest function of newborn-screening policy in 2005 is still to prevent disease or, when that's not possible, to lessen its effect. But my interviews with parents highlight many other consequences of testing, including its substantial influence over the entire context of early parenting, over intra-familial relationships, and over the balance of power and the process of collaboration between parents and their health-care providers. My dissertation research illuminates under-examined aspects of the current technological shift in childhood genetic diagnostics; gives voice to a broader range of parental narratives about their experience of this shift than is generally found in either the popular or the scientific literature; and examines newborn screening as an increasingly important aspect of the "new public health" and its prevailing discourse about risk.

Acknowledgements

I have spent much of my time as a sociology doctoral student exploring the many layers of meaning conveyed by one of the discipline's fundamental claims -- i.e., "context matters." This seems such a self-evident observation, yet I continue to marvel at how profound an insight it really is. And never has the structural power of my own life context been demonstrated to me more concretely than during the process of researching and writing this dissertation. If "becoming a sociologist" confers any authority upon me, I use it to assert that I absolutely could not have completed this work without the advice, support and encouragement of the family members, friends, teachers, colleagues and advisors who have formed my own personal context.

Barbara Katz Rothman has been magnificent in every way. Intellectual mentor, practical advisor, fellow mother, friend – Barbara is all of this and more. The only thing I can fault her for is setting me up to receive scowls and jealous vitriol if I tell the truth about my own doctoral experience during those predictable gripe sessions academics hold to re-hash their painful rites of passage. Julia Wrigley and Paul Attewell both gave me advice and ideas with simultaneous respect and rigor, making the oral exam and the dissertation process into something as close to the ideal type as I can imagine.

The mothers and fathers who volunteered to be interviewed for my study gave generously of their time. They also shared intimate feelings, thoughts and ideas. I am very grateful to all of them, and I wish them and their children all the very best.

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proceed; substantive feedback; ideas about what else I should read; and unfailing confidence that I would indeed finish. Amy Barnett's steadfast friendship has helped me in innumerable ways in the general course of day-to-day life,

My friends and colleagues at Sarah Lawrence College have provided me with support of many kinds. Susan Guma, who has been the "good angel" quietly overseeing most major events in my professional life, once again engineered things so I could get this work done. Marsha Hurst, Caroline Lieber, Linwood Lewis and others have spent many hours with me talking through a wide range of conceptual, practical, ideological, and technical issues. Marsha, in particular, has been a smart, challenging, and energetic intellectual partner for more than fifteen years. Mayra Bloom steadfastly communicated her confidence that I would finish and do it well. Vicki Lyus and Karina Achrich provided skillful and patient research assistance of many kinds, and Vicki Lyus and Shawna Irish both did a terrific job transcribing interviews. I want also to thank Barbara Kaplan and Michele Myers for indirectly supporting my work in a variety of ways.

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There, she pampered me in every way, gave me a beautiful space to write in, and took care of all logistical and household tasks so I could focus entirely on writing and on relaxing after a long summer of work. Muchisimas gracious, Lucy.

Andy has been steadfast, patient, and generous – as ever – throughout this process. He never failed to encourage me; to tell me it was fine for me to go write despite the dishes in the sink or laundry in the basket; to talk with me about my latest interview, idea, or epiphany. The intellectual friendship we began nearly twenty years ago at Wesleyan continues to bear precious fruit, held as it is within the larger, multi-dimensional, always-growing framework of our lives together. I know well that the collective energy we had to focus on my dissertation these past eighteen months meant putting on hold other adventures and other priorities. That Andy could make this sacrifice without begrudging it one iota is a true act of love, and also a true testament to his generosity and goodness.

My mother, Naomi, deserves a paragraph all her own. Since this is the only page in the dissertation that will not benefit from her skillful editing, I cannot dare hope it will convey more than a fraction of my gratitude to and love for her – but here goes. Naomi, more than anyone else, truly shared the dissertation process with me. She read chapters as soon as asked; talked with me about them in depth (or sometimes just listened!); told me how interesting, moving and important she found the work; and helped hone the prose in innumerable ways. And she even stuck around for some mean games of "Casino" after our late-night editing sessions. What more could a daughter ask?

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pleasure so enormous that it fuels all other forms of activity, making difficult things not only imaginable but also possible. I dedicate this dissertation to them.

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Chapter One

Introduction

Overview

Within forty-eight hours after the birth of a baby -- before the mother and her family have gotten to know the infant outside the confines of the hospital, and before they have yet learned how to care for this child or gained confidence in their new roles -- the newborn's heel has been pricked and the blood sent to a laboratory that will screen for inherited conditions. Almost always without parental consent, and usually without parental knowledge or understanding, the newborn's genetic makeup is analyzed for molecular clues to her future. Some of these tests provide information that can and must be acted on quickly to protect the infant's health or even to save her life. Other tests identify mutations that may in fact never cause manifest signs or symptoms. Still others find mutations that will result in serious or fatal disease for which there is no cure. And many provide information about what, for the time being at least, are genetic abstractions -- traits that have not yet been expressed and will not result in recognizable effects for months or years.

This dissertation examines the impact of diagnostic processes on parents, with particular focus on how newborn screening is changing the way they perceive and experience their children's genetic disorders. What is the effect of knowing one's child has such a disorder at birth, and how is this experience different from that of knowing later, after symptoms of the disease emerge? How is it different from knowing earlier, before birth? How is newborn screening changing the relationship, for affected families, between medically-defined "disease" and socially-defined "illness"? In qualitative

interviews with parents who learned about their child's genetic disorder in various ways, I have asked how the diagnostic process unfolded for them, and how their lives were changed as a result. I have also asked more specifically about whether and how the diagnostic process influenced their perception of their child; their parenting practices; and their relationships with health-care professionals.

Recently, technology has emerged that makes it easy and inexpensive to screen infants' heel blood for large numbers of disorders. Eagerly, and with unprecedented speed, policy-makers have seized the opportunity to add more and more new conditions to state newborn-screening panels. There are testing innovations in development which would increase again, many fold, the already-expanded capacity to test. More and more parents are thus getting more and more genetic information about their child right after birth. At the same time, the implications of this genetic information are increasingly complex and ambiguous. This is because states are now testing for more and more conditions that are not clearly treatable, and/or are not predictable either in terms of when (if ever) in the child's life symptoms might emerge, or of how serious those symptoms might be if and when they do emerge.

Now is an opportune moment, then, to ask hard questions about the impact of newborn-screening technology, and about the social policies that guide its use. As Katz Rothman wrote of prenatal testing when those procedures were just becoming widespread, "Technological changes can force us to confront questions we never before faced, to see ourselves and each other in new ways" (1986, 3). My dissertation research is designed to illuminate under-examined aspects of the current technological shift in childhood genetic diagnostics, and to give voice to a broader range of parental narratives

about their experience of this shift than is generally found in either the popular or the scientific literature.

The remainder of this chapter describes the sociological perspectives I have drawn on in constructing my study and analyzing my data; provides background on newborn screening in the United States; places my research within the existing literature; and outlines my research design and methodology.

Sociological Context

This dissertation examines the impact of diagnostic processes on parents largely from the standpoint of parents themselves. My primary data are drawn from twenty-five qualitative interviews with mothers and fathers whose children were diagnosed with cystic fibrosis (CF) either prenatally; by newborn screen; or after symptoms of the disorder emerged. To my knowledge, no published study in the rapidly-growing literature on newborn screening has attempted to understand either the actual *experience* parents have had with the process, or how that experience – which is now becoming more and more common -- differs from the experiences parents have had as they negotiated other diagnostic processes. As political scientist Diane Paul affirms, citing legal scholar Ellen Clayton, "We know little in general about how screening 'programs actually feel to those they touch'" (Paul 1998, 179 including quote from Clayton 1992b, 94). I designed my study to begin filling this gap. In so doing, one of my central commitments has been to listen to and make public the voice of parents who – along with their children -- are the objects of what I argue to be a new form of medicalization, but whose responses, insights,

and feelings have been largely overlooked as a legitimate source of data about newborn screening.

Drawing on the literature of "illness narratives" within medical sociology and anthropology, I have approached my subject with the assumption that there are crucial differences between CF as a *disease* defined by the diagnosticians and treatment-specialists who classify and manage health care for affected children, and CF experienced as an *illness* by parents of the young children who are diagnosed (Kleinman 1988). CF is a complex disorder that expresses itself in diverse ways: symptoms can begin at birth, sometime in the first few months or years of life, or not until much later; it can be debilitating, very mild, or may cause only infertility (in men). In some cases, people have no symptoms whatsoever. Furthermore, the various ways parents now get a CF diagnosis result in a number of different temporal relationships between their learning of the disease as a genetic fact and the onset of symptoms. Yet, in the face of these complex variations in the disease itself and in parents' experience of it, professionals offer parents highly standardized information and highly standardized treatment protocols. Ironically, they do this in part because the very complexity of the disease makes its course difficult to predict. Diagnostic experiences with CF thus offer a rich arena for investigating the relationship between medically-defined disease and the actual experience of CF by parents of genetically-diagnosed children.

Sociologist Arthur Frank writes that "Parson's modernist 'sick role' carries the expectation that ill people get well, cease to be patients, and return to their normal obligations" (1995, 9). But in postmodern times, he continues, the line between "sick" and "well" is much more complex. Frank's own work focuses in particular on the roles

and stories of people who are effectively well, or at least able to function on a day-to-day basis, yet "could never be considered cured" (Ibid., 8). He uses the term "remission society" to describe this group of individuals, along with the family members who share their struggles, and he argues that modernist medicine lacks a story appropriate to describe the experiences of its members. I approached my study with the intention of understanding the parents of children diagnosed with CF as another subgroup of people who are profoundly affected by disease, but who do not fit the classic modernist "sick" role – in this case both because they are caregivers rather than persons directly affected, and because the meanings of the diagnosis are so complex, varied, and unpredictable. I embarked on my interviews with the expectation that these parents would have their own narrative accounts of what the diagnosis has meant to them, and that they would be generous enough to share these accounts with me. As I hope the chapters that follow illustrate, I was not disappointed.

Although my primary objective has been to understand *the perspective of parents* on diagnostic processes, and although I have used a grounded-theory methodology for analyzing and categorizing my data (see below), I have by no means approached the topic as a "blank slate" on which I could inscribe the stories told to me as individually-constituted truths. Like Kleinman and Kleinman, I view experience itself as intersubjective, as "constituted in social space," rather than as a product of individual subjectivity. Experience is itself "the outcome of cultural categories and social structures interacting with psychophysiological processes such that a mediating world is constituted" (as quoted in Kleinman and Seeman 2000, 234). As such, no meaningful sense can be made of it in the absence of *context*, or of an appreciation for the larger

social, cultural, and political influences that shape that context in any particular time and place.

I am indebted to several bodies of literature within my field for providing me with the tools to create a coherent context for my inquiry, and to ground my analysis of primary data – once I obtained and began trying to make sense of them -- within useful "cultural categories" and "social structures." I drew heavily on these traditions as I shaped my research questions and designed my study: how could it be otherwise, since I myself am fully situated within my own social context as an emerging sociologist? At the same time, I tried *not* to let my own theoretical biases over-determine either how I conducted the semi-structured interviews, or how I categorized and interpreted what parents had said once I began my analysis. What follows is therefore a description both of approaches in sociology that helped shape my work from the outset, and of perspectives that have emerged -- through my data analysis -- as recognizable and important frames for the collective experiences parents recounted.

Sociology of Families: Parenting and the "Baby Experts"

My dissertation follows the sociological tradition of studies looking at how the experience and practice of parenting are socially constituted by both material and discursive influences shaping the larger environment in which families' lives unfold. This tradition has repeatedly demonstrated the historically contingent nature of the parent/child relationship, and its susceptibility to change in the face of social transformation, new technology, and changing policy arrangements (Arias 1962; Hochschild 1997; Popenoe 1988; Wrigley 1989, 1995; Hays 1997; Stearns 2003; Hulbert 2003). My own research brings this theoretical perspective on the socially-constituted

nature of parenting -- and the important questions that it enables us to ask about the qualitative impact that health system practices have on families -- to a substantive arena in which such perspectives have to date been largely lacking: i.e., the impact of diagnostic processes on parents and parenting.

The strand of parenting literature within sociology that has been most useful to me focuses on the role of professional expertise in shaping ideas about and approaches to raising children. Social scientists have examined extensively the changing influence, over the last hundred years and more, of "baby experts" -- those outside authorities (usually men) who make it their business to tell parents (usually mothers) what to do and how to do it (Ehrenreich and English 1978; Crane 2000). Since the colonial era, they note, child rearing in the United States has been a concern of political and religious leaders alike. It was not until close to the turn of the last century, however -- with large-scale migration to the cities and the "discovery" of the child as a central actor in the family drama -- that the long-standing "normative tradition of communal concern" about child-rearing was transformed into a distinct social problem to be tackled by social reformers (Halpern and Weiss 1991; Ehrenreich and English 1978). During this period, the unfolding "century of the child," expert advice to parents blossomed in quantity, variety, and influence. Child rearing was quickly converted from instinctual female knowledge -- "… the irreducible core of woman's existence, the last refuge of her skills…" (Ehrenreich and English 1978, 173) -- to a new arena for the exercise of professional authority.

Sociological studies looking at the impact of expert advice on parents have taken up a broad array of specific issues. These include, among many others, the changing view of

children's cognitive development (Wrigley 1989); the medicalization of motherhood via advice from physicians (Litt 2000); and the advent of child-centered, emotionally-involving, and time-consuming "intensive mothering" as an ideology (Hays 1997). Most relevant to my study is the rich literature that investigates the impact of prenatal testing on the experiences of pregnancy and of mothering that was pioneered by Katz Rothman (1986), and subsequently taken up by others (see for example Rapp 2000; Lippman 1991). I have drawn heavily on this work as I conceptualized and designed "Testing Baby," and as I analyzed my primary data.

I have found little in the parenting literature, however, that focuses on the influence of genetic testing beyond the prenatal period. Nelkin, in an article entitled "The Social Dynamics of Genetic Testing: The Case of Fragile-X," comes closest: she analyses, cogently, the influence on parents of messages about genetic testing that began to pervade the parenting literature in the late 1980s, and of the concomitant rapid expansion of genetic screening and testing programs for children in school and health-care settings. "Most of the parent advice literature of the post [World War II] period emphasized the crucial role of the social and family environment in shaping the child," she writes. "But this perspective changed during the 1980s, when parents were inundated with messages stressing genetic determinism." (1996, 544). The new focus on the importance of genetic endowment, she asserts, moved nature to center stage in the nature/nurture debate, and promised parents that "research will give us an 'early peek at genetic endowments'" (LaForge 1991 as quoted on p. 544). Nelkin proceeds in the remainder of this article, as well as in other work (1994; 1995), to examine how messages about diagnostic technologies "exploit [parents'] hope of controlling frightening conditions, of

predicting and eradicating risk" (1996, 546) despite the fact that for many genetic disorders there is no effective treatment. At the same time, she illustrates how in fact "economic and entrepreneurial forces are driving the development and use of emerging predictive tests" (Ibid., 547).

My review of the newborn-screening literature (see below) prior to designing my study certainly suggested to me that Nelkin's general critique of diagnostic testing for children had bearing on the specific procedure of newborn screening as well, though it is not one she herself examined. And as I show in the chapters that follow, the theme of control – the hope of gaining it with respect to CF, and the guilt and devastation that ensue when control feels elusive -- arose again and again in my interviews.

Sociology of Science and of Health and Illness: Diagnostic Procedures in the "Age of Risk"

My dissertation also fits solidly within the sociological tradition of studies examining how science and technology shape human experience in new ways (e.g., Harding 1986; Fox Keller 1985; Latour and Woolgar 1979; Petersen, ed., 1984; Barnes and Edge 1982). Social constructionist perspectives on science have demystified the notion of objectivity, and made transparent the processes by which scientific "facts" are produced and come to be taken for granted. Berger and Luckmann's pioneering work in the sociology of knowledge laid the ground for much of this work. Particularly influential, I would argue, were their insights about the way a social phenomenon can come to "confront the individual as an external and coercive fact" (1966, 58) through processes of institutionalization (i.e., "typifications of habitualized actions that are shared by various actors" (Ibid., 54)), and reification (i.e., "an extreme step in the process of objectification, whereby the objectivated world loses its comprehensibility as a human

enterprise and becomes fixated as a non-human… inert facticity" (Ibid., 89)). Their conviction that all knowledge must be examined as a social and historical product, and their invitation to other sociologists to take up this project, have helped inspire a range of studies.

Most relevant to my project are those social-constructionist studies which have made health-care practices their object. These undertake to deconstruct, to one degree or another, the processes of instutionalization and reification that have led to broad acceptance of various aspects of health, illness, and/or health care as immutable "facts." The traditional view of medicine "sees disease as being located in the body as a physical object of physical state that can be objectively identified and treated as a physiological condition by scientific medical knowledge" (Lupton, 2000, 50). In contrast, social constructionists view health, illness and health care as "sociocultural products," and make it their work to "analyze the nature of their social and cultural representations and the symbolic meanings that surround them" (Ibid.). This rich sub-field within the sociology of health and illness encompasses scholarship on a broad range of issues such as – to name just a very few -- the categorization of people via psychological diagnoses (see for example Szasz 1961; Rieff 1966; Scheff 1999); the conflation of health with masculine views of the ideal female body (Bromberg 2001); and the commodification of pregnancy and motherhood (Katz Rothman 1986, 1989; Ehrenreich and English 1972).

The specific critique, within sociology, of genetics -- viewed both as an ideology and as a medical discipline -- has grown in leaps and bounds in recent years as the medical paradigm has itself shifted towards increased reliance on genetic understandings of health problems and risks (see for example Katz Rothman 1998; Cunningham-Burley 2000;

Duster 2003; Petersen and Bunton 2002; Conrad 2001; Marteau and Richards, eds., 1996; Hubbard and Wald 1999; Hubbard and Lewontin 1996; Rapp 2000; Kerr and Shakespeare 2002; Paul 1997, 1998; Condit 1999). Social scientists have, for example, analyzed at length how genetic explanations of human health and behavior tend to oversimplify complex and highly variable phenomena by implying that there is a simple correlation between the DNA sequence of a gene (genotype) and noticeable traits (phenotype). This emphasis on genes as predictors of disease draws attention away from the importance of interactions between genes and the environment, researchers have argued, and downplays the environment's role in mediating human health. Social scientists have also examined at length the relationship between the genetic paradigm and what is often referred to as the "new eugenics" – i.e., the effort to manipulate and control "what kinds of people we will permit to be born" (Katz Rothman 1998, 177).

As I demonstrate in the chapters that follow, I have found many sociological insights that appear over and over again in the above-referenced literature to be useful in understanding the impact of diagnostic processes on parents. One such insight is that *genetic diagnoses themselves have considerable power to socially construct interpersonal realities* – in this case, as the central findings of my study illustrate, those interpersonal realities are parents' perceptions of their children, and their practices and experiences as care-givers. Another is that micro-processes governing the structure of health care are generally organized around the bureaucratic imperatives of large-scale systems rather than around the needs and wishes of patients. A third is that the relationships among patient- or health- "education," informed consent, and the systemic imperative to induce patients to comply with a given program are complex and vexed.

Simplistic accounts of autonomous, rational subjects making informed decisions based on perfect information certainly have little reality when applied to the complicated arrangements that currently govern "decision-making" about diagnostic tests for CF.

Like others who take a social-constructionist approach to examining specific phenomena relating to health, illness, and the structure of service systems (see for example Lupton 1995; Petersen and Bunton 2002; Petersen 1998; Petersen and Lupton 1996), I have found that poststructuralist theories have much to offer my analysis. Particularly salient is the work of Michel Foucault (1973; 1978; 1979), which I have used here to help make sense of how power relationships are embedded in the micro-practices of public health today. Foucault's studies of how specialized knowledge emerged in the nineteenth century emphasize the role of expertise in making individuals active participants in the prevailing order – i.e., "governable subjects." In his view, power is diffuse and omnipresent, a medium that we all live in rather than something possessed by professionals and used coercively to control subjects. "We must cease once and for all," he writes,

> to describe the effects of power in negative terms: it 'excludes,' it 'represses,' it 'censors,' it 'abstracts,' it 'masks,' it 'conceals.' In fact, power produces; it produces reality; it produces domains of objects and rituals of truth. The individual and the knowledge that may be gained of him belong to this production" (1977, 194).

Certainly the direct use of state authority to mandate newborn screening without parental consent is one aspect of the newborn-screening experience that has consequences for families, and I explore these in the dissertation. But perhaps even more central to the testing process today is the way it contributes to the "production of selves" who view the drive for control and predictability as possible, desirable, and even in some

sense obligatory. The endless processes of self-improvement and self-monitoring that individuals now embrace in their effort to gain control were named by Foucault "technologies of the self." As Lupton describes it, he viewed these as involving "the voluntary internalization of norms governing appropriate behaviour in the interests of achieving the best possible self, including the quest for self-knowledge, self-mastery, and self-care" (Lupton 2000, 57, drawing on Lupton 1995 and Rose 1996). Rose, building on Foucault's work, describes the modern "means of governing the human being" as creating our very identities.

> [O]ur very experience of ourselves as certain sorts of persons – creatures of freedom, of liberty, of personal powers, of self-realization – is the outcome of a range of human technologies, technologies that take modes of being human as their object. Technology, here, refers to any assembly structured by a practical rationality governed by a more or less conscious goal. Human technologies are hybrid assemblages of knowledges, instruments, persons, systems of judgment, buildings and spaces, underpinned at the programmatic level by certain presuppositions and objectives about human beings (1996, 26).

Recent studies of the turn-of-the-twenty-first-century public health apparatus – the "new" public health, of which the newborn-screening programs I examine in this dissertation are a part – have drawn heavily on poststructuralist theory to analyze and deconstruct its techniques of governance. As Petersen and Lupton put it:

> The new public health is, if nothing else, a set of discourses focusing on bodies, and on the regulation of the ways in which those bodies interact within particular arrangements of time and space. Perhaps less obviously, the discourses of the new public health also seek to transform the awareness of individuals in such a way that they become more self-regulating and productive both in serving their own interests and those of society at large. By providing norms by which individuals are monitored and classified, and against which individuals may be measured, the emphasis of the new public health is upon persuading people to conform voluntarily to the goals of the state and other agencies. (1996, 12)

A critical element in this system of monitoring and regulation, sociologists studying the new public health have argued, is the constant awareness and calculation of risk. When viewed as an element of a normative discourse, risk assessment is more than an objective calculation of the chances a health problem will or will not arise. Rather, it is a highly relative and contextualized process of selecting certain dangers as worthy of energy and attention while relegating others to the margins of public consciousness. In the new public health, risk is something individuals are responsible to continually assess and – to the degree that it is possible -- manage. Risk is thus less an external threat that is beyond our control than an *internal* threat that we have a moral obligation to be informed about, and to address through behavior change (Lupton 1995, 1999, 2001; Davison 1996; Kerr and Shakespeare 2002; Petersen and Lupton 1996). When viewed from this social-constructionist perspective, the purpose of analyzing discourses about risk is not to determine which are "real" and which "fake," but rather, in Lupton's words, to understand:

> What statements are used to construct certain kinds of knowledge about risk at a particular historical moment and sociocultural setting? What rules prescribe certain ways of talking about risk and exclude other ways? What types of subject are constructed through risk discourses? How does knowledge about risk acquire authority, a sense of embodying the 'truth' about it? What practices are used in institutions and by individuals for dealing with the subjects of risk discourses? (Lupton, 1999, 33, adapted from Hall 1997, 45-6).

Much social-science critique of the new public health focuses on how it obscures structural threats to human well-being, such as environmental degradation and institutionalized poverty, by emphasizing instead the health risks we incur through "lifestyle choices" (Becker 1986; Petersen and Lupton 1996; Lupton 1995, 1999). The dominant risk discourse teaches its subjects that good health and long life are within

reach for those who eat well, exercise, refrain from smoking, get regular check-ups and screenings. It thus encourages, with a vengeance, the proliferation of "technologies of the self" designed to improve one's own personal "risk profile." Strict adherence to prescribed "self-care regimes" have come to be understood as "the only real means of avoiding cancers, heart diseases and other afflictions that constantly threaten the integrity of the self in a generalised climate of risk" (Petersen and Lupton 1996, 23).

I have brought this same analysis of risk to bear on a different micro-practice within the vast terrain of the new public health: diagnostic testing of children. I have embarked on my study with full appreciation for the lives that have been saved and the suffering that has been averted as a result of these testing programs: it has never been my intention to gather evidence to the contrary. However, like Nelkin and Paul,[1] I have approached my topic with skepticism about the assumption that the utility of diagnostic tests is straightforward, and their widespread use an unmitigated good. The tests are represented as objective and scientific; the technologies that underlie them are represented as creating direct access to hidden information about human health and well-being. The risks of failing to know, from birth, what genetic disorders may threaten one's new baby are represented as disastrous, while the risks of finding out everything technology can reveal about that child beginning when she is one day old are represented as virtually non-existent. But I began with the assumption that this construction of newborn screening – which I explore in more detail just below -- is an over-simplification, and that from my interviews with parents who have experienced diagnostic tests would emerge a more complex reality.

[1] I refer to these two because they are the only social scientists I have identified in the literature who appear to have focused specifically on diagnostic testing of newborns or children.

Newborn Screening: What Is It, and Why Is It Done?

Newborn screening, defined as biochemical testing for inherited disorders by analysis of the infant's blood, began in the mid 1960s when Dr. Robert Guthrie developed a laboratory procedure that rapidly advanced screening capacity for phenylketonuria (PKU), a metabolic disease that causes severe mental retardation if untreated but can be effectively controlled with a phenylalanine-restricted diet if the diet is introduced soon after birth (American Academy of Pediatrics (AAP) 2000; Therrell 2001; Secretary's Advisory Committee on Genetic Testing (SACGT) 2000). Children's advocacy groups, such as the Association for Retarded Citizens and the March of Dimes Birth Defects Foundation, successfully advocated alongside Guthrie – who himself had a family member with the disorder -- to make the new PKU-testing procedure mandatory for all newborns. Within a few years, almost every state had passed a law to establish compulsory screening at birth, and the heel stick became a routine part of well-baby care in hospitals. Today more than four million infants undergo testing every year, making newborn screening the single most widely-utilized form of genetic testing in the United States (Paul 1999; Hiller and Landenberger 1997; AAP 2000).

Although the federal government has supported newborn screening since its inception with guidance, research, and funding, there is no national policy. The programs are governed at the state level, and there are substantial differences among them with respect to nearly every variable. To cite a few: the actual number of tests currently performed ranges from eight to more than forty;[2] mechanisms for making decisions about which

[2] I arrived at these numbers using data published on the National Newborn Screening and Genetics Resource Center's web site (http://genes-r-us.uthscsa.edu) as of June, 2005. It is likely, however, that my

tests to include vary widely, as do provisions for citizen participation in the process; laboratory capacity, technology and standards are inconsistent; provisions about parental education, informed consent, follow-up counseling, medical care, and ongoing services for affected children and their families are all state-specific and vary widely (AAP 2000; ACOG 2003; CDC 2004; Hiller and Landenburger 1997; Gollust et al. 2005; GAO 2003; NYSDOH 2005; Mandl et al. 2002; Hoff and Hoyt 2005).

With relatively little public attention or debate (Clayton 1992a), newborn genetic screening has pioneered and is now expanding new terrain for American medicine and public health: *state-mandated diagnosis of non-infectious disease*. In an effort to provide guidance to states and to make the policy process more consistent and evidence-based, the World Health Organization, the National Academy of Sciences, the Institute of Medicine, the American Academy of Pediatrics, and various national task forces have developed iterative sets of recommended guidelines for newborn-screening programs since as early as 1968, often at the request of Congress (ACMG 2005; Downie and Wildeman 2001; GAO 2003; AAP 2000; Clayton 1992b; Therrell 2001). Over time, these bodies have become more specific in their analyses and have taken into account the impact of ongoing technological changes. But the core criteria for testing and the core protections they have recommended remained fairly constant until just this year (see below). First and foremost, they held that the screen must be clearly beneficial to the affected infant. Further, they specified that conditions should be tested for only if they meet all of the following conditions: if they are relatively prevalent; if effective treatment exists; if the technology can identify the disorder with sufficient accuracy; if early

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numbers are inaccurate even as a snapshot of summer 2005, because as I write states are adding new conditions to their panels with remarkable speed. As recently as 1995, the number of conditions screened for across the states was between three and nine – a dramatic difference in ten years' time.

detection and treatment are necessary to prevent substantial harm; if onset of symptoms is early; and if facilities for diagnosis and treatment are available (Therrell 2001).

The latest in this series of reports was released just earlier this year (2005) by the American College of Medical Genetics (ACMG), which was commissioned by the federal Department of Health and Human Services to analyze the scientific literature on the effectiveness of newborn screening; gather expert opinion on which conditions should be screened for; and recommend a uniform screening panel that could be adopted by all states (ACMG 2005, 7). In its report, ACMG recommends that this uniform panel include fifty-four conditions: twenty-nine in the "core panel," and an additional twenty-five "secondary targets" which would be reported to physicians and families because they would inevitably be identified as part of the differential diagnosis for conditions on the core panel. This report breaks new ground in newborn screening by officially advocating that all states test for a broad range of specific genetic mutations. Notably, it also departs from earlier standards by adding "benefits to family and society" as a core criterion for assessing whether a specific screen should be mandated. If this change in criteria is widely accepted, as it appears it has already begun to be, it will make it even easier for states to screen for conditions that *do not directly benefit* the infant, but are appealing for other reasons such as cost savings for the public, and receipt by parents of genetic information that can "guide future reproductive decision-making" before other babies are conceived (ACMG 2005; Presentation by Therrell at SUNY Albany School of Public Health, July 2005).

Historically, the recommendations of these sequential reports have had relatively little impact on newborn-screening policy: the reality of the program-development process is

that it has remained haphazard within states, and not at all uniform among them (Goodwin et al. 2002; Atkinson et al. 2001; Hiller and Landenburger 1997; Holtzman 1991; Stoddard and Farrell 1997). In the absence of a widely accepted framework for guiding decisions, the policy process has been susceptible to states' efforts to protect economic interests by identifying genetic disorders early and therefore "influencing" future reproductive decisions and preventing the births of additional affected children. Evidence of the state's intent to manipulate procreative decision-making is not hard to find. In policy-making contexts, there are frequent and explicit references to detecting families at risk in order to deliver timely genetic counseling services that may be "of importance in preventing the birth of more affected children in the same family" (Dankert-Roelse et al. 1987, 271; Plauchu et al. as quoted in Clayton 1992b, 102). Moreover, when researchers analyze "outcomes" related to the first child in any family to be diagnosed with a genetic disorder, they place disproportionate focus on future reproductive decision-making (Mischler et al. 1998; Wertz et al. 1992; Ciske et al. 2001). And despite explicit objections on the part of the public to the formation of testing policy on the basis of cost/benefit analyses (Green 1992, 318), proposed expansion of newborn-screening panels is always considered in actuarial terms. [3]

The absence of a uniform framework for newborn-screening laws also leads to what John McDonough has characterized as "the misuse of anecdote" in policy making – i.e., the development of large-scale policies in response to individual narratives that may or may not accurately characterize the experience of a larger group (McDonough 2001).

[3] See, for example, Filiano et al. 2002, who conducted a study comparing the costs of late vs. early diagnosis (by tandem mass spectrometry) of a genetic disorder called propionic acidemia. They document states' interest in the cost-effectiveness of screening tests, and conclude that "just as with the earlier experience with newborn screening, the monetary benefits of expanded newborn screening will outweigh the monetary charges" (204). See also Venditti 2003.

Despite guidance advising that procedural, ethical, and quality assurance considerations should factor heavily in screening-policy decisions, states have largely gone their own way in terms of how they expand their panels, and what they choose to include and exclude (Wilfond 1995; Wilfond and Nolan 1993; Stoddard and Farrell 1997). And just as parents of children with PKU were instrumental in establishing newborn-screening programs to begin with, so family members of children with various screenable conditions have continued to play a prominent role in the programs' expansion (Therrell 2001).

Another factor that has significantly influenced state newborn-screening policy is the availability and affordability of various screening technologies. Until the 1990s, laboratory analysis was conducted by "growing out" bacteria in blood that was placed on filter paper after being taken from the baby's heel, or by other methods of identifying evidence of abnormalities one by one (ACOG 2003). Although additional tests can be and are run on the same few drops of blood that have routinely been collected from every newborn since the 1960s, this form of "piggy-backing" is relatively cumbersome since each new condition requires an additional step in the analysis process – its own test, in essence. With the advent of a new technology for screening, tandem mass spectrometry (also known as MS/MS), testing entered a whole new era. Now, one sample of blood can be analyzed simultaneously and at very low cost for more than thirty distinct gene mutations. By making it easy and cheap to screen for tens of new conditions, the new technology has shifted – as technology inevitably does[4] -- the terms of policy discussions. Although advocates of a more cautious approach to program expansion point out that

[4] For social science and historical explorations of the relationship between technology and society, including arguments about technological determinism, see Winner, 1977; Smith 1994.

there are no effective treatments for a good many of the disorders identified by MS/MS, and that the *capacity* to screen does not imply an *obligation* to screen (Wilcken 2003), the technology has without question been a primary catalyst for increasing the size and scope of screening panels in recent years.

The long-predicted rapid expansion of newborn screening is now a reality. More than forty states have now begun using MS/MS (http://genes-r-us.uthscsa.edu, January 2005), and, remarkably, in the one year that has elapsed from the time I designed my study in 2004 to the time of this writing in 2005, the average number of conditions screened for across the states has risen from an estimated average of *eight* to an average of *twenty* (Hoff and Hoyt 2005; http://genes-r-us.uthscsa.edu). In my own state of New York, the number of conditions has increased from eleven in 2002 to forty-four today, with two more set to be added in the fall of 2005 (Baker 2004; Newborn Screening Program Staff 2005). The gate opened by legislation mandating PKU testing without parental consent has recently become a floodgate, a flood of parentally-unsought information at the birth of each baby. And researchers and state newborn-screening directors are already looking forward to the coming DNA "chip," the new technology that will supersede MS/MS and be capable of identifying ten thousand alleles – ten thousand "conditions" (or perhaps just variations) that each baby carries in her genetic make-up (Wilcken 2003; ACOG 2003; Lloyd-Puryear and Forsman 2002). This technology -- not yet here, but confidently anticipated as was MS/MS just a few short years ago -- will make it even easier and cheaper than it has already become to obtain genetic information about newborns at birth.

Representations and Research: Newborn Screening in the Popular, Scientific, and Social Science Literature

Newborn screening has been the focus of much research, commentary, and media attention, especially over the last several years -- since the March of Dimes launched a major public campaign advocating screening-program expansion.[5] Some stories on the topic in the popular media try to present a balanced view of the complex issues raised by expanded newborn screening. For example, Gina Kolata of the New York Times reports that "while no one argues with the idea of saving babies, [ACMG]'s proposed [uniform panel of 29 diseases] is generating fierce debate" because experts dispute both "whether treatments help," and "how often a baby will test positive but never show signs of serious disease" (2005, 1). The web site www.kidshealth.org also lays out the issues in a balanced (if necessarily simplified) way, with a summary of the benefits and drawbacks of expanded newborn screening, and advice to parents about how to learn about screening processes in their own state.

However, the majority of articles in the popular press and the parenting literature, as well as on the web, are advocacy pieces that call for screening programs to be expanded, or that decry their existing limits. Most often, these articles use first-hand stories about or by affected families to add emotional emphasis (see for example Edelson 2003; Graham 2003; Nawn www.savebabies.org/familystories accessed 2004; Oliver 2002; Stahl 2003; Hehmeyer 2001). They begin with a heart-wrenching story about a baby whose death could have been prevented by newborn screening, or a heart-warming story

[5] To my knowledge, nobody has yet undertaken a systematic study of how newborn screening is represented to the public by the media. Katz Rothman, Hackett and I have developed a methodology for conducting formal research in this area, and are currently seeking funding so that we may proceed. In the meantime, I present here my impressions based on a more informal review of media articles, and on conversations with other researchers immersed in the newborn-screening literature.

about a baby who was saved by it, and then go on to talk about the failure of states to protect the public by expanding their mandatory panels. This introductory text from a recent piece put out by the Associated Press, entitled "Parents Seek Expanded Newborn Screening," is illustrative:

> Debra Gara held 9-month-old Cristal in her arms, singing her to sleep, and then dozed off herself. An hour later, she awoke with a start to find her baby ice-cold and not breathing. An autopsy diagnosed a rare metabolic disease, one treatable if Cristal's parents had known – and one of more than 40 genetic and metabolic disorders that can be diagnosed easily at birth (Associated Press, www.intelihealth.com, accessed 10/31/04).

Headlines of similar articles proclaim, with little content to balance the bold claims in the lead lines, that "early screening will save lives"; that "few states offer adequate newborn screening"; that "early tests can reduce later problems" (www.marchofdimes.com; www.greenvilleonline.com 1/16/03; www.TheChamplainChannel.com, 8/27/03). Conrad's conclusion that the media's overall coverage of genetics is disproportionately focused on success stories without counterbalancing examples of "failures" or "disconfirmations," and that the media therefore "reinforce[s] the idea that genetics can explain more of the world than it can" (1997, 149), appears to hold true for press coverage of newborn screening specifically.[6]

The medical, public-health, and public-policy literatures also tend towards a favorable view of newborn screening, and towards endorsing screening-program expansion (see for example Goodwin et al. 2002; Wright et al. 1992). However, a number of problematic issues have been and continue to be raised by practitioners, researchers, and analysts in

[6] For an excellent analysis of how the press treats issues of science and technology in general, as well as genetics in particular, see Nelkin, 1995, Selling Science: How the Press Covers Science and Technology.

these fields. One recurrent worry is that children who are being promptly diagnosed by newborn screening don't then have adequate access to follow-up services and ongoing treatment, since the mandatory diagnosis is not complemented by a guaranteed right to health care. Other concerns are that states have inadequate capacity for genetic counseling and health education in connection with the screening process; that the practice of testing without parental consent is problematic, especially as state screening panels expand; that the genetic information gathered by newborn screening is not sufficiently protected with respect to either insurance coverage or confidentiality. Critics also question the ethics and legality of screening since this form of mandatory testing acts as a back door for giving parents unsought (and potentially unwanted) genetic information -- information they might or might not voluntarily consent to obtain in the form of prenatal testing -- before they plan or conceive subsequent children. (Clayton 1992a; CDC 2004; Hoff and Hoyt 2005; Nelson et al. 2001; Therrell 2001; Paul 1999; Downie and Wildeman 2001; Annas 1982; Mehlman and Botkin 1998; Warren et al. 1982).

The vast majority of research in the field of newborn screening is designed to determine whether testing produces tangible benefits for children in terms of clinical outcomes, and for families by reducing the stress caused by undiagnosed symptoms. However, some researchers have also begun to examine the "unintended consequences" (Merton 1936) for families of this broad-reaching policy whose *intended* consequence is the prevention and avoidance of disease.

The high rate of false-positive screening results is far and away the "ancillary effect" of greatest concern, and the impact of this unintended consequence of screening is the

one that has been most thoroughly researched – although as Wilcken points out, even this "most obvious" potentially harmful by-product of screening has been investigated "only a few times" (2003, S64). False positives are defined as "initial out-of-range screening results that do not signify a… disorder on further evaluation of the child. Generally, these are not laboratory errors, but rather transient findings"(Waisbren et al. 2003, 2565). The rate of false positives is astoundingly high. The overall rate of false positives is difficult to calculate precisely because of differences in technology and laboratory practices among programs (Paul 1999), but the rate is estimated in one recent report to be *more than fifty false positives for every true positive* (CDC 2004). For some specific conditions, such as congenital adrenal hyperplasia and galactosemia, the rate is *more than two hundred false positives for each confirmed case* (Goodwin et al. 2002, 165). It continues to be the case, despite improved screening technology, that for every infant actually confirmed to have a genetic disorder, many, many parents will be notified of an abnormal screen, and will undergo the worry and suffering of subsequent testing only to find out that their child is not affected. Some of these parents will learn that their child has no mutated gene, and that the false positive was entirely unrelated to any genotypic reality. Others will learn that their child – and thus one of them as well -- is a "carrier" of a mutated gene. This means that the child herself will remain without symptoms, but that she might pass on the gene -- and thus, if her partner is also a carrier, the disorder -- to her own offspring.

In discussions, debates, commentaries, and research studies, the stress and expense caused by false-positive screening results are consistently cited as the most serious unsolved problem associated with screening programs. Goodwin et al., for example, call

the high rate of false positives "[p]erhaps the single major drawback to newborn screening" (2002, 165), and Waisbren et al. describe it as one of only two "major concerns" inherent in screening programs (2003, 2565). Wilfond is summarized in CDC workgroup proceedings as saying ""the greatest harms [associated with screening] may be transient distress or long-term confusion among families who have infants with false-positive results on screening tests" (CDC 1997, 10).

Indeed, the research that *has* been conducted on this most publicized of newborn screening's "unintended consequences" demonstrates that, even after follow-up tests indicate to the satisfaction of *clinicians* that the infant's health is not imperiled, *parents* receiving false-positive results experience increased anxiety levels; remain more vigilant overall; and take their children to the emergency department for a disproportionate number of visits (Tluczek et al. 1992; Clayton 1992a; Waisbren et al. 2003; Paul 1999). A Swedish study found that three of four families who got false-positive newborn screen results had strong reactions at the time they were given the news, and that nearly twenty percent of them continued to be concerned about their child's health six to twelve months later (Bodegard et al. as cited in Clayton 1992a, 119). Other research, which focused on children who screened positive for CF specifically, has shown that "at least 5 percent of parents whose infants had false positive results… believed their infants might have [the disease] one year later, despite consultation" (Farrell as quoted in Holtzman 1991, 803).

Another set of unintended consequences occurs when newborn screening identified a child as ("merely") a carrier of a mutated gene. This result has complex implications that are perhaps even more difficult for parents to fully comprehend -- particularly if, as is so often the case, they don't have adequate access to a genetic counselor or other

knowledgeable health professional (Clayton 1992a; Mischler et al. 1998). One study found that 11% of parents whose children were identified via newborn screening as carriers for CF believed that the disease would still develop later in their child, or were unsure whether this could happen (Dudding et al. 2000, as summarized in CDC 2004, 28). Another study concluded that 43% of parents whose children were found to be carriers of sickle-cell disease believed their child to have the disease, and 66% of this subgroup thought their children required dietary supplements in order to remain healthy (Hampton et al. as cited in Marteau et al. 1992, 24).

The unintended consequences for families of *true* positive results have received considerably less attention than have those of *false* positives. Parents whose children's lives were dramatically saved by timely diagnosis of a treatable early-onset disorder are highly vocal and visible, but what is the experience of parents who receive an unsought diagnosis that changes their lives in other ways? What are the forms and degrees of impact the diagnosis has -- regardless of whether or not it improves the child's physical health?

Some commentators, ethicists, and policy analysts do mention concern about the impact of early diagnosis on parent/child bonding, on parental perceptions of disease, and on the child's own emerging identity. Penticuff, for example, writes:

> There is concern that early diagnosis of untreatable diseases may disrupt parental bonding to their infant if the disease presence or carrier status is determined. How will the knowledge that the child has a gene-linked disorder or is a carrier affect these children's socialization into the customary roles that anticipate future childbearing? (1996, 787).

Mischler et al. also speculate that early diagnosis can have an impact on "the family's ability to bond with a labeled child and on the child's own developing self image" (1998,

2 of 11). In a report on the implications of all kinds of genetic testing of children, jointly authored by the American Society of Human Genetics and the American College of Medical Genetics, the authors note that

> [p]resymptomatic diagnosis in children … has the potential to alter the relationships that exist between parents and their offspring and among siblings (Fanos and Johnson, 1993). A child known to have a deleterious gene may be overindulged, rejected, or treated as a scapegoat (Gardiner, 1969) (ASHG/ACMG Report 1995,1236).

Another concern is that what Green and Slonit identified as the "vulnerable child" syndrome will occur, causing parents to become overprotective in response to their perception of serious illness (Ibid.).

A second set of questions about the unintended consequences of true-positive newborn-screen results focuses on the issue of diagnosing healthy children with genotypic disease. Nobody knows how many such children will be identified over time, since the epidemiological data now being collected via newborn-screening programs has never before been available (Newborn Screening Program staff 2005). Many of the conditions screened for have complex and varied forms of expression, and it is not at all clear how many unaffected people have been living with disease genotypes all their lives without being diagnosed. The only data that exist so far are anecdotal. Waisbren et al. (2003), for example, found eight healthy older siblings with genotypes for metabolic disease in the course of identifying thirty-three newborns with the same disorders. They also note that in their sample, only four of eighteen children (22%) who tested positive for the disorder MCADD in their newborn screen group had two copies of the most common and severe mutation associated with the disorder, while all four of the *clinically-identified* children (100%) had two copies of that mutation. Wilcken et al. (2003)

reported finding fifty-five infants with metabolic disorders in three years of newborn screening, but only thirty-three cases of the same disorders had been identified clinically in the same population in the three years prior to the expanded screening. They suggest that the 60% difference may be attributed to the fact that the other affected children in the first three-year period were either healthy, or died without being identified. Other data suggest that substantial numbers of individuals with two copies of a mutated CFTR gene are living well into adulthood without symptoms. For example, ACMG's 2005 Technical Standards and Guideline notes that while carriers of CF are asymptomatic, carrier screening programs are identifying an unspecified number of adults who "carry two mutations in the CFTR gene but are asymptomatic or present with nonclassical, mild symptoms or have a late onset presentation."

The phenomenon of diagnosing healthy children by newborn-screening programs is referred to by researchers mostly as a potentially confounding factor in research studies designed to assess the impact of screening on children's health outcomes. If some of the children would have been healthy without any sort of intervention, researchers note, it is more difficult to tell how effective early intervention was in improving children's health. Reference to the actual consequences *for families* of identifying healthy children with genotypic disease is less common, and is almost always made as an afterthought. Scotet et al., for example, who found at least five asymptomatic children with CF mutations out of one hundred and eighteen in their study, note that "[t]he uncertainty of the evolution of the disease can be a source of anxiety for some couples, so genetic counseling remains of the utmost importance in the management of these families" (2000, 793). And Bonham et al. write that one of the costs of newborn screening for CF is that it is "a difficult

condition to define and early asymptomatic detection may exacerbate these difficulties" (2003, S44). But the experience of parents getting a genetic diagnosis for a healthy child has been considered, when at all, as a theoretical "side effect" of current testing policy rather than as an empirical reality worthy of research in its own right.

Actual research designed to address speculations about any of these unintended consequences of true-positive results is even more scant than the speculations themselves. One recent large-scale study funded with federal dollars through the Human Genome Project's "Ethical, Legal and Social Issues" (ELSI) program did compare "impact on the family" of newborn-screening versus clinical diagnoses for metabolic disorders -- but only as one minor component of a much larger project whose primary objective was to "assess the impact on families of a false-positive screening result compared with a normal result" (Waisbren et al. 2003, 2564). Based on analysis of parents' scores on the standardized "Parenting Stress Index," these study authors found that "[p]arents of children who were newborn screened compared with parents of children who were clinically identified expressed lower levels of stress and greater satisfaction with their support network" (Ibid., 2570). They attribute this difference partly to the fact that children of parents in the former group had markedly fewer developmental and health problems. They also note that – as suggested above -- results may reflect the fact that newborn-screening programs appear to be identifying children with mild or benign forms of genetic disorders compared to the phenotypic norm in the clinically-identified group (Ibid., 2571).

A few smaller quantitative studies have also examined the impact of a cystic fibrosis diagnosis via newborn screening on parenting practices, parental stress levels, and

attitudes towards diagnostic processes (Thompson and Thompson 1986, as cited in Clayton 1992a, 112; Al-Jader et al. 1990; Boland and Thompson 1990). As I document in the chapters that follow, these studies generated some intriguing preliminary data. Not unsurprisingly, they found that receiving a newborn-screen diagnosis is less stressful for parents than a prolonged "diagnostic odyssey" (CDC 2004) during which their child is symptomatic and physicians fail to identify the problem correctly. They also discovered that "…neonatal screening for CF can have a psychological impact on the parent-child bonding" (Al-Jader et al. 1990, 460),[7] and that "… the experiences [with diagnosis] of mothers of apparently healthy children are qualitatively different" from the experience of those whose children are symptomatic (Boland and Thompson 1990, 1244). However, because they used standardized testing instruments and a survey research design, even these few published studies do not adequately address important questions about the qualitative aspects of parents' experience with diagnostic processes. My dissertation research was designed to begin filling a critical gap in the existing literature by addressing under-examined aspects of the unintended as well as the intended consequences of these processes on the lives of affected parents and their children.

My Approach to the Issues

Research Questions and Methodology

The central questions I set out to address in my research were:

[7] Wilcken seems to dismiss these published data when she asserts that "… studies of patients correctly diagnosed with a disorder have not shown early diagnosis to affect adversely the parent-child interaction" (2003, S64). Since she offers no citations to back up this statement, it is unclear what data she relied on in reaching this conclusion.

1. What are the consequences for parents of foreknowledge of genetic disease in their newborn as identified by newborn screening tests? How are these consequences different from those experienced by parents who get a prenatal diagnosis? How does foreknowledge of disease shape parenting in ways that are distinct from how the disease itself shapes it (to be assessed by differences in experience between those who learned about the diagnosis before symptoms and those who encountered symptoms before the diagnosis)?

2. What are parents' experiences with the various processes of genetic diagnosis for children that are currently the norm, and what are their perceptions about how well these processes function?

3. How do power dynamics unfold in relationships between parents of genetically-diagnosed children and the professionals who provide services to them and their children? Do different diagnostic pathways have noticeably different effects on these relationships? More specifically, do different diagnostic pathways have an impact on the deployment and assertion of lay knowledge in everyday caregiving practiced by parents?

"Testing Baby" is a qualitative research study. Within Robert Alford's typology of sociological "arguments," it would fit most comfortably within the category "foreground interpretive" – i.e., in his words, a study focused

> …on the theoretical importance of the symbolic construction of meanings in social interaction. Such meanings are inferred from observations of behavior, from interpretations of texts, and from depth interviews that interrogate individuals about the way in which they interpret their experiences and social relations. The observer is seen as a participant in the co-construction of meanings, not as a separate, isolated, neutral, and 'objective' scientific analyst (Alford 1998, 85).

I relied heavily on the methodologies of grounded theory in designing and implementing my study, since I share this tradition's commitment to inductive theory-building and its belief that much useful social science research emerges through the iterative process of study design, data collection, interpretation, and re-design. Grounded theory is also well suited to an exploration of complex social phenomena such as those I set out to explore: I did not anticipate, nor did I discover, linear relationships between diagnostic systems for genetic disorders and human interactions among affected children, parents, and health care professionals. While I did hypothesize, as noted above, that diagnostic systems *matter* and are likely to have complicated effects on the social phenomena of interest to me, I tried to approach the inquiry itself with as few assumptions about the nature of these effects as I could manage. I set out to "discover… develop… and provisionally verify [theory] through systematic data collection and analysis…" (Strauss and Corbin 1990, 23), rather than to posit a specific theory and proceed to prove or disprove it.

I investigated the questions central to my study through research of two kinds: "interpretation of texts," and "depth interviews that interrogate individuals about the way in which they interpret their experiences and social relations." As noted above and documented in the attached bibliography, I consulted a broad range of secondary sources related to my topic. These included government reports; public-health information sheets and pamphlets; web sites; institutional policies and procedures; personal stories and narratives written for publication; and both theoretical and applied work in sociology and other fields of social science. I also spoke informally with parents of children with genetic diagnoses whom I know through personal and/or professional connections; and

with genetic counselors, physicians, public health workers, and other professionals involved in the formulation and delivery of newborn-screening services. These early conversations helped me enormously to test out my ideas, shape my study, and formulate germane interview questions for my primary research (see below). Throughout the year that I spent actually preparing for and conducting interviews, I was also fortunate to have regular contact with a number of researchers, scholars, and practitioners whose work is related to my study, and to visit a state newborn-screening program and tour its laboratory (including the new MS/MS equipment it had recently acquired). These opportunities to discuss ideas, double-check data, talk through early interview findings, and continually challenge my own thinking were invaluable.

The primary research I undertook was a series of twenty-five interviews with parents of children who were diagnosed with cystic fibrosis either via newborn screen; prenatally; or after the development of symptoms. Because I was interested in the qualitative aspects of parents' experience, I used a semi-structured design. I developed at the outset an Interview Guide made up of questions organized to elicit parents' stories about the diagnostic experience and its impact on their parenting; their perceptions of the child; and their relationships with professionals (see Appendix B). As I proceeded with the interviews and began to see themes emerge from the data, I revised the Interview Guide and added several new questions. However, with the exception of the first few interviews and one or two later ones with notably reticent parents, I found little need to rely on the guide. The interviews flowed very naturally, and it was easy to find seamless ways of moving from one topic to the next in an order that felt respectful and right within the context of the particular interview. Often, little or no prompting was necessary on my

part: responses to the questions I had written in my Interview Guide were given spontaneously by parents as they recounted their experiences in their own ways.

The parents I interviewed were generally extremely eager to talk about their experiences with diagnostic processes, and to tell stories about their lives as parents of children diagnosed with CF. Parents' generosity in sharing both their time and their very personal, often very emotional narratives was a great gift. My gratitude for it is not in the least lessened by my clear sense that the interviews were also meaningful to the parents who volunteered for the study. The fact that my research questions coincided so well with what parents wanted to talk about encouraged me throughout the research process, and I consider the very richness of the interviews a sort of "finding" in itself – a crucial piece of evidence that it matters to parents to have their illness narratives recognized, and that they want to claim their own voices with respect to their diagnostic experiences rather than allowing them to be "reduced to 'clinical material'" (Frank 1995, 12).

I began recruiting parents for "Testing Baby" by sharing my IRB-approved flier (see Appendix A) with genetic counselors and other professionals who are associated with CF specialty centers, and asking them to help me identify parents appropriate for the study. Although most of the professionals I contacted in connection with recruitment at the beginning of my study were not able to officially recruit "patients" without IRB approval from their own institutions, one of these professionals was herself the parent of a child with CF, and thus able and willing to volunteer for the study on her own. From that first interview, and from one other contact with a parent-who-is-also-a-professional, I proceeded with "snowball sampling," i.e., asking each participant to assist in identifying and recruiting other eligible families. Several parents soon posted copies of my flier on

their CF list serves, and a steady stream of people who read about the study on line proceeded to contact me, volunteering to participate. Towards the middle of my research -- when it became evident that I was finding a disproportionate number of parents with later diagnoses and an insufficient number with newborn-screen or prenatal diagnoses -- I supplemented my snowball sample strategy by reaching out again to genetic counselors at CF specialty centers in states that have been conducting newborn screening for CF. These professionals generously took time to post or hand out copies of my flier, and my study sample was successfully brought into better balance (see below).

All but one of my twenty-five interviews were conducted by telephone. In many cases, this was a necessity because of the physical distance between my home in New York and where the particular parent lived. However, even for those parents who lived close enough for an in-person interview to have been possible, most preferred the telephone for both for their own convenience and – I gathered – because of the freedom they felt to speak plainly without having to be face-to-face. I mailed IRB-approved informed-consent forms to each parent prior to our interview, and always began the conversation by asking if they had questions or concerns about the study and their participation in it. I also verified their consent to have the interview tape-recorded. Parents mailed one copy of the signed consent form back to me, and kept another for their own records.

I drew heavily on the methodologies of grounded theory in my analysis, just as I did in my study design. Each interview was transcribed word for word from the tape, usually very soon after it was conducted. [8] This enabled me to read the transcripts from

[8] As noted in my acknowledgments, I am very grateful to several Sarah Lawrence College graduate students for assistance with this process.

interviews that had just been completed even as I was scheduling and holding subsequent ones. I continually derived concepts from my memories and interview notes, which I then verified through review, re-review and comparison of the written transcripts. As I proceeded to work with concepts and gain confidence in their "theory-observation" congruence (Corbin and Strauss 1990, 7), I began grouping these together into the more complex categories that I subsequently used as sub-headings in the chapters that follow. For example, I initially identified from the interview data a variety of concepts related to how control over children's treatment is negotiated between parents and providers. These included "losing control to the doctors once the diagnosis is made"; "being trusted by the doctors"; "trusting the doctors"; "not trusting the doctors"; "reliance on the doctors and lack of parental confidence"; "power and information shifts between parents and providers over time"; and "exerting parental expertise." In order to verify the validity of these conceptual categories once they began to emerge from the data, I reviewed the interview transcripts for specific quotes and anecdotes that fit each conceptual heading. I then reviewed and re-reviewed both the original transcripts and the data, which had been re-grouped according to these conceptual categories. This analysis revealed patterns in the data connecting parents who had had specific kinds of diagnostic experiences with specific kinds of parent/professional arrangements regarding control over treatment. It also revealed changes in parent/professional relationships over time. I thus generated the categories "power of professionals after an early diagnosis" and "diverse pathways to parental empowerment," and subsequently presented them as discrete sections within the larger topic of "parents and professionals" (see Chapter Five). In extrapolating more theoretical implications from these data – in this case, for example, that early diagnosis of

a genetic disorder constitutes a specific form of medicalization that increases parents' reliance on health care professionals and decreases their confidence in their own parental knowledge – I drew also on my knowledge (gained primarily from secondary sources) of what Corbin and Strauss call "[b]roader conditions affecting the phenomenon," such as "economic conditions, cultural values, political trends, social movements, and so on" (Ibid., 11). Throughout the dissertation I have used extended quotes from the transcripts – edited slightly to make them more readable by removing some of the "ums" and "you knows" -- to illustrate thematic findings and give texture to my work.

My data analysis generated many more intriguing concepts and categories than I was able to use in one dissertation. I have listed in Appendix C those I did not use extensively, in order to give the reader a more complete feel for the range of issues that arose in the interviews than would otherwise emerge from what I have written. A few of these are also referred to in footnotes within the text. I hope to continue with analysis of some or all of these to-date-under-used data in connection with future projects and publications.

The Choice to Focus on Cystic Fibrosis

Cystic fibrosis (CF) is an "autosomal recessive" genetic disease. This means that an individual must have two defective copies of the gene – in this case, the cystic fibrosis transmembrane conductance regulator, or CFTR, gene -- in order to express the disorder. Mutated CFTR genes can cause problems with a specific protein that is critical for the healthy functioning of many organs, including the gastrointestinal tract, pancreas, liver, lungs, sweat glands, and genitourinary tract. As a result, people with CF often have problems digesting food, experience recurrent lung infections, and are infertile. Average

life span for people with CF in the United States has increased from fourteen years in 1969, to thirty-three years as of 2001 (CDC 2004, 4). Death is usually caused by respiratory failure as a result of chronic obstructive pulmonary disease.[9] Clinical diagnosis in children can be delayed since many of the initial symptoms of CF, such as wheezing, coughing, and diarrhea, are also signs of various other childhood illnesses (CDC 2004; Wilfond 1995; ACMG 2005; Moskowitz et al. 2005; Wilfond 1995).

I decided to organize my study around parents of children with CF for several reasons. First, I assumed that my data would be more internally consistent, and that identifiable themes would more likely emerge, if I interviewed a group of parents who share a diagnosis rather than a group whose children have an array of different disorders.

Second, I wanted my cohort of parents to include some whose children were not symptomatic at birth. As I noted above, CF is a complex genetic disorder that manifests in many different ways ranging from severe symptoms beginning in infancy, to no symptoms until adulthood, or indeed, ever. This is because there are so many different variations the gene can have: more than one thousand distinct mutations have been identified, and along with them many different expressions of the disorder.[10] CF's broad spectrum of disease expression is also explained by the fact that even the most common mutations (e.g., DeltaF508, which is thought to account for up to two thirds of all CF mutations world wide (CDC 2004, 2)) have what is known as "poor genotype/phenotype correlation" – i.e., individuals with that same mutation do not all have the same traits and symptoms (Tsui and Durie 2001). I therefore hypothesized – correctly, as it turned out –

[9] People with cystic fibrosis who live into adulthood are beginning to tell their own stories about their experience of CF, either by autobiographical work or by participating in qualitative research studies. See for example Rothenberg 2003; Schubert and Murphy 2005.

[10] In the United States, there are twenty-four mutations that account for 80% of all identified CF cases, while the remaining 20% of affected persons have much less common alleles.

that if I chose as my group of interviewees parents of children with a CF diagnosis, I would be able to find some parents who had received a diagnosis before symptoms emerged.

Third, I needed to select a condition that at least some states include in their newborn screening panels, and one that is sufficiently prevalent to allow me to recruit an adequate number of parents into my study. Here again CF fit my requirements very well. With the exception of sickle-cell disease, CF is the most common "life-shortening… inherited disorder in the United States" (CDC 2004, 1), with an overall birth prevalence of one out of every three thousand seven hundred births. The disorder is most common among "non-Hispanic whites," where it occurs in one of every twenty-five to thirty-five hundred live births (Ibid.; ACMG 2005). Prevalence among "Hispanics" and "non-Hispanic Blacks" are one in four to ten thousand, and one in fifteen to twenty thousand, respectively (CDC 2004, 2). One of every twenty-five persons of northern European descent is a carrier, as is one of every twenty-six Ashkenazi Jews, one of every forty-six "non-white Hispanics," one of every sixty-five African Americans, and one of every ninety Asians (Genzyme, 2005). Colorado began screening newborns for CF in 1982, and Wisconsin and Wyoming followed suit in 1985 and 1988 respectively. Between 1998 and the middle of 2004, six other states added state-wide screening programs, and a number of others opted to make state-wide their regional or hospital-based pilot screening programs, bringing the total number of states screening for CF to thirteen. According to a CDC workgroup convened in 2003 to make recommendations about CF and newborn screening, the number of children screened in the U.S. doubled between the year 2000

and the year 2004, from four hundred thousand (or 10% of U.S. births) to eight hundred thousand (20% of U.S. births) (CDC 2004, 5).

CF was also an apt choice because it is among the conditions for which the direct health benefits to the child of early identification are much less clear than they are for certain "poster child" conditions on newborn screening panels, such as PKU.[11] In fact, my reading of the literature suggests that CF has been the focus of more controversy with respect to newborn screening than has any other single genetic condition. The debate about whether CF is a disorder appropriate for newborn screening has centered mostly on treatment efficacy, although other factors -- such as the (assumed) benefit to families of learning about a CF diagnosis before conceiving subsequent children; the efficacy of screening technology; and the adverse impact of false-positive results -- have also been considered. There is no cure for CF, and no clear way to prevent symptoms from developing altogether. However, a number of "palliative" treatments are available to address the complicated array of digestive and pulmonary symptoms CF can cause. These include medications to control infections; manual "chest percussions" or

[11] PKU is cited repeatedly as "'the paradigm therapeutic case' of postnatal diagnosis and proof of the value of genetic medicine" (Paul 1998, 176, including quote from Kelves 1985, 254). This is because, as noted above, it is a serious disorder that manifests within days or weeks of birth, and because it can be successfully treated with dietary supplements. However, Paul's research on this most widely-celebrated example of newborn screening's success reveals that even here the story is more complex than it is typically represented to be, and that the "PKU story is infinitely plastic, employed by both celebrants and skeptics of genetic medicine" (Ibid., 182). "Mass screening has indeed prevented retardation in tens of thousands of individuals," she writes. "There is no doubt that the vast majority of affected individuals are better off with treatment than they would have been in its absence. But PKU screening is not an unqualified success" (Ibid.). In addition to high rates of false-negative and false-positive test results, early PKU screening resulted in inappropriate diagnosis of babies with a relatively benign condition called hyperphenylalaninemia and subsequent dietary treatment of these individuals that was damaging. In addition, "initial assumptions about the required length, effectiveness, ease of management, and psychological effects of therapy were much too sanguine" (Ibid.,178). Unanticipated problems include the difficulties of inducing diagnosed individuals to remain on the diet once they enter adolescence and beyond; the expense of treatments in a society that does not offer universal coverage for medical care; and the extreme risk of severe mental retardation in the next generation of babies born if affected women do not resume their diet prior to conception and maintain it during pregnancy (Ibid., 177 – 182).

mechanical vests used to clear mucus out of the lungs; and replacement enzymes to improve digestion and nutritional status (ACMG 2005). The central question in the CF newborn-screening debate has been this: Do the available medical interventions make a notable difference in children's health if started at birth?

Two large-scale, longitudinal, randomized clinical trials (RCTs) have been designed and implemented in an attempt to answer this question – one in Wisconsin that ran from 1985 to 1994, and one in the United Kingdom that ran from 1985 to 1989. These studies randomly assigned neonates to either a screened or a control group, and then followed all infants with CF (diagnosed either by newborn screen, or – in the control group – later, with the onset of symptoms) over time to assess health outcomes. A number of cohort and registry studies have also been published. These use retrospective data to see if there were significant differences in childhood health between newborn-screened and clinically-identified children. Data from these studies have been interpreted in two diametrically-opposed ways: as clearly demonstrating the health benefits of newborn screening for CF (particularly in terms of improved nutritional status and improved growth for children identified at birth); and as clearly failing to demonstrate significant benefit. In the United States, investigators in the Wisconsin study have published a number of articles arguing that the trial clearly demonstrated nutritional benefits of early diagnosis, as well as improved long-term growth for screened infants (see for example Farrell et al. 1997, 2001). But these results have been challenged on methodological grounds, as well as on the grounds that policy recommendations regarding newborn screening for CF should not be made on the basis of improved nutritional status alone (see for example Grosse et al., 2001; Farrell et al. 2001). The Wisconsin study also

discovered a number of cases of early infection of diagnosed infants (many otherwise asymptomatic) with pseudomonas and other bacteria because they come into contact with older, symptomatic children in CF specialty centers. This issue was downplayed by promoters of newborn screening and emphasized by its critics. Researchers in other countries have concluded, on the one hand, that newborn screening results in clear "beneficial effects" for children, and on the other, that while there is "preliminary evidence for improved growth in the screened population… the main benefits of screening result from the improved efficiency of the health care system and reduction in the length of time required to make a diagnosis" (see Dankert-Roelse and Meerman 1995 and Bonham et al. 2003, respectively).

Although debate about CF continues to rage, even in the single year that has passed since I designed my study in the summer of 2004 the tide of professional opinion appears to have turned in favor of screening for it. In late 2004, the CDC released the findings of its "working group on CF" in the Morbidity and Mortality Weekly Report. In this report (CDC 2004), they reversed their earlier finding that there was insufficient evidence of benefit from early diagnosis to support CF screening (CDC 1997) and concluded that "the magnitude of the health benefits from screening for CF is sufficient that states should consider including routine newborn screening for CF in conjunction with systems to ensure access to high-quality care" (CDC 2004, RR-13). The 2005 ACMG report referred to above also recommends that CF be one of the fifty-four conditions included on a uniform national screening panel. Notably, however, CF's rating within the methodology of that study placed it precisely at the cut-off mark between conditions that warrant universal screening and those that do not. With a score of exactly 1,200 in a

system that made 1,200 the "logical separation point between high scoring conditions… and low scoring [ones]" (ACMG 2005, 9), CF continues to be a test case for newborn screening even from within the screening paradigm itself.[12]

The Use of Comparative Data in the Study

As I have noted above, it was the rapid expansion of newborn-screening programs, and the concomitant changes I hypothesized must be occurring in parents' experiences with genetic diagnosis, that originally excited my interest in "Testing Baby" as a dissertation topic. In order to understand how newborn screening may be changing the way parents perceive, experience, and respond to their children's genetic disorders, however, it was important to interview not only parents who received a diagnosis for their child through newborn screening, but also their counterparts who learned that their child had CF either via prenatal testing[13], or after the emergence of clinical symptoms. As I had hoped, this comparative design – i.e., conducting interviews with parents of children with the same condition who arrived at genetic diagnosis through various routes – was very helpful in illuminating the impact of newborn screening, and in my attempt to isolate its effects. All of the interviews were incredibly rich, but the differences in experience were often striking. I was also aware throughout the interviewing process that the accounts of parents who received a later diagnosis may prove very valuable in future, since it may well be that clinical identification of CF will become a thing of the past before long.

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[12] It should also be noted that ASHG's methodology is currently being criticized on a number of grounds. Doubts are being raised, for example, about who participated in the process, since the group that was assembled was made up entirely of newborn screening "insiders" and did not include anyone with an "outsider" perspective. ASHG is also coming under criticism for releasing its recommendations to the public *before* publishing its report, and for *still* failing to make public some aspects of its methodology.

[13] Because a central focus of my research is the relationship between diagnostic processes and subsequent parenting of the diagnosed child, I did not interview women who chose to terminate their pregnancies after receiving a prenatal diagnosis of CF.

The fact that I interviewed parents with different diagnostic experiences should not be taken to imply, however, that I employed a strict comparative design. For one thing, I did not set out to interview equal numbers of parents in each of the three groups, and in the end my sample was uneven (see below). For another, I found during the research process that there were actually many more sub-groups and defining categories than I had initially anticipated. For example, some experiences -- though by no means all of them! -- were shared among all parents who received what I call in my text an "early diagnosis," regardless of whether that diagnosis was by prenatal testing; newborn screening; or diagnosis immediately after birth because of tell-tale symptoms. Other experiences -- though again only a subset -- were common to those with asymptomatic children, or, again, to those with actively ill children, regardless of the age of the affected child or of how the diagnosis occurred. Even in these cases, however, the elements of comparative design as I initially conceived them continued to be useful with respect to extrapolating the potential impact of newborn-screening policy on social phenomena. For example, all parents with an early diagnosis might struggle with what it means to know that their children have CF before they have gotten to know those children in other ways. However, it can be reasonably expected that as more and more children are screened for CF at birth, the number of parents with an early diagnosis will grow relative to the number of those with a later diagnosis. Similarly, the number of asymptomatic children with genetic diagnoses will likely increase as newborn screening for CF becomes more commonplace.

Elements of comparative design were also helpful because they kept before me throughout my research what remains constant for all parents regardless of diagnostic

routes – i.e., the pain of learning that something is or could be devastatingly wrong with their child. I discovered through these interviews many ways in which parents' experience of CF is in fact socially constructed by diagnostic processes, and I believe that how we as a society organize these experiences makes a profound difference. At the same time, I was humbled by constant reminders that there is no good way to get hard news, and that the terrible fact of a child's impaired health is the dominant reality for many parents of children with CF. In the day-to-day life of parents whose children are symptomatic, being forced to bear witness as CF "wreaks havoc" on their children's bodies (to quote one mother) is the most profoundly "structuring" experience they confront. It is this that keeps them up at night, and that makes them weep as they tell me their stories. They would give their own lives to free their child from CF if they could, but the reality is that nothing – not the complex medical interventions available today, and not the most perfect of diagnostic processes we could conceivably design – can answer this fervent wish for their child's good health and long life.

Study Sample

Of the twenty-five parents I interviewed for "Testing Baby," eight parents received a newborn-screening diagnosis; two parents received a prenatal diagnosis for a first affected child, and one for a subsequent affected child; four parents received a diagnosis within a few days or weeks of birth for a symptomatic child; and eleven received a later diagnosis (between ages two months and seven years) for a symptomatic child or children.[14] Most children were between the ages of one and seven at the time of my

[14] One of these eleven parents had still not received a conclusive diagnosis for her child, age one, at the time of my interview, and it appears unlikely that CF is the cause of the child's symptoms. However, since the interview was already underway by the time this situation became clear to me, and since many of this

interview with their parent, but there were two extended outliers on the upper end of the spectrum, and two infants. The total range of the children's ages at the time of the interview was thus three-and-a-half months to twenty-two years, with an average age of five. There was a healthy diversity of ages at the time of interview within both the group who had had a newborn-screen diagnosis and the group who had had a diagnosis after the emergence of symptoms.

Eleven of the thirty children with CF in my study were diagnosed before symptoms, and their ages at the time of diagnosis ranged from before birth (via prenatal testing) to thirty months old. As I discuss in Chapter Three, six parents in my study proceeded to have another child after they already knew about the CF diagnosis of an older child. Two had already had one or more additional children with CF before finding out about CF in the older child. At least one child in my study is considered by her doctors to have a form of CF that will not result in symptoms at any time, and at least two others have also been identified with "mild" forms of the disease which may or may not cause health problems in later life.

I interviewed a total of twenty-three mothers and two fathers. One of the two fathers is married to a mother who also participated in the study. Three of the mothers in my study are not living in the same house with the affected child's father.

Twenty-four of the parents I interviewed identified themselves as "white" or "Caucasian" when I asked them to describe their ethnic identity, and one identified her/himself as Hispanic. One other parent noted that her husband is of Indian descent.

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mother's experiences with the diagnostic process are so similar to the experiences of parents whose child did get an eventual CF diagnosis, I have not excluded her from my sample.

Given that CF is a disease most common among non-Hispanic white people, these demographics are not surprising.

I asked parents to tell me their occupation, and that of their spouse if they had one, as a proxy for class. Because the interviews were conducted by telephone, and because the subject matter was so personal and emotional, I did not feel my research would be well served by asking more explicit questions about socio-economic status. However, it was very difficult to tell much about class issues from occupational data alone. I deduced from the data on their occupations, and on the basis of other points and perspectives shared during the interviews, that my sample included a more or less equal mix of working- and middle-class families. Any breakdown more explicit than this approximation would risk significant inaccuracy.

Mothers and Fathers

I intentionally designed "Testing Baby" as a study of *parents'* experience rather than of *mothers'* experiences. This is largely because my review of the literature led me to agree with Morgan et al. that "[t]he experience of fathers has been generally neglected in parent-child research" (2002, 222). It is also because of ways my own experience has colored and contextualized my research. The father of my two children has been such a completely equal partner in the parenting process, and my own father has played such a central role in raising and nurturing both me and my children, that the idea of excluding fathers from the outset *felt* wrong -- even though I am well aware (as a member of the social world, as a sociologist[15], as a feminist) that mothers do a hugely disproportionate amount of caregiving work of every kind.

[15] See for example, Grob and Katz Rothman 2005.

When I recruited interviewees for my study, I explicitly called for "parents" rather than "mothers" to volunteer. As I proceeded with the interviews and saw how few fathers were responding, I asked each mother -- if she offered to help identify additional parents -- whether she knew any *fathers* who might be willing to talk with me. However, as noted above, in the end I succeeded in recruiting only two men -- and one of these became interested in the research only at his wife's urging.

While I did not manage to recruit a significant number of fathers into my study, I did begin to learn from my interviews with mothers some important things about how differently mothers and fathers respond to the diagnosis – despite the fact that I did not explicitly set out to investigate this relationship. My interview data – preliminary as they are – suggests that the gendered division of labor that leaves mothers in charge of most aspects of child-rearing becomes even more pronounced once a child is identified as sick. Some mothers in my study spoke about how involved the child's father is in parenting, and/or how they work together to care for the affected child. Most mothers who raised gender issues, however, described the father as being "helpful" in what is clearly an ancillary role, and many of these talked about how the different responses they and their partners had to the diagnosis created a distance between them. As one woman put it in a dramatic but not entirely uncharacteristic anecdote:

> When I told my husband that he was the [cystic fibrosis] carrier also, his reaction was, 'Oh, I'm so sorry this is happening to you.' And I was like, 'Happening to me? It's happening to both of us and our whole family!'

In future research about the experience of diagnostic testing, it would be important to resolve study-design issues around gender in a manner more satisfactory than I have managed here. Possibilities might include: to conduct a study interviewing only fathers;

to interview equal numbers of men and women (using a recruitment strategy other than snowball sampling) and ask each to reflect specifically on gender issues; to exclude men from the study altogether and collect more complete and explicit data from women about their perceptions of gender issues. In the meantime, what I have done in this dissertation is to include fully the data from the two men included in my sample, and to retain the term "parenting" as a descriptor for the group I spoke to at large. However, I have reverted to the term "mothers" when discussing phenomena that were notably specific only to women in my sample. I have also drawn attention to gender differences in particular instances where my data were sufficiently consistent to merit comment – though I have refrained from making a comparison of mothering and fathering a major focus of analysis because my overall data in this area are too partial.

Limitations of My Study Design

I have already alluded to the limitations of "Testing Baby" with respect to both gender and class. Another limitation is the small size of my sample. I am comfortable that I reached a legitimate saturation point in my primary research because as I drew near the end of the interview process – at least with parents getting a newborn screen or clinical diagnosis -- unfamiliar themes ceased to emerge. Nonetheless, I am aware that twenty-five interviews constitute only pilot data. My assertions in the chapters that follow, however, are stated without repetition of this caveat.

Because parents who participated in the study volunteered to do so, the sample is also biased in favor of those who were motivated – for whatever reason – to join. This may have resulted in an over-representation of parents who had dramatic experiences of one sort or another – either very satisfactory or very unsatisfactory – and were therefore eager

to tell their story. It may also have resulted in an over-representation of highly conscientious parents as reflected by the fact that they found out about the study through a network of contacts in the CF world they had already built for themselves.

The small number of mothers in my sample who received a prenatal diagnosis is yet another limitation. Despite having talked with only three such mothers, I included the very rich data from these interviews in my analysis at appropriate junctures just as I included the data from my interviews with fathers. I was also able to supplement my primary data about mothers' experience of prenatal diagnosis with excellent data from secondary sources, since (as already noted) this group has already been the focus of considerable sociological inquiry.

What Follows

The remainder of "Testing Baby" consists of four chapters presenting and analyzing the data I gathered during my research, and a final chapter concluding the dissertation. The chapter immediately following this one provides an overview of the variable role CF diagnosis plays in the lives of families depending on when and how the diagnosis is made. More specifically, it examines the *timing* of the diagnosis in terms of its proximity to pregnancy and birth, and how parents feel about and handle the process of "going public" with diagnostic information. In Chapter Three, I describe current screening and testing procedures in prenatal, newborn, and pediatric settings, and then explore aspects of this experience that emerged as central in my interviews: the waiting period; issues of education and consent; and the vexed nature of "choice" about screening and testing. In Chapter Four, I discuss the impact of a CF diagnosis on parenting, and in Chapter Five, I focus on how it affects the relationship between parents and professionals and the

development of care-taking practices at home. Chapter Six summarizes my findings and their sociological significance, and also outlines policy implications, changes in newborn screening practice, and future research that might usefully follow from what I have written here.

Chapter Two

From Sober Confirmation To Devastating Surprise: An Overview Of Parents' Experience With CF Diagnosis

No sociological research is needed to determine whether it is difficult for parents to receive news that their child has a potentially very serious genetic disorder. Regardless of how and when the diagnosis comes, it brings with it a range of painful responses: shock, grief, anger, numbness, fear, and very often the feeling that, as more than one mother put it, "our little world just crashed." Terror at the prospect of outliving the child is a constant for all parents who learn that their child has cystic fibrosis. Paige voices here emotions that nearly all parents in the study expressed in one way or another.

> I had when she was born so many hopes and dreams and aspirations for this child and she is, she is the most beautiful child I have ever laid eyes on and so smart, and I love her so much and I want so many things for her in this life, and yet it scares the shit out of me, excuse my language…to think of, you know, what's it, what's it gonna be like, how sick is she gonna be, how many years does she have and the hardest thing is nobody can answer my questions.

Betty raises another, related theme when she talks about how her daughter's diagnosis a few days after birth signaled the end of normalcy for her family, the end of her heretofore little-examined yet dearly-held assumption that her children would outlive her.

> Part of my brain was just reeling from the fact that there was something that was wrong with this child, that she was chronically ill and that from that day forward my life would be drastically different. That people would perceive us different as a family… that there wasn't going to be any of the months of normalcy from now on, that I had to worry about life expectancy for one of my children….

Many also expressed feelings of panic: "The more I would read on cystic fibrosis the more upset I would get," says Kim, "until I was like blacking out, couldn't breathe, my heart was pounding a mile a minute all the time…. it was terrible of course because you think she is going to die immediately." And from Catherine, "I remember the day before we went for the test…I took Joseph for a walk and I could not breathe…. I was trying to take a breath and I was so nervous that my heart wouldn't unclamp. And I just thought, I was trying to tell myself, 'Your son has cystic fibrosis.'" Those of us who have not experienced the heart-wrenching depth of a mother's grief and terror at this moment can only try to stand back and bear witness.

For all of the parents I spoke to, the diagnosis also brought them into immediate contact with health-care providers, and thus with the realization that cystic fibrosis means not just a threat to health and longevity, but also a permanent regime of daily treatment. "It was just very overwhelming," says Catherine.

> Cause like you get a diagnosis one day and the next you are being pounded with information like your son is going to be taking medication every day for the rest of his life, every time you put the food in his mouth. That's really the most mind-blowing thing of it all. That like he'll always have to have it…. That was a really hard thing to accept….

Also overwhelming is the sudden inclusion of multiple unfamiliar health-care providers within the inner circle of adults responsible for the child's welfare – a whole "army" of professionals, as one mother put it. The complex system of care that parents are connected to immediately after the diagnosis can feel strange and confusing,

signifying as it does such a clear demarcation between before CF and after.[16] Parents often meet, in one day, nutritionists, pulmonologists, respiratory therapists, social workers, dietitians, gastroenterologists, and more. This throng of professionals is very valuable to the family in the long run, but their abrupt introduction as permanent features of the new post-CF landscape is decidedly overpowering at first. "I just couldn't handle all that," remembers Judy. "All these people, that was just so overwhelming."

Anguish, heart-wrenching worry about the future, a sense that life is irretrievably altered – these responses are shared, the somber joint province of all parents receiving their child's cystic fibrosis diagnosis. But other aspects of parents' experience vary significantly, not just because of individual differences in emotional make-up and ways of coping under stress, but also according to when and how and under what circumstances the diagnosis is made. The remainder of this chapter provides an overview of some fundamental ways that diagnostic processes affect parents' encounters with CF, their experiences of early parenting, and their sense of the child's emerging identity. The themes laid out here are ones that parents brought up again and again in the interviews, and I present them in this first chapter almost entirely in narrative voice. The next three chapters then proceed to examine in greater depth, and with reference to other research and sociological literature, the issues introduced here.

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[16] My study did not include any parents who were not directly connected to abundant medical resources. However, one concern of note with respect to newborn screening is that no coherent mechanism exists for assuring that children confirmed positive for a genetic disorder receive long-term services, or that they establish connections with an appropriate specialty center (Clayton, 1992). In New York State, for example, public health officials are not able to say what happens to diagnosed children once confirmatory testing is complete since they have no mechanism for tracking families in the long term (newborn screening program personnel, New York State Department of Health, interview by author, June 21, 2005). In a national survey of newborn screening programs, Hoff and Hoyt found that fully half the states do not conduct long-term follow-up for infants with abnormal screens (2005).

The Variable Role of CF in Constructing Children's Identity

Diagnosis of a genetic disorder can mean the long-sought answer to the question "what is wrong with my child?" It can mean an unsuspected, unsolicited revelation of invisible chromosomal abnormalities within the body of a healthy, or of a seemingly healthy, newborn. It can mean the relief of finally having a label and some appropriate medical treatment; the confirmation of a half-conscious suspicion; the death of blissful ignorance; the advent of needless worry. In each case, what preceded the diagnosis, and what brought parent and child to the point of getting it, profoundly influence its impact.

Despite CF's high prevalence, pediatricians often do not recognize its clinical manifestations in babies and young children. As a result, some families have been coping with a sick child for months or years before a diagnosis is made. Many have felt helpless in the face of their child's repeated illnesses, apparent failure to thrive, digestive problems, or other symptoms. All too often, parents have also had their observations, suggestions, and concerns dismissed by health care professionals (see Chapter Five). Not surprisingly, then, the sadness and fear these parents feel when receiving the CF diagnosis is mixed with tremendous relief. As Annie put it, recalling her son's diagnosis at age seven,

> … when we got it I was totally relieved. Even though he had cystic fibrosis and I knew what it was and I knew the outcome of it, it was a relief, because I knew he was gonna be treated correctly. I knew… that I wasn't crazy, that I wasn't looking for something to be wrong with him, you know?

For these parents, the reality of their child's suffering has already unavoidably asserted itself, and the idea of a healthy, "perfect" child has already receded – sometimes long since. What matters now is to find out what the problem is and take action to make

things better for the child. In Jody's words, "We knew that there was something wrong, so tell us what it is and we'll go forward. [After the diagnosis] we had a name. At least we had something to deal with – it's better than nothing. We had some understanding of what it was." And, perhaps, some medical treatment able to alleviate suffering and prolong life.

Catherine's baby was only seven months, not seven years, old when the diagnosis came. But her child had been gravely ill, and she had been trying to convince others that there was a serious problem for quite some time. A "mixture of horror and relief" was her response to confirmation that CF was the cause. At least, she goes on, it wasn't some exotic, unknown condition.

> I had envisioned flying around the world looking for specialists trying to figure out what was wrong with this child. But it was a relief to know that there was help you know, at hand and that they knew about the disease and it could be, I don't want to say treated, but you know what I mean.

A positive CF test is confirmation of established suspicions for some parents, but for others, the news arrives unbidden and unexpected, a devastating surprise. In my study, parents who received a positive newborn screen for an infant who had shown no signs of illness were clearly least prepared for the presumptive diagnosis, both emotionally and in terms of their knowledge of the testing process and of the disorder itself. For these parents, it is not a deterioration in their child's health that rocks their world, but rather a phone call from or visit to the doctor's office.

Francesca got the call from her pediatrician's office when Tess was about a month old. She was asked to come in and discuss the baby's "blood work." When the doctor

proceeded to inform her that the baby had screened positive for CF and would need further testing, her surprise was absolute.

> Even when she was born there was nothing… she was born and she was seven pounds eight ounces, just I mean everything seemed perfect… I didn't know…. even now when I think about Tess having CF it's so weird cause we taped her birth and everything and I look at it and I'm like I don't get how this happened because everything was so perfect… everything seemed to be so right so it was a, it was a big, it was a huge shock I guess, after, to find out she has the CF.

Francesca had received little information about the newborn screen in general, and none about CF in particular, at the time the blood was taken from Tess' heel. The pediatrician provided her only "a little bit of explanation" when he gave her the screening results. She describes the next week, before she went to the CF clinic for further testing and education, as the "hardest part," a haze of fear, guilt, sorrow and confusion.

> 'Til my first clinic I really didn't know what was truly going on or what was the prognosis or anything… it took me like two days to quit crying… I cried a lot, I mean that, and I slept with her even for nights before I could let her just go back in her own crib and stuff like that because you just want to hold on so tight because you don't know…. It was like a blaming thing, like what did I do wrong and how is she gonna be with it, you know, [and] is she hurting now? I didn't know anything about it and I got sent home with a little bit of information but it was like a week later before I had my first clinic…where I got you know the most knowledgeable information.… So that was a long week waiting to see kind-of where this was going or what is it. I remember I still remember I went home… and I was just bawling and I was there all by myself and calling like half of [city name] it felt like trying to find somebody that… was home and that could talk you know…. They told me it was genetics but I didn't understand what genetics truly meant either so I was like well what did I do and you know what could I have done different.…

His infant was two months old when Anthony got the unexpected phone call. Baby Denise, a second child, had appeared perfectly healthy. Her body chemistry did seem a little different from her older sister's, in retrospect, but certainly there were no symptoms

or worrisome signs of any kind. The phone call from the pediatrician "came as… very much a surprise to us, just because we didn't even know that the test had been run," he remembers. Like Francesca, Anthony and his wife knew very little about CF, and had to seek out whatever information they could in the interim between the call with the screening results and the confirmatory sweat test.

> And it was actually just horrible timing… 5:15, 5:30 P.M.…two days before the big holiday, before the big Christmas holiday, and nobody was around. I actually called some of my colleagues who I know are very well versed in birth defects and so on and they weren't even available. They had left… so it was just you know bad timing on that. I ended up speaking to a nurse over the telephone and contacting the San Diego Cystic Fibrosis Foundation [which was still open] … I needed a person, I didn't wanna just look online and speak online… I needed to speak to somebody immediately.

Suzanne had even less time than Francesca or Anthony before getting a CF diagnosis for a newborn she thought was just fine. Since she and the baby's father had both tested positive as carriers for CF during the prenatal period, she had consented before Quinn's birth to have his cord blood tested immediately. Nevertheless, it came as a surprise to Suzanne when the test results came back on Quinn's fourth day. Aside from some iatrogenic problems with his umbilical cord, which kept both her and the baby in the hospital beyond the time at which they might otherwise have been discharged, she had found him relatively healthy since birth. Completely focused on getting to know the baby and enjoying him, her firstborn, she hadn't been thinking about the test at all. "It didn't even occur to me [to think about the CF test] even when the doctor came in to tell me. I think I was more wrapped up in the euphoria of having him… I didn't really think about it until he came in and told me." However, once the results were in, Quinn's identity was immediately transformed from that of a healthy child who could be cared for

primarily by his mother to that of a sick child who needed to be treated by the health-care professionals in Suzanne's hospital setting. "I had to learn everything about him," she recalls, "… how to treat him. [I] had to learn the physio and about the enzymes and different medications that he had to take…. It was like starting again really… It was huge, it was absolutely huge." Learning "about him" becomes, with the test result, a process of learning about CF and learning how to begin preventive treatment – a radically transformed context for early parenting. Her son is no longer seen as, constructed as, just "a normal baby who you were allowed to hold and try to nurse." Rather, he is a newborn with a genetic disorder, a baby who requires expert treatment from professionals and whose mother cannot, at first, be entrusted with his care.

For most parents, the newborn-screen result and subsequent testing mark the end of their experience of their child as "normal" and the beginning of their life of caring for a child with special health-care needs. For some, however, the "diagnosis" turns out to be a genetic profile without manifest or foreseeable clinical implications. A CFTR mutation has been identified and the result has been given to the parent, but the child is healthy and will in all likelihood remain so.

Margo, a little girl who was two-and-a-half at the time I interviewed her mother, is a child who falls into the latter category. Shannon, who just happened to be in her pediatrician's office when the newborn-screen result came in, had never heard anything about CF before her doctor informed her of the positive screen. "[Margo was] chunky," she recalls. "She was over nine pounds at birth, so I mean there was no indicators, you know, I mean visually, you know looking at her there wasn't anything to, to um, you know think there was anything wrong with her…." Getting the newborn screen was

"very terrifying you know, and I mean you're sitting there and it's like you're holding what you thought was a really healthy baby…."

Shannon had not explicitly sought out diagnostic testing for her baby, and had no reason to believe anything might be wrong. She was aware that newborn screening is part of routine infant care in the hospital, but did not know what conditions were included on the screening panel in her state. The positive CF screen brought Shannon – as it had Francesca -- abruptly face to face with the contradiction of an abstract test result suggesting that her baby suffers from a serious and potentially fatal genetic disorder, and the very concrete reality of the thriving infant in her arms. Unwittingly, Margo was subject to what Nelkin calls "a form of predictive diagnosis…[that] allows the anticipation of problems or conditions that are not necessarily visibly expressed in overt symptoms." Shannon's assumption that her daughter was fine because she nursed well, slept well, looked well was suddenly called into question, the experiential evidence on which a mother bases her sense of her children trumped by DNA analysis. Margo's identity was suddenly reconstructed by her genetic test, which like all genetic tests "treat[s] molecular signs as more important than behavioral or physical expressions, and allow[s] the classification of people on the basis of biological abstractions." (Nelkin and Tancredi 1994, as cited in Nelkin 1996, 539)

Shannon and her husband left through the back door of the pediatrician's office that they had entered blithely, from the front, an hour before; it was too overwhelming to contemplate being in public as their "world came crashing down." Shannon recalls the sorrow, fear, and contradictions that pervaded the time after the screen.

> The hardest part about that whole time… it's just knowing that this is something that…could potentially take her life. Having been a mom already, the hard part was that at times I felt like I was scared to fall in love with her because I didn't know how long I'd have her and so that was the hardest part, because I mean those, those [weeks and months] that we were waiting for the information back, um, I have to say I did look at her a lot and think about it and, you know, and it's hard to look at her and go well… nothing's wrong with her cause she's beautiful and she's, you know, just the epitome of healthy…. [When she was a newborn I wondered] you know, 'Okay, who are you?' And then to find this out a week and a half after she's born, so then that compounded on top of it… it's kind-of like okay… I'm trying, now's the time when I'm supposed to be falling in love with you, but I'm scared to.

Margo looked the same after the test as before. She acted the same as well, and her health remained unchanged. But, as I discuss in the chapters that follow, the positive screen and subsequent sweat and genetic tests confirming that she carries two copies of a CF mutation changed many things for her and her family.

For a third group of parents, CF diagnosis is neither the end point of a long search for answers nor an unexpected revelation, but something in between. These are the parents of children who get a diagnosis during their child's first weeks or months, but whose children have already begun to show symptoms of one kind or another, causing the parents to suspect there is a problem. In my study, parents who received the diagnosis in this intermediate zone -- recently sensing or perceiving a problem, but not yet acclimated to the idea of a sick child -- fall into two primary subgroups.

The first of these is made up of people whose children experienced significant medical problems at or soon after birth. The most frequent issue, and one that plagues approximately 15% of all newborns with CF, is meconium ileus (MI). This intestinal blockage generally results in a distended abdomen, inability to pass stool, and/or vomiting. It nearly always requires surgical intervention within a few days of birth.

Because it is most likely (though not always) indicative of CF, a diagnosis of MI is regarded by many as a presumptive diagnosis for CF (Moskowitz et al. 2005).

Four mothers in my study had babies with MI, and therefore got the CF diagnosis within the first ten days or so after birth. For these parents, joy over the baby's birth, worry about an emergent health issue, and shock at the CF diagnosis all crowd together in rapid succession. Betty recounts, with emotion, how for her, the newness of her baby daughter, the symptoms of the moment, and the looming possibility of a genetic disorder are densely intertwined.

> Initially she let out a little cry, but she needed to be intubated because there was so much fluid in her tummy that she couldn't expand her lungs to breath. And they took her. I did not see her, just heard her tiny little cry and that was that…. She had meconium ileus and…we were told of the possibility [of CF] then because of the severity of the meconium ileus that she'd had. The surgeon said do you guys have cystic fibrosis in your family and we're like "no." That was like not even a possibility for us, we're just like well that can't be true…. It was about ten days later, we got the results back that it was positive for CF. And we were just in shock…. I was just, it was just shocking um and we just, we were numb, we were stupefied [and] didn't hear much of what the doctor had to say….

Although baby Rose had been ill from the moment she was born, for her mother Betty there was a tremendous gulf between having a sick newborn, and having a child diagnosed with a serious, often fatal, genetic disorder. Finding out from the doctor that Rose had tested positive for CF sent her into freefall across that distance.

> This was my perfect baby that only had this one little issue that we were gonna fix and bring her home and everything was gonna be great. You know that was what the resident said…. I was holding Rose you know, [saying to her] oh you know here she is and… we're gonna fix her, we're going to fix her…. [Then] we go into this meeting and he tells us that there is something you know devastatingly wrong with our child and um we were very distraught, we were very upset, we went into the nursery for just a few minutes and just held and (deep breath) we're just like we just have to go. We have to go absorb this….

For Sheri, too, it felt like the bad news just kept on coming. She had loved being pregnant, and was greatly looking forward to the arrival of her first child. When he was born, however, he was taken straight to the NICU, where they determined he needed to be flown to a larger hospital for surgery. "They gave us 15 minutes," Sheri remembers, to hold him since "we hadn't even held him yet" Close on the heels of the news that baby Jasper needed surgery came word that this was not just a discrete problem, but that the diagnosis would probably be CF. "So I remember when the surgeon said that…. he most likely has CF I you know just sat there for a few minutes and then I just ran from the room saying 'When do we get some good news?' So we were in quite a bit of shock um and I was kind of in denial at first."

Parents in this situation are overwhelmed all at once by the general caregiving responsibilities associated with any new baby, fears for the future, and the technical difficulties of tending for an ailing infant. Parents wonder, does the CF diagnosis mean the child will always be sick like this, or will things get better? Betty describes this uncertainty with eloquence:

> While we were waiting for this next surgery to reconnect [Rose's intestines], she started her first lung exacerbation and she had bronchitis…. [S]he got really mucusy, really coughy, she just really, she was just really bad. And it scared me because I just thought that was the way she was always going to be. I thought that was natural progression and no one took the time to really explain that you know there's going to be certain times in her life where she's going to be so sick that she's going to need IV antibiotics and that kind of thing…. I mean everybody's like, 'do you have any questions?' Well, I didn't know the right questions to ask.

All parents of sick children have to worry about caring for them in the moment. For those whose child is also diagnosed during infancy with a genetic disorder, this concern

is compounded by preoccupation with the future and how the child's needs will be met in the longer run. Parents wonder, how can I possibly care for this child at home after all the medical intervention and equipment that has been required during the first days or weeks? And – especially for first-time parents – how can I master all the technical requirements associated with these medical interventions while at the same time managing the already-complex mechanics of infant care? As Leslie put it,

> I really didn't wanna hold John at first…. I mean I just I had heck of a time with all the tubes. I would change his diaper and I was getting the IV lines in it and everything was getting tangled up and its just like 'ahh….' I'm not all that coordinated and a teeny tiny baby and teeny tiny baby who poops like 40 times a day… [I thought if] I'd stand there maybe the nurse will do this, but no…. [And] I didn't know how to operate his car seat…. [or] how to strap him in, and then he's got all this, all the you know all the baby stuff …. A couple of days before we were supposed to leave then it's like 'Oh, by the way, you're going home with all these medications.' And then I remember just before we were getting ready to leave then the doctor added another one, it's like 'ahhhh….'

Among parents of already-symptomatic children, there is a second subgroup: those who, despite their observation of their child's health problem before it was diagnosed, did not yet have a firm view of their child as significantly ill. This group itself includes both parents who received a newborn-screening diagnosis, and parents who got a later diagnosis for a child who had only recently begun to exhibit symptoms. In my study, there were three parents who had the latter experience, and four parents (out of eight tested at birth) who told me they had begun to wonder if something might be wrong before the positive newborn-screen result came back. This is consistent with what Boland and Thompson (1990) found in their study population of infants screened for CF (thirteen of twenty-nine had begun to show some symptoms), and a lower percentage than found by Al-Jader et al. (1990) (five out of eighteen screened infants in that study

had begun to show symptoms). These parents are suspended, at the time of diagnosis, between their hope and/or knowledge of a healthy baby, and their sense – sometimes conscious, sometimes partly unconscious – that something outside the normal range is at play.[17] The diagnosis confirms a partially-formed fear, abruptly asserting one version of the various futures parents have contemplated for their child, one version of who they are, as primary.

Andrea describes knowing something was "different" about Bobby, her third child, from the moment he was born, and maybe even while she was still pregnant. Then, during the first few days at home, Bobby's appetite seemed ravenous, disproportionate. She was concerned, and on the phone with the nurses seeking advice. Neither her instinctual knowledge nor her empirical observations of worrisome signs prepared her, however, for the call from the doctor's office with the positive newborn-screen result, which she likens to "a kick in the head." It's one thing to have a feeling, to have a concern, and another thing entirely to be confronted with the harsh reality of a genetic screening. The screening numbers above the normal range are irrefutable, undeniable, more real than either instinct or observation in their consequences and import.

Like Andrea, Lilly had a sense that there might be a problem before the newborn-screen result came back. Baby Mia was so unlike her older brother, so much smaller and hungrier. But at the same time, she didn't actually believe anything could be wrong. "In the back of my mind [I] just thought, well maybe her digestive tract isn't all set up or ready to accept the formula the right way yet, and it'll just take a few days." Lilly

[17] Because these interviews were conducted after the children's diagnoses had already been firmly established, it is possible -- and would, I think, be perfectly natural -- that some of the recollections parents recounted with respect to this issue were influenced (maybe a little, maybe a lot, maybe not at all) by hindsight.

remembers – though it was thirteen years ago – that when the call from the doctor came, she was out in her barn. Her vague suspicions hadn't prepared her at all for what he had to say.

> He said he was very very sorry to have to call me and tell me this, but did I remember that in the hospital they did this heel test? And I, I remember them doing the heel test but I did not remember them saying what that was all for… and he explained to us that she tested positive for um ind- indications of cystic fibrosis. And I just broke down and started to cry and my husband was standing right there by me at the time… and I ran in the house cause she was sleeping in the house and we had the monitor by her and I just sat by her and said, 'Oh, my little baby, there can't be anything wrong with you.' And then I started calling relatives and trying to find out about it, what exactly it was. Cause I had heard of cystic fibrosis but I didn't know what it was… I was concerned about what was gonna happen you know, is she gonna be okay, what do I have to do, where do I go, what do I, what steps do I have to take, exactly what is it?

Getting a diagnosis in the first weeks when you're a first-time parent – whether you have suspected a problem or not -- is perhaps uniquely difficult, since there is neither experience with another child nor a history with the affected child to act as an emotional buffer. Paige describes -- with a tremor in her voice, though her newborn-screen diagnosis had happened more than four years before our interview -- how she had an instinct, as a mother, that something was wrong, that the baby was not healthy, that extra checks at the doctor were necessary. Nonetheless, when the doctor responded to her call of concern with the news that the newborn screen for CF had in fact come back positive and more testing would be needed, Paige was in a state of shock. "I didn't really know what cystic fibrosis was… I knew it was chronic, I knew it was fatal, but that was really about it…. It's overwhelming at first," she goes on,

> when a parent has their first child, just to learn about being a parent. And then on top of it to learn about this disease that has now overshadowed so much of our

> lives…. All I remember was it was like, all my world coming crashing down around me.…

Suzanne notes that where she lives, parents of first-born children with CF have a very difficult time, and that special support services have been set up for them by more experienced parents. Francesca, who doesn't live anywhere near Suzanne, would likely have benefited from such assistance. Instead, she had to cope on her own. "As a new mom," she recollects,

> you're always skeptical. Do I put her down, or you know should I go check on her? Should I not check on her? And with CF, it's like okay now I have this newborn and I have CF, I can't put this baby down. I have to sleep with her right next to me because I don't know what's gonna happen and what's going on. I think it just as a mom it made me a little more paranoid.

Paige had a very similar experience as a first-time parent.

> Kaya slept next to my bed in a little cradle, where I would fall asleep and my arm could reach over on her stomach so I could feel her breathing, because I was afraid she was going to stop breathing in the middle of the night, I was going to wake up and my baby would be dead, until she was seven months old, and just that fear is so intense.

Experienced parents on the whole seemed to share the perception that news of the diagnosis is somewhat less shocking if there's a child in the home already. For example Lilly, who had a healthy four-year-old at home when she got the newborn-screen diagnosis for Mia, imagines how difficult it must be to manage when one is a first-time parent. "You're going from no baby ever to having a baby and now trying to work all your life around… it and now you have to do all this extra health care with this baby."

After pausing, she adds "I think if it would have been my first child I would have felt like I missed out on having a normal child."

The "Cursed Blessing" of Newborn Screening: Timing and the CF Diagnosis

In her moving essay exploring the effect of newborn hearing screening on the beginning of her life as a parent, Jennifer Rosner reflects on what is lost as well as what is gained from the unsought diagnosis of a genetic disorder during the first hours or days of a child's life. "I have… had two years to live with my mixed feelings about the cursed blessing of newborn screening," she writes.

> At once the grateful beneficiary of information that has enabled Sophia to hear and speak without delay, yet left longing for the pure joy of new motherhood, I cannot help wishing, still, that I had had just a bit more time unravaged by the news, at least more than a meager six hours – time to sing lullabies to Sophia, without worrying about whether or not she could hear them (Rosner 2004, 21).

The heartfelt ambivalence about early detection voiced here is an aspect of newborn screening that is little considered in policy debates or research studies. By most existing standards (Wright et al. 1992), there is nothing subtle or debatable about Sophia's story. The early identification of her congenital deafness led to early medical interventions, which in turn enabled her to compensate for her hearing loss and to function in the normal range. According to the most widely-accepted criteria for assessing newborn screening (American College of Medical Genetics 2005), this case is a winner: the condition is identified at birth; there is available treatment; the treatment results in health benefits for the child and the family, and in cost savings for society. But for Jennifer, the immediate diagnosis also meant that she did not have a post-birth relationship with her daughter before she knew she was deaf. In coming to know her and to make decisions

about her health care she had "little else to go on but her deafness," no other dimensions of her identity to counterbalance this early signifier of disability. "Singing lullabies felt idiotic," she recalls. "Indeed, all impulses to make sound were stifled by sadness and anger before they could leave my throat…. That I had only six hours of untempered joy at the arrival of my baby was maddening." (Rosner 2004, 20)

When it comes to early diagnosis, there are criteria that matter to parents besides those outlined by the American College of Medical Genetics and other research and expert bodies. Though I certainly heard, during my interviews, a number of heart-wrenching stories from mothers whose children were diagnosed after symptoms developed (detailing suffering that was alleviated once a diagnosis was finally made, and improvements in health status that occurred once causes were identified and treatment begun), nonetheless, in a complex echo of Sophia's mother's lament over lost lullabies and the "cursed blessing of newborn screening," parents of children with CF also express the actual or anticipated loss of something very precious when diagnosis is made at birth.

Kaya was about 10 days old when Paige got the positive newborn-screen result. Before the diagnosis Paige had intermittently suspected there was a problem with Kaya's health. She also, however, had basked in the glow of "falling in love" with her gorgeous new baby. Those days have served, in the four years since the diagnosis, as a sort of reservoir of remembrance, a memory to return to when emotional refreshment or respite is needed.

> Do I wish I would have had maybe a month, maybe two or three months, where she was healthy and we didn't know she had cystic fibrosis? Absolutely! Because I express to my husband, I say this all the time, some days I just wish I could go back to that time, to that first week, her birth, being elated about her birth. She was so beautiful, she seemed so healthy, that first week where

> everything, we were this little family and we had our first little house, and everything was so perfect and just as planned. I, it was just so content and so peaceful, and I was so happy, sometimes I wish I could go back to that time, so absolutely I wish I would have had a month of that, or two or three, before the diagnosis.

Other mothers who got the diagnosis either at or before birth speak wistfully about wishing their relationship with their child had not always been colored by CF, wishing they had had a time free of the sadness and stress that the diagnosis brought. Crystal, who got a prenatal diagnosis for her son Martin, wishes she could have "just one day where I didn't know." When I ask her what that day would be like for her, she replies that she doesn't know, she can't even imagine. But that's precisely why she wants it. "I would like it," she supposes, "'cause even when I'm not thinking about cystic fibrosis, it's in the back of my head."

For parents who got a later diagnosis, the question of timing is viewed from a very different standpoint. These parents did have time with their child before knowledge of CF arrived – sometimes months, sometimes years; sometimes plagued by serious health issues, sometimes virtually without health problems or entirely symptom-free. Their responses to questions about what might have been gained, and what lost, by the timing of their diagnosis were complex. They all, each and every one, clearly believe that early diagnosis has beneficial results for the health of children with CF in general. They absolutely wanted to go on record supporting newborn screening as a matter of policy. At the same time, they spoke eloquently about the emotional importance of the time they had without CF.

Nancy and Paul capture very articulately the complex feelings a number of parents expressed about the timing of the diagnosis, that tricky push-pull between their own

experience and their knowledge of what professionals believe is state of the art in CF treatment. Their daughter, Alexandra, had been relatively healthy for her first year. Her growth had slowed some – a phenomenon noted empirically at the time, but significant only in retrospect. Neither her parents nor her pediatrician were overly concerned about her health or development before she suffered the rectal prolapse that led to prompt CF testing. The positive result shocked both the family and their health-care provider, since Alexandra had been so relatively healthy to date. Looking back on how things went, it's difficult for Nancy and Paul to separate out their own experience – which they both describe as pretty much optimal with respect to timing of the diagnosis – from what they know is being advocated in the professional world of CF. In Nancy's words:

> Newborn testing you know they say that that would be best, if she would have been diagnosed, I mean the earlier the diagnosis the better is what they say. But I don't know. I think about this sometimes and I just, it was great having one year of you know blissful ignorance basically, so I don't think I would have changed it. But I think the diagnosis came at a, a good time. I definitely wouldn't put it out any further… I'm glad, I'm actually pretty glad for when we got the diagnosis.

And from Paul:

> It's hard for me to think that any [other] way [of getting the CF diagnosis] would have benefited *us* more…. The only thing that I can think of is that with cystic fibrosis, scientifically and medically it is better to be diagnosed earlier, to get a jump start on treatment and maintaining good nutritional habits as well as eliminating unnecessary exposure…. I think only for the scientific and medical standpoint I would say it'd be better to know at least at birth.

Catherine, like a number of other women I interviewed, wishes she had had an earlier diagnosis for Joseph; he suffered terrible malnutrition during his first seven months, and she is certain that at least some of the multiple developmental issues he continues to

struggle with at age seven could have been prevented with early treatment. This was a terrible, a terrifically sorrowful consequence of how the diagnostic process unfolded for her. Even so, she knows she would have lost something precious if she had had, like Sophia's mother, only a brief few hours or days getting to know her son just for himself, rather than as someone with a genetic abnormality.

> I'm actually grateful for what I had [with a later diagnosis], because I did have my moment in the sun. Even if it was that one day in the hospital with all the guests and the flowers and the balloons and thinking I have this beautiful healthy baby. It wasn't robbed from me from the get-go where other people know right away and they never have that moment in the sun. They always have to be anxious, when is it coming, what's going to happen.

In Lorraine's case, the diagnosis of her symptomatic daughter, at age two months, precipitated also the utterly surprising diagnosis of her older son. Luke was two and a half when his parents learned his younger sister Jessica had CF. Lorraine and her husband had insisted – over the objections of a physician who resisted doing the test on such a healthy sibling – that he be tested too. For Lorraine this always-devastating process was made substantially easier coming as it did at a later age, when she knew already that CF need not mean poor health and suffering.

> I was actually really glad that my son was two and a half when they were both diagnosed because I, my own experience with my son was as a very healthy kid. And so when I got the diagnosis I didn't all of a sudden put him into a category that all of a sudden he's an invalid. And I had with my daughter because she was so thin and fragile at the point she was diagnosed… but having my son being so hale and hearty I knew that that was not necessarily something that was predestined. I knew that we had a CF kid who was perfectly healthy.

When parents remember their experiences with the timing of CF diagnosis, and imagine what it might have been like if the timing were otherwise, several other life circumstances are introduced over and over as important factors. One of these is whether the affected child is first born or not, as discussed above. Another is the vulnerability of the post-partum period.[18] For most women, this is a momentous and highly emotional time. The physical demands are always significant, and sometimes overwhelming. Between 50 and 80 percent of birthing women go through "the postpartum baby blues" during the first weeks after birth, experiencing a range of symptoms including tearfulness, depression, insomnia, emotional instability, elation, headaches, fatigue, confusion, and poor concentration (Rothman 1993, 311). Ten to twenty percent also suffer post-partum depression, a syndrome whose "usual symptoms are sustained depressed mood, heightened concern about her own and the baby's health, anxiety, somatic preoccupation, indecisiveness, fatigue, irritability, and sleep disturbance." (Ibid.) Getting or imagining a CF diagnosis during this alternately fragile and exuberant time presents specific challenges, which Paige describes like this:

> I had never been so happy in my life than the day she was born and then I just remember I-I went into kind-of a depression… I was there for her and … I was still a very good mom, but I was just sad a lot and I did require some, some medication to kind-of help me get through that phase of my life. That lasted about six months, where I just really couldn't snap out of it. I, oh, it was just very traumatizing… it was hard, I just kind-of remember being on autopilot… for a while.

Catherine says she has thought about what it would have been like to get a newborn-screen diagnosis, to find out about CF in that immediate period after birth when her son

[18] This study did not include any parents who adopted a child with CF. However, the issue of how adoptive parents get and respond to diagnosis is important to study and understand, and something that would be useful to take up in future research.

seemed perfectly healthy. "I think it definitely would have been like a punch in the stomach," she says.

> You're lying there… I was not well; other mothers might be basking in the glow of their baby. And then you are told that there is something wrong. I think things would have very quickly disintegrated because you know, there's also post partum issues there too and it could get really ugly. Cause you know the whole newborn thing anyway, you're tired, you're exhausted, you're recovering, you're this, you're that. You don't know what you're doing, you don't know what to expect and then to get a diagnosis on top of it, just seems even more looming and frightening.

Nancy concurs:

> [the NBS diagnosis] would have of course been just as shocking and I, I just think as a new parent that would have been almost too overwhelming for me. I was really overwhelmed with a newborn anyway….When we did get the diagnosis, it almost did feel like we had a newborn again. We had to learn so much about her and give her you know enzymes and figure out how to do this and how to do that…. I just think with a newborn it would have been, I can't even imagine what that would have been like. To have had both at the same time.

The CF diagnosis brings with it – among other things -- a crushing, sometimes disabling sadness, a chronic sorrow that, as Erica puts it, wasn't there before. "I remember waking for… nine months when [my children] were first diagnosed…," she goes on, "[and] within two minutes it would hit me and I felt like I was kicked in the stomach…. It wasn't a death, but it was the death of a dream of a healthy child." For Kim, the depression she fell into after the diagnosis was debilitating. "I don't know how I would be today," she says, if she hadn't gotten help in the form of medication.

If a child is ill, there can be no question of separating the moment of diagnosis from the post-partum period; the disease is manifest, and must be dealt with immediately. But

what is the effect of layering the genetic diagnosis of an apparently healthy newborn on top of all the other post-partum issues? We know that young children can be significantly affected by their mother's post-partum depression, but the current literature does not seem to address the relationship between this well-documented phenomenon and newborn screening. Evidence from my study suggests such research is warranted.

Going Public with the Diagnosis: Social Construction of the Child's CF Identity Within the Family and Community

Children belong, in some sense, to their extended families and communities as well as to their parents. Most children have several adults in their world – and sometimes many -- who care for them and about them, who want the best for them. [19] These adults will have their own spectrum of reactions in the face of substantial threats to the child's health and well-being. CF is one such threat, and parents in my study – particularly those who received an immediately-post-birth diagnosis -- clearly indicated that the process of telling family members and friends about the diagnosis was an important aspect of their own experience at that time.[20]

Parents of children who receive a CF diagnosis manage the dispensing of bad news in a variety of ways, of course. Some share with others their fears and suspicions, the worrisome signs or medical news they receive, right from the beginning. Others wait until test results are in and the situation has been more fully clarified before bringing

[19] Of course, this is by no means a universal truth. However, it is a generalization that holds for the group of families who participated in this study.

[20] The importance of family and community in coping with the diagnosis, and in constructing the child's identity as a person with CF, emerged through first-level analysis as I conducted my research. I then proceeded to revise my interview guide, and to collect richer and more complete data in this area from parents I talked to later in the study. For a future project, it would be worthwhile to go back and ask participants from the earlier part of the study to talk more about the role family and community played for them.

even intimates inside their trouble. Some look to people outside the nuclear family for support, assistance, advice; others feel they have to "manage" the reactions of other people and that giving them information is therefore a kind of burden. Certainly part of this variability is attributable to personal and interpersonal style and psychology; to how physically and emotionally close the parents are to family and friends; to what else is going on in everyone's life at the time. But also important is how, when, and within what context parents themselves receive the news.

For obvious reasons, parents whose children have had chronic health problems over a period of months or years before the diagnosis are least likely to be alone with their sense that there is a problem by the time the diagnosis comes. Often these parents have already talked with others about their concerns, asked for help, sought referrals to health-care providers, and otherwise "gone public" with their knowledge that something is wrong – or they suspect something is wrong -- with their child. Sometimes other family members and friends have seen evidence of the child's poor health or impeded development first hand. By a process of accretion, the child's identity has already begun to be constructed -- both within and outside the household -- as someone with health issues. The CF diagnosis is often, as noted above, more a confirmation than a surprise -- not just to the parent, but to the extended family and community as well. Knowing that CF is to blame can still be a devastating shock for everyone involved, but it is unlikely to bring with it a complete reconstruction of how people understand and perceive the child. These parents therefore spoke little about the challenges associated with telling others the news, focusing instead on the role of people beyond the nuclear family in connection with caregiving; with the impact of the diagnosis on relatives' reproductive decisions and

sense of self;[21] and with the negotiation of shared space at family gatherings and in other settings where contagion is a concern.

Parents who receive a positive newborn screen for a child with no, or only few and private, manifest health issues are in quite a different position when it comes to telling family and friends the troubling news. With the call from the doctor's office, a set of complex interpersonal dynamics is immediately set in play. Parents wonder, do we tell other people about the positive screen, or wait for the confirmatory sweat and/or genetic testing? Do we wait until we know more about the condition and its implications, or ask others for help navigating this new terrain? How will family and friends respond to the news, and how will their response color my own as I adjust to my new knowledge? And underneath all this -- implicit yet palpable in the interviews: Will telling other people make this unreal news more real? Will it make my baby forever a sick baby instead of a well one in the eyes of the world, and thus in actuality?

Shannon, the mother of a well child with CF introduced above, felt acutely the difficulty and confusion of "going public." She describes wrestling with what to tell people, when to tell them, and the subsequent need to cope with their responses, as "the hardest part of [the] entire process" – quite an assertion for a parent going through an

[21] The fact that identification of genetic disorders in one family member influences not just that individual but the entire family is well-documented in the genetics literature (see for example Kerr and Shakespeare 2002; Marteau and Richards 1996). Disclosure of positive test results is thus a complex process for all involved, since others besides the diagnosed person can suddenly understand themselves to be "at risk" as a result. A number of parents I interviewed talked about these dynamics in one way or another. For example, one mother recounted how "When you start telling [family members] it's genetic, well then some people can kind-of get a little freaked because it's like what do you mean, that means I could have it or they could have it, you know… people are really shocked when you tell them that like 1 in 28 people's a carrier because that's a really high number." Another mother talked about how difficult it was to deal with family members whose primary response to the diagnosis seems to be "How's this gonna affect me?" Analysis of these data would be an interesting future project. However, for the purposes of this dissertation, I am choosing to bracket this discussion and focus instead on data related to the role of other people in constructing the child's identity and the parents' experience of the post-diagnosis period.

experience so profoundly difficult in so many ways. In the time between the positive screen and the confirmatory diagnosis, she and her husband

> didn't tell anybody what was going on because we didn't have enough information, as far as, not because they didn't provide us, but we didn't, we didn't want to tell people, a bunch of people that there might be something when there might not be…why get everybody worked up, and also, too, it is [a] very… hard thing to explain because nobody has any experience with the disease….

Again and again Shannon returns to her reluctance to share the screening results, to get other people involved. Partly, she was being self-protective, wary of their reactions. "I've got enough to keep my own emotions in check right now," she says. "I don't need to have to try to…take care of ya'll, I need to take care of myself and my kids and my husband and that's it." But during the time between the screen and the test there was also a deep hesitation about putting Margo's public identity in question, bursting the bubble of congratulatory joy that accompanied the baby's birth and replacing it with… what? The idea of a daughter who seems well, adorable and chubby, but is actually carrying a potentially deadly genetic disorder within her week-old body? A future that suddenly seems ominous, yet is unclear in terms of actual implications? Shannon and her husband, like many parents in this situation, were themselves swinging back and forth between hope and fear, between a sense that nothing could possibly be wrong and a deep foreboding that something was. Why encumber the process with the weight of other people's responses? Why give other people the power to participate in constructing the child's identity while the parents are still struggling to do so themselves?

After the confirmatory test, going public brings another set of issues. The reality of the genetic mutation is now irrefutable from a scientific standpoint, but the process of understanding what this means, of conceiving who this child really is and what role CF

will play in her life – that has just begun. Parents must begin at this point to cope with the diagnosis themselves, to hold this shocking news about their child's life and identity in whatever way they choose or are compelled to hold it. Other people, when they are told and therefore unavoidably included in this process, can disrupt the parents' preferred modes of adjustment in a number of ways. Parents of newborns sense keenly how fundamentally the baby's identity is "socially bestowed, socially sustained and socially transformed" (Berger 1966, 98), and they endeavor mightily to protect both themselves and the baby by controlling this process during infancy as much as possible. However, their capacity to manage the responses of other people is limited, and the resulting distress can be significant.

Betty recalls that for her, telling other family members about the diagnosis – which came shortly after birth, for a child already quite ill with MI and other complications – was most difficult because others would react more strongly than she. They would see CF as more dire, and Rose as more vulnerable, than Betty herself did. "It was just a really, really hard day," she recalls,

> going home to tell other people, because my parents were watching my other daughter that night and it happened to be their wedding anniversary that we found out. Go home on their wedding anniversary and tell them, by the way she has CF? And of course they don't know what it is, and we're not in a position yet to know what it is exactly ourselves. And it was just really hard to call, you know it's harder to call and tell everybody else…. I just sent out a group email to a bunch of friends and family. I mean I didn't have it all together to tell everybody…. Their reaction was probably the hardest part, rather than you know hearing it ourselves, because it made them feel helpless, it made them not know what to say and I think that they felt worse for us than what we felt for ourselves.

For Betty, going public presents the risk of having her child's situation be immediately construed as disastrous in that tender time when the baby and the diagnosis

are both new. For parents reeling from the news themselves, parents with "little else but the diagnosis to go on" in apprehending their infant and getting to know her, it's not helpful to have others predict or foresee a bleak future, a limited life span. It's not helpful to "get people calling and saying 'we hear your baby's dying'" (as one mother reported) while they hold and tend their newborn. Such responses, or the fear of them, force families inward, changing the newborn period from a time of shared joy to a period of self-induced isolation, a time when you "don't wanna… talk about [the baby and the diagnosis], not, I shouldn't say not being able to, but choosing not to."

Paige's articulation captures with fullness another common response – the sense that others cannot truly comprehend the seismic shift the early diagnosis has caused, or how it has transformed what it means to mother the new child.

> In the beginning everybody's in shock. And your whole family hears about the news, and there's… visitors pouring in to visit and… sympathy cards of 'I'm so sorry,' and phone calls of 'oh my gosh I can't believe this is happening,' and family members who want to buy air purifiers for the home or give money for this or make you meals. But then after it settles in… everybody goes on with their day-to-day life, which is normal. But your life isn't normal anymore… not normal, *per se*. And now you're doing, people don't know, you're getting up in the morning, you're doing breathing treatments, you're doing chest physiotherapy, and you're dealing with emotional issues, and financial [issues]. I mean the dynamics of the disease are huge, and those phone calls stop… and the in-kinds stop, you know. And that's normal, but it's just so lonely….I can't tell you how many times I would have just loved to just pour out all of my feelings and cry about it, and I think people are hesitant to ask….it's just very, it's just very lonely, with cystic fibrosis.

Like women who find themselves taking that self-protective turn inward after the diagnosis, mothers who experience the awkward falling-off of inquiries and support end up feeling very much alone, walled off from loved ones and community. As a parent interviewed by Taner-Leff and Walizer puts it, "One is never more alone than after one's child has… been found to be ill or disabled. One encounters rejection and distancing

from others – even those from whom one has a 'right' to expect better. One withdraws, bruised and hurt." (1992, 98) When the diagnosis becomes, as it inevitably must, a public as well as a private event, it affects not only the baby's household (see Chapter Four), but all of the "embedded contexts" -- household, extended family, community, social support systems (Bronfenbrenner 1979) -- in which early childhood emerges.

Only one parent in my study described "an act of social recognition" (Ibid., p. 99) after an early diagnosis that made her feel less instead of more isolated. Crystal received an unsought prenatal diagnosis during the second trimester of a pregnancy she was always determined to carry to term. Her anticipatory grief at the news, the way it stunted her joy in the pregnancy, was palpable. "I couldn't sleep at night," says Crystal.

> I had a hard time eating, and… I found myself… not having like happy thoughts about having a baby, just dread…. I felt like when I was walking around I didn't want people to see that I was pregnant, I didn't want them to ask me questions, I didn't want to talk about it… I didn't have a baby shower.

She did trust and confide in her mother, however, and after a while that confidence bore very precious fruit.

> I was staying with my parents [during the pregnancy] and one day [my mother] got out her knitting needles, `cause she always knits the babies a sweater. And she started knitting a sweater, and… I realized she was seeing something happy, you know… so like a few days later we were out shopping and I bought the baby a hat, and it just felt really good, like something normal to do.

This chapter has begun to examine the differences in experience between parents who encounter illness before a diagnosis, and those who get a diagnosis at the very start -- sometimes long before any symptoms, but always before the child's identity and social

standing have been clearly established apart from CF. The latter experience became increasingly common as *prenatal* diagnostic tests were introduced and quickly became the norm beginning more than 20 years ago (Katz Rothman 1986; Rapp 2000). Now, with the rapid growth across the states of *newborn screening* for CF and myriad other genetic conditions, mass screening and pre-symptomatic testing affect ever-larger numbers of parents. As a result, the relationship between parental knowledge of the child and scientific knowledge of the child, between the experience of CF as a physical illness and the experience of it as a diagnostic prediction, is shifting again. The next three chapters explore this shift from various angles: how diagnostic processes themselves shape experience; how parenting practices are influenced by diagnostic information; and finally, how relationships between parents and health-care providers, between parental knowledge and professional expertise, are affected by the change.

Chapter Three

The Diagnostic Process: Procedures, Timing and Consent

CF is described in the medical literature as the most common genetic disorder affecting people of Northern European descent, and among the most common in the U.S. population at large. As noted in Chapter One, 1 in 3,700 newborns in the general population has the genotype (CDC 2004). Between 20,000 and 25,000 people in the U.S. are living with the disease (CDC 1997). While these statistics are well known to clinicians, most people outside of medical circles still don't know much about CF. Downs Syndrome is a genetic disorder that has gradually made its way into the public consciousness, and certainly the inherited factor at play in breast cancer rapidly became common knowledge after the BRCA gene was identified in the 1990s, though it in fact contributes to the disease in only 5-7% of cases (Katz Rothman 1998). But CF has not yet become part of the popular lexicon of risk, prevention and disease. It is not something parents actively worry about during pregnancy, nor is it something they are likely to think of as the culprit if their child develops one or more early symptoms of the disease -- especially since these symptoms can also signify many other childhood illnesses.

In my study, most parents either had never heard of CF before the testing process began, or had only a hazy sense of what it is.[22] No parent had any specific concern about her child's risk for the disease ahead of time. In this context of little or no knowledge from elsewhere, the diagnostic process itself -- the rolling out of procedures designed to

[22] The one exception to this generalization is Andrea, who had done extensive research on her own to determine what was wrong with her sick son, and arrived at the conviction that he had CF well before his health-care providers agreed to consider seriously that this might be the cause.

determine if the disease is in fact present and the current technological and policy arrangements that shape them -- can loom very large for families. This chapter first describes how that diagnostic process varies for parents in differing situations, and then examines its various aspects and their impact on parents' experience.

Current CF Screening and Testing Procedures

Diagnostic procedures for CF vary significantly depending on when the process is initiated. Here are the various scenarios that face parents and their children while they are undergoing testing.

Diagnosis with Symptoms

If testing for a child is prompted by clinical signs potentially consistent with CF – i.e., "one or more characteristic phenotypic features of CF" – then clinicians will move directly to a confirmatory test (Moskowitz et al. 2005, 2-3). This may happen at any time in the child's life, but the median age of CF diagnosis on the basis of symptoms and signs other than MI is fourteen-and-a-half months (CDC 2004, 4). Parents may have been observing their child's symptoms for quite some time, or symptoms may have emerged relatively recently; symptoms may already be severe, or they may still be mild. Most often what is performed is a "sweat test," a "simple and painless procedure [that] measures the amount of salt in the sweat." Results of this test are available to parents within hours of when it is performed, and CF is diagnosed if the salt level is high. (http://www.cff.org/about_cf; accessed June 18th, 2005). Alternatively, or if sweat testing is inconclusive (as it not uncommonly is), the child's blood might be sent off to a laboratory where it will be subjected to DNA mutation analysis. If two disease-causing

mutations are identified as present in the cystic fibrosis transmembrane conductance regulator (CFTR) gene, then a CF diagnosis is established (Moskowitz et al. 2004, 1-3).

Newborn Screening

When newborn screening prompts the chain of events leading to diagnosis, the process is quite different. Blood will have been taken from the heel of the newborn in the hospital before discharge,[23] and sent to a state health department or regional laboratory to be screened for whichever genetic conditions are included in the newborn screening panel mandated by law in that particular state. As of 2005, in all thirteen states that mandated newborn screening for CF, the protocols begin with a test that measures immunoreactive trypsinogen (IRT), a possible indicator of CF, in the dried blood that was taken from the infant. Standards for what is considered an elevated IRT vary from state to state. If the newborn's IRT levels placed her/him in the abnormal range for the state in which she/he was born, a second test will be run. In some states (eight of the thirteen with CF screening programs), the second test is a DNA analysis of the same blood sample that was originally obtained in the hospital. DNA analysis may look for only the most common CF mutation (DeltaF508), or for multiple mutations.[24] If one or more mutations are identified, the baby's doctor, and/or the clinic or hospital of birth, will be notified of the positive screen.[25] The infant's parents will then be contacted and told to bring the infant in for further testing – using the same sweat test described above.

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[23] Blood specimens taken from the newborn before 24 hours of age will have only limited accuracy. Those collected between 24 and 48 hours after birth are considered "satisfactory," and those collected between 48 and 72 hours are "optimal" (NYSDOH 2003, 2-1). However, given that most hospital stays are less than 72 hours, specimens are usually collected on the second day of life.

[24] New York, for example, analyzes for 33 mutations (NYSDOH 2003, 7-9).

[25] In some of these same eight states, even if *no* mutations were identified, a sufficiently high IRT by that state's standard will result in parental notification of a positive screen. This is because mutation analysis detects only a fraction of the possible mutations of the CFTR gene, and some states consider that an

In the five remaining states of the thirteen that test for CF, an elevated IRT will prompt direct notification of the baby's health-care provider, and then of the parents. In this case, parents will be told to bring the baby in for a second heel prick when she is about two weeks old so another IRT test can be run. At this age, "values are more specific for CF because IRT values decrease with age in infants without CF" (CDC 2004, 6). If the IRT is still above whatever value the state has set as a cutoff for this second screen, the child will be referred for confirmatory sweat testing (Ibid.).[26]

So with newborn screening, a positive screening result is just a first step, a possible indication of a problem. In one state, that result may reach the parents after just one test, and the baby may then have to undergo a second IRT screening test and then -- if indicated -- a third test to confirm or rule out the diagnosis. In another state, the child's blood may have already been "double tested" by a combination of IRT and DNA analysis by the time parents know about it, and the number of additional tests and waiting periods for families is cut in half. But whatever the next step is, that first positive screen result certainly does not mean that the child is actually affected. In fact, as noted in Chapter One, the chance of being among the "false positive" group is far greater than the chance that a given child with a positive screen actually has CF. Even the state reporting the

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extremely elevated IRT may indicate the presence of a mutation not included among those for which the analysis tests.

[26] It is striking what a perfect example this is of how micro-processes structure scientific definitions of disease. In this case, it is also striking how widely these processes, and therefore the resulting definition of disease, can vary, even within the United States. For a fascinating examination of how "genetic explanations play a role in the reclassification of Cystic Fibrosis" at the more general level, see Hedgecoe's 1993 article "Expansion and Uncertainty: Cystic Fibrosis, Classification and Genetics." In this paper, Hedgecoe argues that the availability of genetic tests for CFTR mutations played a critical role in constructing male infertility as a form of CF. "[T]he CF classification system has settled down to include certain cases of male infertility where mutations in the CFTR gene have been detected," he concludes. But this outcome "… should not obscure the socially constructed, contingent nature of this (and any other) classification system. CABVD [i.e., male infertility where the CFTR gene is implicated] does not *have* to be classed as a mild form of Cystic Fibrosis: 'Typically, the genotype in those patients [CBAVD] consists of at least one very mild mutation uncharacteristic for CF patients' (Zielenski 2000, 126)" (1993, 59).

lowest false-positive rate identifies only one true CF positive for every five positive screens. In other states, the ratio of false-positive to true-positive cases ranges from 9.5:1 to 25:1 (CDC 2004, 9).[27] To give a more concrete example, in New York State in the year 2003, a total of 1,384 children were "presumptive positive" for CF based on the screening result. Only forty-eight of these – or a mere 3.4% - were actually confirmed positive (New York State Department of Health Annual Report 2003, http://www.wadsworth.org/newborn/annualrept/annsum.htm).[28]

The public health system responsible for conducting the newborn screen for CF does not communicate with parents directly.[29] Rather, the laboratory that ran the screen contacts the baby's pediatrician (if one was indicated on the newborn screening form filled out at the hospital); and/or the hospital where the child was born; and/or a local geneticist or approved "specialty center" for cystic fibrosis where various health care professionals with expertise in treating the condition are gathered under one umbrella (Mandl et al. 2002; NYS Health Department personnel, 2005). This process seems to

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[27] As political scientist Diane Paul points out, the rate of false positives is very difficult to compile reliably, given "the lack of consistency among jurisdictions in what counts as a false positive result" (Paul 1999, 10 of 18). However, as noted in Chapter One, the CDC estimates that for conditions other than CF, the number of false-positive results is often greater than *fifty* times the number of cases of diagnosed disease (CDC 2004, 9). The rate of false-positive screens for CF is thus considered moderate in comparison to rates for other conditions.

[28] Cumulatively in that same year there were 21,151 babies "presumptive positive" for one of the eleven conditions for which New York State screened at the time. A total of 572 of those were confirmed positive. Another 11,701 of those infants were determined to have sickle cell or a related trait (i.e., they have only one copy of the mutated gene, and are therefore carriers; some of these children may also have some relatively mild form of symptoms). 817 were identified as carrying HIV antibodies. (NYDOH Annual Report 2003, http://www.wadsworth.org/newborn/annualrept/annsum.htm).

[29] As far as I can tell, no state's newborn screening program contacts families directly with positive screening results for CF, probably because the presumptive positive for this condition is not considered a medical emergency. Mandl et al. found that in addition to notifying the infant's doctor of all positive results of all screening tests, nine states notified the infant's parents directly of positive results for some tests. Eight notified the geneticist in addition to the infant's doctor, and twenty notified the birth hospital in addition to the infant's doctor (Mandl et al. 2002, 270).

take between one and three weeks on average after the heel stick is done[30], though it can take longer, especially if the pediatrician requests a repeat analysis of the blood the laboratory already has before contacting the parent. Once the child's pediatrician, clinic, or birth hospital gets the result, someone from one of these settings will contact the parents to tell them that the screen was positive and that further testing is necessary. Usually this news will be delivered by telephone, although in some cases – as happened with Francesca (see Chapter Two) -- the pediatrician might ask the mother or parents to come into the office in person. It might be a health-care provider the parents already know on the other end of the line, or it might be a complete or comparative stranger.

Confirmatory testing, usually by sweat test, is generally done at the CF specialty center. As is the case with children diagnosed clinically, if the sweat test is inconclusive, the child's blood or saliva will be sent to a special laboratory equipped to conduct mutation analysis in order to confirm or rule out CF. The time from when the parents first hear that the screening test was positive to when the diagnosis is ruled out or confirmed can be anywhere from a week to six weeks or more, depending on how many steps there were in the process; whether any tests were inconclusive; and how long it took to schedule appointments. The child's diagnosis by newborn screen may come any time between two weeks of age and eight or ten weeks, but the average age at diagnosis is .5 months according to the CDC (CDC 2004, 4).[31]

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[30] This assertion is based on what I could gather from my discussions with newborn-screening staff, and from my interviews with parents. I was not able to locate any published research calculating the average time, nationally, between when heel blood is taken and when results are made available, nor could staff at the National Newborn Screening and Genetics Resource Center give me these data or tell me where I might find them.

[31] NYSDOH seems to disagree with this estimate on the part of CDC. It estimates that the average age of those diagnosed by positive screening is twelve weeks (2003, p. 7-8, http://www.wadsworth.org/newborn/annualrept/annsum.htm).

Prenatal Testing

If parents get a CF diagnosis for their fetus prenatally, the testing route will be entirely different from that of either newborn screening or post-symptomatic testing. As a first step, the baby's mother will be offered carrier screening during the first trimester of her prenatal care. If she agrees, or if she at least signs a form that says she has agreed, her blood will be analyzed to see if she carries a mutation in her CFTR gene. If she is found to have a CFTR mutation, she will be notified and carrier testing (by the same route) will then be offered to the fetus' father to see if he too has a single mutation, since the fetus will be at risk for the disease only if it is possible to inherit one defective gene from each parent.[32] If the man is a carrier as well, then the woman will be offered amniocentesis, a more invasive procedure whereby amniotic fluid is extracted from her womb and analyzed to determine how many copies of the mutated gene – if any – the fetus has. This three-step procedure, with waiting periods associated with each step, can take up to two months from the time the mother learned of her positive carrier-screen to the time of a confirmed fetal diagnosis.

Parents' experiences with finding out their child has CF thus vary tremendously based on *when* they get the diagnosis, and on *how* that diagnosis is therefore delivered. Whether children with a later diagnosis have been symptomatic for a long time or for just a short time, CF is established as the suspected problem in a clinical setting. Health-care providers speak with parents about their recommendation for sweat testing, and make the

[32] As noted in Chapter One, CF is an autosomal recessive disorder (see page 38). This means that people who carry one copy of the defective gene will be "carriers" of the disorder but will not be symptomatic. In order for the disease to be expressed, two copies of the mutation must be present. If a child's parents are both carriers, the child will have a 25% chance of having no copies of the gene, a 50% chance of having one copy, and a 25% chance of having two copies.

appropriate referral immediately. On the other hand, parents with early diagnoses, whether by newborn screen or by prenatal testing, are often unaware or only dimly aware that CF testing had been initiated at all. Once a first positive screen is returned, they receive information piecemeal over an extended period of time until the actual diagnosis is established.

The Waiting Period

The length of time between suspected and confirmed CF diagnosis is one important factor in the testing process that varies significantly depending on whether testing was initiated in response to symptoms or not. All parents feel a sense of urgency about performing the test once CF is mentioned as a possible diagnosis. However, the time between a recommendation that the test be conducted and the test's completion is usually very short for parents with a later diagnosis -- a matter of hours or days, rather than the weeks it takes when testing is done prenatally or via newborn screen. If their child has been symptomatic for a long period of time, these parents may have already suffered a devastating diagnostic delay because of health-care providers' overall failure to take their observations seriously and recognize the reality of a problem (see Chapter Five). But the delay is in realizing that CF might be the culprit at all, not in getting through the various procedural stages between a positive screen and an established diagnosis. In my interviews, seven out of eleven parents[33] with a later diagnosis described enormous stress before CF was identified as a potential cause, but none of them described problems associated with the actual testing process itself once it was put in motion by their health-

[33] Both the "seven" and the "eleven" numbers here include the mother I interviewed whose child does not in fact appear to have CF, but who has certainly undergone a "diagnostic odyssey."

care provider. This is consistent with the research of Al-Jader et al., who found both that all parents are "extremely anxious while waiting for the sweat test to be arranged," and that those with a newborn-screen diagnosis are far more likely to experience a distressing delay than are those whose children are tested once symptoms have emerged (1990, 462).

Lorraine was the only parent with a later diagnosis who mentioned the threat of any sort of unplanned delay in actually obtaining the CF test.[34] Her need to act quickly, to circumvent a waiting period if at all possible, illustrates just how strong the need for immediate action is for most parents once they are alerted to the possibility of CF.

> [The doctor] said, 'do you have any cystic fibrosis in the family?' And we said no. And she said, 'Well, I want to test her just to rule that out.' It was a Friday afternoon, though, on the 4th of July weekend and she said 'Well, you're not going to be able to get a test set up now though so you'll have to wait [until after the weekend].' And I was like 'No, we're going to get a test done.' So we got on the phone to various hospitals and through a combination of pleading and making our case and tears and all other types of discussion with various lab techs we got some poor guy who agreed to stay late to administer the test to my daughter. So we ran over there and he administered the test on two arms, it was a sweat test.

Parents who receive a newborn-screen result and those who get the diagnosis prenatally share the burden of longer waits, given the multi-stage nature of the testing process and the time that inevitably elapses between the steps. For these parents – in contrast to those with a later diagnosis, who are generally most focused on lived experiences with symptoms -- each step in the diagnostic process itself looms very large in experience and memory, and they all agree that the waiting is terrible. It is "full of tears" and "a lot of sleepless nights," full of worry and anticipatory grieving, even while

[34] One other parent was advised by the doctors to have her child put on weight between the suspected diagnosis and the sweat test, but this was a planned delay and one she did not seem to find objectionable.

there is yet hope that the next screen or the next test will be negative. Kate, who got a prenatal diagnosis, describes the waiting period like this:

> We started on this up and down emotional thing, and I'll tell you that period between me finding out I was a carrier and actually getting the results of the amnio was the worst time of any time. Even knowing the official diagnosis was better.

Parents in my interviews thus certainly corroborated data from a study of one hundred and four parents in Wisconsin who indicated that while waiting for sweat test results after a positive newborn screen they felt concerned (96%), depressed (77%), and shocked (76%) (CDC 2004, 24).

As I discussed in Chapter Two, difficult aspects of the waiting period include the way the new child's identity is held suspended during this time, and the social isolation parents experience as they wait for the final outcome. A third major source of distress that parents talked about repeatedly is unmediated encounters with outdated information about the disease. People waiting for more CF testing can certainly, like one of the parents quoted by Taner-Leff and Walizer, be driven to distraction by the combination of "the professional brush-off," and the "lack of information[,] especially current information…" (1992, 23) that is characteristic of the waiting period.

Often, the pediatrician or other health professional who communicates the positive screen result to the parent provides only the most minimal information about what CF is and what it might mean for the baby. This is partly because the structure of the process does not allow for in-depth interaction (see below), and partly because the provider in question may know little about the condition herself. The general lack of knowledge about genetics and genomics among primary care doctors is well documented (Greendale

and Pyeritz, 2001), as is limited familiarity with CF in particular (Boulton and Williamson 1995). Add to that the astonishingly rapid rate at which the number of conditions being screened for at birth is increasing, and the chances that a doctor can responsibly tell parents what they need to know regarding a positive result shrink even further.

Parents are often left to research CF on their own, then, after they find out about the abnormal screen – left, as Anthony puts it, "just trying to understand it without having [a] professional guide, or somebody who'd seen it many times before and could give us more concrete information." Paige, for example, says "After I got off the phone with the doctor, I had picked up an old medical book. The copyright was like 1982, and it was talking about cystic fibrosis, and life expectancy was eighteen years old and this and that, and it was just very grim." And from Shannon, "The data is changing so much that if you read anything with any age to it, you know, you're gonna read a death certificate… there's so many people who still have them dying before they're thirteen." Catherine, who also had a waiting period between the recommendation for a sweat test and its completion, found the predictions similarly dire.

> [W]hen you read the literature it's scary, man. They're basically telling you they [the babies] are going to die. And it's going to be scary, and it's going to be suffering, and it's going to be hard. And you need help. And its like, there's no gleam of a normal life in there at all.

Sometimes physicians will warn parents that there is misinformation out there, and advise that they stay away from the internet in particular. But parents are generally desperate to know more, to learn whatever they can as they wait for further tests, and they

are therefore vulnerable to misinformation and the terrible anxiety it breeds. As Lorraine puts it,

> For me the most important thing is to get people who are learning about CF the most accurate current information about the disease and in the beginning. I don't think I… got that. I only got it when I went to an internet web page that hooked me up to other parents. Because… when I read you know official things, even the CF foundation web page… started out with something like 'CF is the most common incurable fatal genetic disease.' That was the first line of the CF foundation web page. And in fact as I've worked with them over the years I've gotten them to change that because I don't think that should be the gateway. Having you worry about this disease. I mean you should not have that kind of preconception about a disease that at this point I have a lot of confidence [will] become a highly treatable disease that people live well into adulthood with.

Getting the Diagnosis

Another major difference between those with an earlier and those with a later diagnosis is the way in which the news is delivered. Parents with a later diagnosis are generally on site in the health-care provider's office or hospital when the sweat test results come back, or else they have agreed with the provider ahead of time on clear follow-up procedures: how to get the result by phone, what to do next if it is positive. The diagnosis may still be a shock, but it is unfolding in the context of the symptoms that prompted the test to begin with. As Nancy describes learning about CF after her relatively healthy child had a rectal prolapse at age one,

> … so when we took her in to her pediatrician she ordered a sweat test and sent us to the GI people to figure out what to do with this prolapse. And they admitted us to the hospital because they needed to lightly sedate her because when they would reduce it, or push it back in she would you know feel it…. And so after she was admitted to the hospital a few hours later the pediatrician came back and said that the sweat test was positive. And she said herself she would have bet a hundred thousand dollars that it would have been negative because other than being a little small you know she'd had one respiratory infection but got over it with no trouble and otherwise was healthy. So it was quite a shock…. she stayed one night for [the rectal prolapse] and then so then the next day we got the spiel from the CF

> doctor and the nutrition and then what not on 'this is what cystic fibrosis is and this is what things are going to be like.'

Erica too was told about her child's diagnosis in the health-care setting, and was connected immediately with the specialty resources she needed to begin addressing her daughter's symptoms. As a parent confronting CF for the first time, Erica found -- as have other parents receiving diagnoses before her (Taner-Leff and Walizer 1992, 95) -- that face-to-face contact with doctors, "that personal touch… the person in front of you" was invaluable. So was immediate access to other resources.

> I needed that information at that point, I think, as a new [CF mother]. I think parents need to leave with papers, you need to walk out with something in your hand. Also just an appointment makes a difference because you feel like, after you get your diagnosis, like you are blown away. It doesn't matter what it is. It could be leukemia, anything, but you need to know you are going to another doctor because then you feel like, you feel like it'll be ok.

When Erica subsequently tested her younger children to confirm her sense that they too had CF, however, she asked for and got the results over the phone. In those cases, she says, "I liked the phone thing better…because I knew already [that the test would be positive], and I think it was also, for me, it is more private…. And I was able to just hang up the phone and cry."

Parents receiving news of a positive newborn screen, on the other hand, generally have no way to walk out of the conversation with "papers in their hands." Most often, notification that their child had an abnormal result for CF, and that either repeat screening or confirmatory testing is needed, arrives by phone. As I have already shown, this call arrives entirely unexpectedly, even for those who suspected something might be wrong with their infant – a sudden reach from the medical world into their homes without any

advance warning. This relatively impersonal way of communicating potentially life-altering news is objectionable for many parents. Joan recounts her experience like this:

> When she was about 2 weeks old we received a call from her pediatrician and her pediatrician informed us that one of her newborn screenings had come, that her pancreas wasn't working, and that she was presumptive positive for cystic fibrosis and that we had to follow up with sweat test. And he kind of made me mad because he told us over the phone. It was kind of like, 'get her to the hospital because we need to do tests.' It just wasn't the correct way to tell us because we were like 'What is cystic fibrosis? Is she going to die?'… I think that could have been done in a better way. I think he could have explained a little more what it was…. I think he could have done it in person. I think over the phone was a little impersonal…. Especially something that is affecting your child and is going to affect them for the rest of their lives. And you can't tell me in person?

Anthony had a similar response to the phone call. His pediatrician had asked that the lab repeat the screening test twice more after the first positive screen in an effort to be sure she was not dealing with a false positive. When she got the positive screen back for the third time, it was on Christmas Eve, just before she was due to go off on vacation. She called the family to tell them the news, and then left on her trip. "I think I was grateful knowing it's been several times before she shared any information with us," says Anthony. But he goes on:

> I guess the down side to that was perhaps she could have shared the news a little bit differently with us. You know it wouldn't change any outcomes, [my daughter] wasn't symptomatic in anyway. There was nothing that was necessarily immediate that maybe was reason for alarm. But I guess if she, had shared with us a little sooner so that it wasn't a phone call you know sort of at the 11th hour, and 'Sorry I'm not going to be around to answer any questions, but you know here [the screening result] is. And there's resources out there available for you, there's resources available online and so on. And you don't have to do this right away but you might want to look into finding a new specialist, I know [name of hospital] has a great clinic.' That was the kind of information we received from our primary care [doctor].

Anthony is voicing here a dissatisfaction with the standard method of communicating positive screen results that most parents in my study who shared this experience echoed. Newborn screening programs, and the physicians who get the results, have developed systems for dealing with positive screens that are generic, that help them cope with a large number of tests as efficiently as possible. From the perspective of the system, it is just too costly and work-intensive to arrange a home visit or even an office visit so that each abnormal screening result can be explained in person. After all, the number of positive screen results is very large, and the vast majority are false positives and therefore signify little or nothing about the actual health of the baby. But for parents, for *each* parent getting the news that something may be terribly wrong with his or her child, the statistical likelihood that the positive screen does not indicate a real health problem is little comfort.[35] As Crystal (who, ironically, is a statistician by profession) put it, "I know odds are different from… reality, what actually happens… I mean at that point I didn't care about one in four, I just cared about my baby."

Education and Consent

During the diagnostic process and beyond, as Anthony indicates in the quote above, information and knowledge matter tremendously to parents. How much they knew about the testing and the disease at each stage, and how they came to know it, were themes that arose consistently in the interviews.

The anxiety parents feel when they suspect or are told something may be seriously wrong with their child is acute. For most, that anxiety is made far worse when the natural

[35] The telephone can also be problematic for those with false positive results. Waisbren et al. found that mothers receiving repeat screening results after a false positive newborn screening test were considerably less stressed when they received these results in person rather than by phone or mail (2003, p. 2570).

tendency towards fear-induced theorizing and supposition is not mediated by adequate access to health-care professionals who can knowledgeably answer their questions and help them digest whatever amount of information they request and are ready to receive. As will be discussed in Chapter Five, parents' relationship to research, scientific explanations, and prognostications about CF is complex: it is not the case that more information is always better *per se*. Rather, what matters to parents is that their questions - whatever they are, however general or specific -- be answered quickly, reliably, and with compassion. Similarly, parents have complicated feelings, perceptions, and assumptions about testing itself, and how decisions about these procedures should be made.

Diagnosis with Symptoms

In my study, parents whose child got a later diagnosis were all aware that testing for CF was being done, and that the purpose of the test was to establish or to rule out CF. Parents had varying levels of familiarity with the disease at the time of the test, but none of them found that the procedure itself had been performed without their knowledge or against their will.[36] This is partly because, as mentioned above, the test takes place in the context of a health care environment where active symptoms are being addressed and a search for diagnostic information has been explicitly undertaken. It is also because in this case the patient is a child - not a fetus, not a newborn in the liminal hospital environment where professionals retain significant control, but a tangible, individuated child who is symptomatic and whose parents are seeking information about what is wrong. These

[36] It is my assumption that at a minimum verbal consent for the sweat test was given by parents with a later diagnosis, since it is standard practice to seek parental consent for any procedure proposed for a minor. However, it is a limitation of this study that I relied on parents to recount the testing process in their own words, and did not ask explicitly about consent during interviews with those whose child received a later diagnosis.

children are in the custody of their parents, and it is their parents who bring them to health-care providers to ask for assistance. Although many parents were very critical of the way their providers handled these encounters (see Chapter Five), the problems they experienced were generally with having their own knowledge and observations dismissed or belittled before the diagnosis rather than with issues of education and consent for testing.

Newborn Screening

Parents receiving a positive newborn screen result are in a very different situation. Newborn screening programs are *mandatory*. As such, they are implemented without the informed parental consent required for virtually all other health-care interventions - including prenatal testing *for some of the same conditions* screened for at birth. As political scientist Diane Paul points out, in the early 1960s when mass screening for PKU became routine across the United States, the shift away from medical paternalism and towards increased patient autonomy had not yet begun in earnest and "the question of whether to require explicit consent for testing was not on the agenda" (Paul 1999, p. 3 of 18). This, coupled with the fact that PKU testing was broadly perceived as a method of preventing mental retardation rather than as a form of genetic testing, meant that legislation giving public health authorities the right to mandate newborn screening was established with only limited public input or debate.

Since the 1970s, in the wake of highly publicized scandals involving abuse of human subjects, such as Willowbrook and Tuskegee[37], the issue of consent has been raised more

[37] The Tuskegee study of untreated syphilis in black men is "…the longest nontherapeutic experiment on human beings in medical history" (Thomas and Quinn 1991, 1498). The study was run by the United States Public Health Service, and it involved the intentional withholding of antibiotic treatment known to be effective so that the impact of the untreated disease on living people could continue to be studied. The

vigorously (Ibid.). Predictions of early critics that increasing numbers of conditions would be added to state screening panels, and that unlike PKU many of these conditions would not have early onset, or be easy to diagnose and/or treat, have indeed proven correct (Annas 1982). However, as the number of diseases screened for expands, those who have raised questions about how appropriate it is to continue mandating tests without consent have so far had little success in altering the already-established mandatory basis of the programs (Bonham etal. 2003; Mischler et al. 1998; Downie and Wildeman 2001; Paul 1998). Only three states obtain signed consent from parents to screen for mandated conditions, and even in those states it is questionable how consistently and meaningfully the legal requirement is implemented (Mandl 2002; Paul 1999; Hiller and Landenberger 1997). Other states have sought written parental consent or dissent over a limited time period for specific conditions -- including CF -- while they are "pilot tested" and researched to determine if state-wide testing is warranted (Atkinson et al. 2001; Farrell et al. 1997, 2001).

Parents do have a putative right to refuse testing on religious grounds in all but one state, and on religious and/or personal grounds in seventeen states, but these policies too have been demonstrated to have little material reality since most parents are not made aware either of the testing or of their right to refuse it (Ibid.; GAO 2003; Hiller and Landenberger 1997). Furthermore, as Downie and Wildeman point out, the option of

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blatant violation of human rights that occurred in this experiment, and the debilitating illness and untimely death suffered by the men on whom it was conducted, have been much studied in the past several decades, and Tuskegee is widely considered to have laid the groundwork for subsequent laws and ethical principles designed to protect human subjects. (For more on the Tuskegee study and its implications for research ethics, see Jones 1981; Shamoo and Khin-Maung-Gyi 2002; Reverby ed. 2000) Willowbrook was the name of a facility in New York State where children with developmental disabilities went when – usually in response to medical advice – their parents institutionalized them. In 1972, television reporter Geraldo Rivera visited Willowbrook. His subsequent expose of the inhuman living conditions and pervasive mistreatment of the inmates eventually led to the institution's closing. Like "Tuskegee," "Willowbrook" has since become shorthand for a signal event in the development of human subject protections.

refusal must be exercised in the "total institution" hospital setting where parents are likely to experience vulnerability because of the physical stress of the infant's birth, as well as imbalances between themselves and health-care providers with respect to both information and power. "All these factors considered, parents may feel that raising an objection is in essence disturbing the status quo, and so may not feel comfortable expressing a refusal. Those who wish to refuse may instead maintain a frustrated (or confused) silence." (2001, p. 21 of 39).

The right to informed consent for newborn screening is thus virtually non-existent in the United States. In lieu of an established legal entitlement, official recommendations and state policies have focused on provisions for notifying parents about the screening in advance, and for providing them with educational materials (Downie and Wildeman 2001). However, even parent education is not a requirement everywhere. Thirty-eight states now require that parents be notified before the mandatory sample is collected, but ten states continue to collect it without explicitly telling the parents. All but two states reportedly make some educational materials available in a more general fashion (Hiller and Landenberger 1997; Mandl et al. 2002), although what those materials are and how they are "made available" varies tremendously within states as well as between them. At this time, there is no evidence that either prenatal or hospital care routinely includes meaningful health education practices around newborn screening, and studies have consistently shown that parents whose children were screened did not feel adequately informed or have a good understanding of the process (see for example Waisbren et al. 2003).

Furthermore, "education" as it is conceptualized in official guidance on this issue to the states is primarily a mechanism for increasing parents' compliance with newborn screening program procedures, and for lessening their anxiety with respect to the lengthy and flawed testing process. Take for example the American Academy of Pediatrics' (AAP) statements on "shared decision-making" in its recent high-profile report on newborn screening (AAP 2000). AAP shied away in that report from a strong stance in favor of informed consent, but firmly recommended that substantial parental education be incorporated in all programs.[38] "A greater emphasis on parental education may improve parent understanding and *increase the number of parents who comply with recommendations for further testing and follow-up*," write the report's authors (emphasis added). And they continue, "Such education may also help parents deal with the anxiety associated with equivocal results, repeated tests, and false-positive results." (AAP 2000, 410) The 2004 CDC report recommending newborn screening for CF indicates a similar view about the purpose of education: "If providers were to educate parents in a relaxed manner before labor and delivery, a positive screening result might be met with greater willingness to obtain follow-up and with less worry about the meaning of a positive test." (CDC 2004, 26) The states, in turn, convey directly to health-care providers the message that parents must be informed so they can comply. The "Education" section of a 2003 New York State Department of Health's Newborn Screening "Guide for Health Professionals," for example, begins with the observation that "Public Health Law 2500a

[38] Notably, the report also specified that such education should take place during the third trimester of prenatal care, when the mother is most likely to be capable of attending to and absorbing information. The same information should then be re-imparted after birth (AAP 2000, 410-11).

mandates that parents be informed of the purpose and need for newborn screening." It continues:

> Informed parents are better prepared to follow-up on presumptive positive test results and to facilitate timely evaluation, diagnosis and treatment of affected infants…. Prior knowledge of the newborn screening program by parents and discussion of test results with the infant's health-care provider further assure that all infants receive the benefits of this most important public health program. Prior knowledge of the screening also reduces the stress that may be associated with requests for a repeat test (2003).

Similarly, the state of Maryland tells its doctors that

> It is misleading to refer to the newborn screen as 'a PKU test,' because tests for other diseases are included. Parent confusion about the full scope of testing may impact compliance with follow-up should the newborn screen be abnormal" (Maryland Department of Mental Health and Hygiene, 2004).

Parents in my study knew little or nothing about the newborn screen for CF before it occurred. To a person they describe themselves as poorly informed by their health-care providers, and confused about what the testing meant. Francesca, for example, says she was aware that some blood work was being done on her baby, and that she may have even signed a form, but she had no idea what would happen with the blood once it was taken. "I didn't think to even ask what they were screening for," she says.

> I figured it was like diabetes, cancer, you know, [or] just what [blood] type she was…. I didn't know what cystic fibrosis was even, so I had no clue. And then even when the doctor called me and told me to come into the office, and after he had told me what was going on, I went home and told everybody she had CP [cerebral palsy], because when he called it CF I never heard of it.… The only thing I remember, somehow it sticks out in my head they were going to test for what blood type she was. I thought [this] was probably normal, in case something came up that had to do [with] any kind of transfusions or whatever. But nobody ever said to me, this is the things we're testing for specifically, this is what we're looking for.

Other parents too were given no clear sense of what the test was for, and thus were left with just a vague impression that "they were testing for a type of retardation" or for diabetes. As Joan put it, "I just thought they tested their hearing, their reflexes and that stuff. I didn't know that it got into genetics and I had no idea that we would come out of it with a diagnosis for something."

Only one of the parents I spoke with described a meaningful verbal exchange with her health-care provider before the blood was taken, a conversation in which the doctor explained

> … that the newborn screening was to make sure that there were no genetic diseases that were not obvious. And they do that as a precaution just in case, because some of the diseases are pretty bad and hard to diagnose without this [test], just with symptoms. So they try to catch it at birth so they can start treatment right away.

For the rest, those who described any formal notification in advance of the screening at all remembered only receiving a pamphlet or booklet about the program. However, this health information was just one piece of paper among many to them at the time, one more thing in a pile of pamphlets and paraphernalia. Paige describes how irrelevant and unimportant the material seemed when it was given to her like this:

> When we were in the hospital, somebody had given me a booklet saying 'we test for these metabolic diseases.' And I just kind-of glanced through it and set it aside with the other bazillion things that you get when you have a new baby, you know……the diaper bag and there's formula samples and the wipes…. So, it just kind-of got stuffed in the diaper bag and put off to the side. I really didn't pay much attention to it, um, until I got the phone call, and then I was digging for anything I could find.

Francesca had a very similar experience:

> There's so much going on… you had a baby and everything else. Or if they gave [information] to me before I had her heaven knows I wasn't even listening, if I'm in labor, what ever, 'just give me some drugs! Where do I sign? What do you need me to sign, because I'm in pain, come on.' Yeah, so I didn't know what they tested for and still to this day I don't, I mean I know now that they test for CF, [but] I don't know what else they even [test for on the panel].

Anthony too finds it understandably hard to remember precisely what information he and his wife may have been given before the screening. However, he gives his health-care providers the benefit of the doubt, assuming that they did their duty to inform him and even to ask for him to sign in consent.

> Yeah part of me says [details about the screening process] could have been offered. I just didn't pay that close attention because I didn't, maybe its just me, [but I thought] that's all the other people, it never happens in my children. Perhaps they did give us all that information and it was among 80 other pieces of paper they you know handed [us] before [and] during the birth process…I don't remember the exact time and you know when we signed [a form], but I knew that newborn screening existed and basically the breakdown of it…. I just sort of went with the program.

This assumption that parental consent was officially sought -- albeit without any kind of accompanying information that would facilitate understanding or meaningful choice rather than simple legal acquiescence with "the program"[39] -- was explicitly noted by half of the parents who got a newborn screen diagnosis (four out of eight). Their memories of the process are of course foggy: it occurred long ago; they had no reason to take particular note of it at the time; the exigencies of the birth process and the new baby eclipsed most other aspects of the hospital experience. Nonetheless, they presume that it was their legal right to give or withhold consent, and that an effort was made on the part of the hospital to comply with the procedural if not the substantive aspects of this right.

[39] For an excellent discussion of the difference between "consent" and "informed consent," and of what constitutes meaningful information in the context of seeking consent, see Annas 2004, Chapter VI.

As Lilly put it, struggling to remember what happened in the hospital at the time her daughter's heel was pricked, "I'm sure I had to sign some consent forms." But her conviction, and those of the others, is more that the process couldn't have happened without her signing than that the signing actually occurred. Further, the shared assumption is that any form parents were given with respect to newborn screening was a consent form rather than some other sort of paperwork -- such as the pink copy of the laboratory screening form parents are routinely given in the hospital in New York State.

Only the hospital's medical record would show precisely what forms were signed at the time of the screening. However, none of the families I spoke to reside in any of the three states that require written parental consent as a condition for newborn screening, and only two live in states where consent was possibly sought for CF testing on a pilot basis at the time of her/his child's diagnosis.[40] It therefore seems probable that parents' assumptions about their legal rights vis-à-vis newborn screening do not emerge entirely from their own experience. Rather, they may be influenced at least in part by the now broadly-shared public view that "people have a right to decide whether or not to have their bodies invaded" in medical settings (Annas 2004, 113), and that parents make all such decisions on behalf of their children. The fact that newborn screening is an extraordinary exception to the doctrine of informed consent is esoteric knowledge from the point of view of women going into the hospital to have a baby: debate on the issue is not highly publicized, and most people would have no reason to give it a second thought.

Once the positive screen has been reported, though, details of how the system works are no longer irrelevant or little-considered aspects of medicalized birth. A routine

[40] In one of these states at that time, research has shown that three quarters of parents did not know that CF was included in the screening program, and only thirty-seven percent realized they could refuse to participate in the pilot program (Tluczek et al. 1992).

hospital procedure turns out to be a life-changing event, and parents are left "digging for anything they can find" among the papers and forms they may have carted home. Alternatively, they are on the phone to family and friends trying to get more information, or on the internet attempting to sort through the mass of data and commentary available there – including the outdated information that causes such distress for many parents (see above).

Thinking back on the experience, two mothers said that despite the difficulties they experienced with not having any information in advance about the newborn screening process for CF, they were not sure they would have wanted much more knowledge. Details about the conditions being screened for would have given them "too much to worry about" between when the heel prick was done and when the results came back, and would have caused excessive "aggravation and worry." "I'm kind of torn," one of these mothers said, but

> I know what it is to be a first-time Mom. With my daughter if they'd have explained to me these are the 10 or 15 diseases that we check for and if you get a positive hit on any of them, this is what we're gonna do, I think that would have just made me worry more. So I guess, I guess kind of no [I wouldn't have wanted more information ahead of time]. I think I would have gotten myself into a real state by the time they called me.

Other parents, however, fervently wish they had known more from the beginning about the test and what might happen. "I would have wanted to know exactly what they were testing for and why and what exactly it was," says one mother.

> Like, where do we go from here if it is positive? I mean it just seemed like such a whirlwind at the time. They were like "we think she has CF, we're not going to tell you what it is, you have to go here and do this." And those people were like "well we did the test, and it looks like it's positive but we're not sure. You have

> to go here and do this." And we just felt that we were running in circles, just not knowing.

Another mother, Francesca, describes her desire to have had precisely the kind of parental education recommended by the AAP's 2000 report.

> I guess if they could give you more information during your last trimester about the newborn screening in your state, and 'here's what we test for, do you have any questions, and do you want any more information on it?' that would be so much [better]. Maybe it's just because I've had an experience now with the screening where something came back positive, maybe I'd be different if I never did…. but just to me now looking back that would have been a time where at least I was thinking about it…. I would have [heard] the word before. You know even like, 'CF, that's right, they told me about that they were gonna test for it in the newborn screen.' And I maybe wouldn't have been so panicky about it earlier…, maybe I wouldn't have been such a basket case over a word I'd never heard before.

This mother is the same one who described in more detail, in Chapter Two, the nature of her panic after the positive screen: her fear that the child was in immediate pain; her guilt that perhaps CF was her fault; her anxiety that the baby might die right away; her regret that she had gone to the doctor alone to get the result, not realizing it would be bad news (see pp 58-9, 105-6). Like most parents in my study, she is modest about her own knowledge, and doubtful of her authority to speak on behalf of anyone but herself. Yet her experience of panic, of being a "basket case" because she had no idea what CF was, seems to me a critical piece of the story about the impact of mandatory screening without consistent parental education. It is not the only kind of experience parents have with the screen, but in my study it is not uncharacteristic either. Rather, it's the most dramatic example of a more generally shared sense that the health care system inadequately prepared parents to cope with a positive screen. To date, this human cost of current screening policy has been overlooked. We know from research in Maryland, where

informed consent *and thus reliable parental education* has in fact been required for more than twenty years, that voluntary screening can be effective: parents in that state knew more about genetic issues post-screening than parents from whom consent was not sought, and fifty-four percent of those surveyed stated that they prefer that their consent be asked (Holtzman et al. 1983; Faden et al. 1982; Annas 1982). Furthermore, contrary to the widely-shared assumption that logistical obstacles make meaningful informed consent impossible, Faden et al. found that "a parental consent requirement is a feasible institutional requirement" (1982, p. 1348). Yet informed consent has neither become standard practice in the U.S., nor has research ever been conducted to determine what the consequences are to parents of *not* being informed before the test.[41] This unnecessary suffering has been left out of the calculus altogether, its toll neither calculated nor reckoned with. The data from my interviews suggest it's time an accounting was made.

At the same time, the reticence of some parents to know more about newborn screening in advance may signal a form of self-protection from the endless warnings and calculations about risk that now mark everyday life. As a number of social scientists have shown, medical and public health practices focus more and more on analyzing the inner workings of the asymptomatic body so as to identify and circumvent disease before it is manifest (Lupton 2001, 1995; Petersen and Lupton 1996; Nelkin 1996; Nelkin and Tancredi 1989). "All bodies are constructed as 'at risk' from one or more conditions or illnesses… [since] all people, whether or not they are experiencing symptoms, may harbour 'risk factors' potentially leading to illness" (Petersen and Lupton 1996, 48).

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[41] Notably, during my interviews parents focused almost exclusively on the pros and cons of being *informed*, and very little on the issue of whether they had a legal right to *consent* or not. Because this became clear to me only after most of the interviews were completed, I was unable to examine this "finding" in greater depth. However, I do examine the nuances of the informed consent issue in more detail from the policy perspective in Chapter Six.

Perhaps for some parents, imagining a meaningful discussion before the test is done about newborn screening and all it entails means bringing this discourse of danger into the newborn period in an overly prominent and explicit form. Perhaps they have had enough of this risk assessment and testing from the prenatal setting where -- as I discuss below -- it is already an inescapable standard of care. When providers "explain about ten or fifteen diseases" and that the baby must be tested for each, they actively label the baby as at risk, and "to be labeled as being 'at risk' means entering a state in which an apparently healthy body moves into a sphere of danger" (Ibid.).

It is perfectly understandable that some parents would like to avoid knowing about this particular "sphere of danger" ahead of time, and thus to avoid adding worry about the tests to the list of anxieties flooding the newborn period. It is also entirely reasonable that for others, it is critical to know everything that is happening to their child, and to avoid being blind-sided by a devastating test result from a clear blue sky. But if the choice is between medical paternalism on the one hand, and increased submission to the discourse of risk prevention and danger on the other, it seems to me – as I discuss in Chapter Six -- it's time to redefine the terms of the debate.

Prenatal Testing

Pregnant women undergoing carrier screening for CF during pregnancy have experiences with education and consent that are distinct from both of the other groups just discussed. Although there are a number of similarities between prenatal and newborn screening (as suggested above) with respect to the experience of "early" diagnosis for an asymptomatic child, there are also substantial differences. From a legal standpoint, at minimum, education and consent constitute one such area of difference.

Women cannot, in theory, be subjected to medical tests or treatments without their informed consent: they must be allowed to agree to or refuse procedures at will. In this sense, they may be protected -- at least to some degree -- from the dangers of an experience like Francesca's, where testing is done without parental knowledge and in the absence of any meaningful information about what the result might mean. At the same time, for pregnant women the experience of a pregnancy free of these harrowing decisions no longer exists (Katz Rothman 1986, 1998; Rapp 2000; Lippman 1991). Prenatal care even for the most healthy and "normal" of pregnancies is marked by frequent recommendations for diagnostic testing. Although women can and do refuse these tests --- and some even find their way to midwives and a less technology-dependent model of care -- social norms, health care practices, and technological imperatives have militated in favor of increased testing for at least two decades (Ibid.; Shakespeare 1995, 1998). As Katz Rothman put it in her seminal book The Tentative Pregnancy, "more and more we believe one should have all the information, one *should* be prepared, act rationally, not bury your head in the sand. The pressure to get information comes from all sides" (p. 84). In this context, informed consent is not just a legal requirement. It's also a social norm that takes as fact that "action is based on information, and the fullest possible information is needed to determine action responsibly." Further, conscious decision-making based on that information -- i.e., "taking charge of your life" – is seen as the only mode of mature, responsible behavior (Ibid., 83)

Prenatal testing, as Katz Rothman predicted, has in fact become entirely routine, and parents now receive increasingly extensive information about the status of the fetus as a matter of course. However, due in large part to reluctance on the part of policy-makers to

make mandatory a set of genetic tests which most often result in pregnancy termination, women's right to "choose" when it comes to prenatal genetic testing has been preserved. The tests are certainly offered to every pregnant woman receiving prenatal care: guidelines from both the American Academy of Pediatrics (AAP) and the American College of Obstetricians and Gynecologists (ACOG) actually emphasize that physicians have an *obligation* to inform them about the "existence and uses" of various prenatal tests. But at the same time, the genetic counseling that health care professionals are supposed to provide is meant to give the woman medical facts and risks, inform her of her testing options, and allow her to reach her own decision about which procedures - if any -- are appropriate for her (Annas 2004).

CF carrier testing has been available selectively for a number of years, but only recently has it begun to move into the mainstream of prenatal testing in the U.S.. In September of 2001, ACOG and the American College of Medical Genetics (ACMG) issued guidelines recommending CF carrier screening for all couples in which at least one partner is white, once prenatal care is being sought or a pregnancy is being planned (Zoler 2003; McNamara 2004; www.acog.org). Although implementation of this recommendation remains only partial to date and the test is still not available to the majority of pregnant women, screening numbers have increased rapidly over the last few years. Best estimates for 2003 are that roughly 20-25% of obstetricians were offering the test, and that approximately 300,000 - 500,000 women were screened in 2003, up from just a few thousand in 2000 (Zoler 2003; Harmon 2004; Medical Director for March of Dimes, 2005).

Research to date on the impact of carrier screening in the U.S. has tended to focus on actuarial calculations of the expenses and savings associated with the program, and on its relationship to reproductive decision-making. In a 2003 report on Kaiser Permanente's prenatal cystic fibrosis screening program, for example, the director of that program estimated the costs of screening and the costs of caring for children with the disease and noted that the "prevented cases" – i.e., the terminated pregnancies of women whose fetuses were predicted to be affected by CF – resulted in savings roughly equal to the cost of the screening program (Bates 2003).[42] Other studies examine the psychological and social consequences of CF carrier screening in the general population (Watson and Mayall 1992; Williamson et al. 1989); how well information about CF is retained in relationship to how it is taught (Clayton et al. 1995); and how much interest non-pregnant couples have in screening tests (Clayton et al. 1996). In contrast, to my knowledge relatively little research has been done regarding pregnant women's actual *experience* of carrier screening in the U.S., as carrier screening rolls out in more and more prenatal-care settings. It remains unclear, for example, both what percentage of women refuse the carrier screening in actuality, and what percentage were explicitly aware that the screening was being done before the test was run. Marteau and Dormandy observe that with respect to prenatal testing in general, we are "unaware of how women are counseled, the information and support they receive, and how this affects the quality and type of decisions they make (2001, 189). Testing for CF is no exception.

[42] That two of the eleven aborted fetuses were "predicted to be mildly affected *or unaffected*" (italics added) by cystic fibrosis does not stop the author of the article from referring to all 11 as "prevented cases" (Bates 2003).

In my study, I was able to find only two women who had gotten a fetal diagnosis and then carried the baby[43] to term.[44] My difficulty in recruiting such participants may be due in part to the limitations of my study's scope and design. However, parents I interviewed throughout my research repeatedly noted that they did not know anyone who had decided to proceed with the pregnancy when a fetus was diagnosed with CF. Several recounted stories of being contacted by pregnant women after they got a fetal diagnosis, and of sharing their own experiences raising a child with CF with these women as they were deciding whether to continue the pregnancy or not. Despite the feeling on the part of the mothers living with CF that they had been encouraging, had talked about how wonderful their child or children are and how the disease can be managed, the pregnant mothers who called – none of whom yet had a child with CF -- all chose abortion.[45] Quantitative data also suggest that a significant proportion of the still-restricted number of women who get a prenatal CF diagnosis choose to terminate their pregnancies. According to one study, 95% of those actually receiving the diagnosis terminated, and only 5% carried to term. (Witt, as quoted in Harmon 2004). Studies of projected *intent* to abort, however,

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[43] I use the term "fetus" when talking about a time when the pregnant woman might have considered abortion an option, and the term "baby" when that option has been clearly ruled out.

[44] As noted in Chapter One, I also interviewed a mother who sought and obtained prenatal diagnosis via amniocentesis for a second child, the younger sibling of an affected child.

[45] The distress over the choice of others to abort affected fetuses that was indicated by several mothers I spoke to, combined with the reluctance of all the women in my study to consider terminating a subsequent pregnancy of their own, may reflect the effect on reproductive decision-making of already having a child with a disability/diagnosis. Collins et al. note that: "A study of the attitudes of women to prenatal diagnosis in France showed that those who had no acquaintance or personal relationship with a handicapped child were more willing to undergo amniocentesis (Julian-Reynier et al. 1993). The personal relationship between parents and the child makes the issue of prenatal diagnosis more difficult." One mother in their own study did not want genetic counseling in connection with a subsequent pregnancy because "'That would be like saying we shouldn't have had (*her [older] child with DS*) in the first place or he's not perfect enough.'" (Collins et al. 2001, 65, 69). Although my own data regarding this issue were neither substantial nor systematically collected, they do seem to corroborate the findings of others that having a child with a disability/disease changes the way women feel about subsequent prenatal testing and abortion of affected fetuses.

put the numbers much lower -- at 36% in one case (Watson and Mayall 1992); 29% in another (Botkin and Alegmagno 1992, as cited in Wilfond and Fost 1992, 641); and as low as 20% in a third (Wertz et al. 1992).

The two mothers with a prenatal diagnosis whom I did interview both reported being the first woman in their prenatal care and hospital settings to have a baby with a known CF diagnosis. One of these mothers described how her obstetricians repeatedly asked her, at each visit over a period of six weeks or more, if she had considered her option to terminate even though she "didn't have any interest in having that discussion." They made the assumption, she goes on, that "because I did the amnio, of course I would terminate if it's positive, and that actually wasn't the case. We thought information would be very helpful in learning about the disease should, should the outcome be positive." This use of test procedures is not in fact uncommon: Read, for example, found:

> Parents use prenatal diagnosis for something other than as a basis for decisions about abortion: such reasons might include a desire to prepare for the birth of a child with special needs, to ensure that the newborn gets proper care immediately after birth, to conform to the perceived wishes of their doctor, or to have information but defer a decision about a medically positive result until if and when it must be confronted" (2002, 272).

However, many health-care providers continue to presume that abortion will follow a positive test result.

Neither one of the mothers I interviewed recalled knowing about or giving consent for the test. However, like the parents receiving a newborn screen diagnosis, they don't doubt that information about it may have been given to them. Kate describes what happened to her like this:

> At my hospital,… when they know you're pregnant they want to see you when you're eight weeks pregnant. So I went in at eight weeks pregnant, and they took blood, and at that time apparently they told me they were gonna test me for CF carrier status, I don't exactly remember that conversation but I'm not doubting that happened. Because, and I explained this to the doctors too, because at that time you're so excited, 'I'm pregnant,' and they are giving you so much information at once, very little is actually getting in except 'I'm pregnant!'

Crystal's experience was similar.

> I didn't even know I was having it done. With [my first born] twins all they offered me was this, um, test for abnormalities like Down Syndrome…and I decided not to do it because I was 30, I think, at the time. And then he offered it again to me this pregnancy. I was 35, so I decided to do it. I didn't realize other things that they were testing for even, so and then he calls me and said I was a carrier for cystic fibrosis, I didn't even know what that was… They just said it was the new recommendation in our state…is to be, for Caucasians to be screened or tested when they're pregnant…

The lack of warning about the initial test, combined as it naturally is with very little information about what CF is and what it might mean for the baby and family, was nearly as stressful for these mothers as it was for those getting the unexpected newborn screen result - a similar carousel of hope and fear. The time between the first positive carrier screen and the ultimate diagnosis was between six and eight weeks long - even more prolonged than the waiting period for confirmation of positive newborn screen results. During that period, as I discussed above with respect to newborn screening, lack of reliable information caused significant stress and anxiety, and access to good sources was felt as a godsend.

Kate's experience was that her obstetricians were just plain uninformed, and her genetic counselor -- whose specific role it is to provide information and counseling to pregnant women undergoing testing -- was radically misinformed. For one thing, the

genetic counselor told Kate the doctor would give her anesthesia during the amnio procedure. When she got there, however, the doctor told her he had stopped this practice long ago and went ahead without it. More importantly, the genetic counselor told Kate and her husband that their baby's particular CF mutation was unequivocal bad news.

> This is where I could have killed the genetic [counselor], because our question was, 'it's our understanding… that this disease really varies and the same mutations could show up very differently?' And she's like 'oh no no, you have the most severe, your child is really in big trouble.' And we were like 'oh my God.' But [later we learned] that's not true, because there's so many factors that play into this. Yes she does have the most common, historically it is the most severe, yes. But they cannot project what is going to happen to her…

When Kate tried to seek out information on her own in the face of this poor performance by her health-care providers, things didn't get much better.

> There is so much outdated and misinformation out in the world about cystic fibrosis…. If you say to someone, 'what do you know about cystic fibrosis?' they'll say 'I knew so and so who died when they were four, or there was a neighbor who died when they were twelve.' And that was true, I mean it was definitely the case and it, while it still happens today, there is so much that has been done in the last fifteen years, I mean they didn't even find the gene until 1989…the world is different now in CF. And so it wasn't until we actually talked to the CF doctors, which was the day after we heard the official diagnosis -- when we talked to two different CF doctors, they were like full of hope…. They said, 'yes, this sucks, but it's not an abnormal life… these kids are amazing.'

Like her counterparts waiting for follow-up tests after newborn screening, Kate certainly experienced the ill effects of outdated information. But in her situation, other factors were also at play. The presumption that a woman receiving fetal diagnosis of an abnormality will not carry the baby to term means that those who do continue the pregnancy are subject both to incredulity (or worse) on the part of health-care providers, and to written and verbal information skewed towards a negative view of CF and its

implications. As Lippman and Wilfond have documented, the stories about genetic disorders told to pregnant women differ substantially from those told to parents after the birth of an affected baby.

> The *before*-birth information is largely negative, focusing on technical matters and describing the array of potential medical complications and physical limitations that may occur in children with the condition, while *after*-birth information tends to be more positive, focusing on compensating aspects of the conditions, highlighting the availability of medical and social resources, and stressing hope for the future… In the *before* stories… the dominant message appears oriented to avoiding the birth of a child with… cystic fibrosis. In the *after* stories, the message is oriented to caring for a child with one of these conditions (1992, 936).

In Crystal's case, the genetic counselor her doctors referred her to at the beginning of the diagnostic process turned out to be an important resource. "I liked her when I went in the first time," she says. "We talked for at least an hour, and she was very helpful. I told her… how much detail I wanted… and I felt comfortable talking to her." It was invaluable for Crystal to have a relationship with someone knowledgeable and sympathetic, who also was willing to follow cues with respect to how much information to provide and how to provide it.[46]

Certainly there are many issues with genetic counseling – including the way it is implemented (as illustrated by Kate's experience), and the way it is conceived as a "nondirective" intervention ostensibly designed to facilitate individual decision-making, but inevitably "directing the woman's attention toward some questions, and away from others" (Katz Rothman 1986, 46). As Patterson and Satz put it, the very act of offering genetic counseling about disabilities and defects of fetuses in the health care setting,

[46] See Chapter Five for more in-depth discussion of how important it is for parents to obtain just the right amount of information for them at the right time, and how critical it is for health-care providers to understand and respond to this need.

where abortion is a clear option, communicates to patients an underlying assumption that it's a bad thing to give birth to a child with a genetic condition. "The implied goal of medicine is to eliminate diseases, defects, and abnormalities. Based on this medical model, the implied goal of genetic counseling is to provide information that reduces the likelihood of propagating these conditions" (2002, 123, 130). And this goal is not always just implied: the literature on genetic counseling often refers to its "… importance in preventing the birth of more affected children in the same family (Dankert-Roelse et al. 1987, 271).[47] Nonetheless, the fairly widespread availability of this resource to women during prenatal testing means there is an at least potentially helpful professional whose job it is to answer the woman's questions; to provide up-to-date information complete with however many details the woman requests; and to mediate the testing process. For most women getting a *newborn* screen result, in contrast, no such resource exists until after the diagnosis has been confirmed, if at all.

Neither Crystal nor Kate sought CF carrier screening initially, but both felt compelled to complete the next carrier screen, and then the amniocentesis, once they were told of their carrier status. As Crystal put it, "[I] decided I wanted to find out if the baby had cystic fibrosis because otherwise I'd spend the rest of my pregnancy worrying about it…so, I just wanted to know…." Kate had an even stronger reaction. " I felt like this [first carrier screening result] was thrown at me. [But] I felt like once the can of worms was opened, I couldn't hide it…. But I did not and would not have sought it out…. No, definitely I-I would not have looked for it at all."

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[47] For a more general discussion of the "new eugenics," disability, and prenatal testing, see Shakespeare 1995, 1998; Katz Rothman 1998; Hershey 1994; Kerr and Shakespeare 2002. For a more specific examination of the attribution of control and blame following the birth of a child with a disability or disease, see Marteau and Drake 1995; Land 1998; Berube 1996.

There can be no clearer example of "the pressure to get more information" that "comes from all sides." But still, as suggested above, the amount of decision-making power vested in women depending on *when* genetic screening happens remains an important difference between prenatal and newborn screening. Mothers of infants with a positive newborn screen can hardly be said to have an option with respect to subsequent testing. Faced with the information that their child might or might not have a serious, even a life-threatening disease, and with instructions from the doctor to show up for confirmatory testing, not many parents are likely to "opt out" of discovering whether the diagnosis is a false positive or a genetic reality. Furthermore, public health systems in most states (forty-seven of fifty-one) are designed to "track" abnormally screened infants at least until a diagnosis is made (Mandl et al. 2002), and in at least some states child-protective services will be alerted and will “visit” parents if they fail to comply with recommended follow-up (State Newborn Screening Director, 2005).

Pregnant women and their partners, on the other hand, can realistically choose whether to proceed with carrier screening for the father, and then with amniocentesis if that screen is positive. Yes, as the women in my study illustrate, this decision is heavily influenced by the imperative to find out whatever can be known, even if the first step in the testing process was taken unwittingly. But some women still do refuse further testing. One study found that 33% of women said they would not pursue a prenatal diagnosis after positive carrier screening (Botkin and Alegmagno 1992, as cited in Wilfond and Fost 1992, 641). Another reported that of women who already had a child with CF, only 26% (8 out of 31 women) consented to prenatal diagnosis when pregnant with their next child (Mischler et al. 1998). As anthropologist Rayna Rapp put it in her

discussion of why prenatal diagnosis is sometimes refused, "although amniocentesis has rapidly diffused and become part of routine prenatal care… some choose not to accept its complicated benefits and burdens…. [A] routinizing technology does not always stay *en route*" (1998, 50).

In my own study, one of the three women who got carrier testing during pregnancy decided not to proceed with fetal diagnosis, opting instead for immediate testing after birth. For her, amniocentesis was just too risky. Perhaps even more important, she didn't see any purpose in knowing earlier. "There's nothing that can be done for you," she said. "[And] especially if it's a first child or you don't have one with it…. you don't know what to expect and fear of the unknown is often worse than reality." Unlike Kate and Crystal, who wanted to know everything there was to find out once they were aware their babies were at risk, Suzanne preferred to preserve the joy of the pregnancy unspoiled, and cope with whatever was to come after birth.

The Vexed Nature of Choice

The data presented above suggest that choice with respect to diagnostic information is a complex and vexed terrain. Choice can mean the specific power to consent to or refuse a procedure. This kind of choice is available to parents in prenatal settings, and if their child has a later diagnosis, but it is virtually out of reach for parents with respect to newborn screening. Even for those who do have legal power to exercise decision-making autonomy, social context exerts a huge influence: as "'choices' become available, they all too rapidly become compulsions to 'choose' the socially-endorsed alternative" (Hubbard as quoted in Katz Rothman 1986, 12.) But choices can also mean the freedom

– less directly exercised, but intentional and meaningful in the minds of parents nonetheless – *not* to be forced to make an explicit decision, *not* to take on the moral burden of deciding what to know and when, or even of worrying over an ever-increasing number of perils before it is determined that any personal, concrete threat is real. This kind of freedom no longer exists in the prenatal setting. With newborn screening, on the other hand, the mandatory nature of the test means both that parents don't enjoy the privilege of informed consent, and that the weight of making an explicit judgment about the test doesn't fall on them.

How the parents in my study first learned about their child's CF was determined largely by circumstance, and very little by active choice. It was not an effort of will that brought some to an early diagnosis by newborn screen, and others to a later one after symptoms had emerged. Even for those with a fetal diagnosis of a first child, it was the new policy of "offering" universal CF carrier screening in prenatal settings that set off the chain of events leading to the diagnosis rather than an explicit search on the mothers' part for this information. However, with the significant exception of those parents who endured a true "diagnostic odyssey" throughout which the symptoms of CF were repeatedly overlooked or dismissed by professionals (see below, and Chapter Five), parents felt grateful that they found out about CF when they did. Like other researchers describing parental responses to chronic illness or disability, I found that the parents I interviewed normalized their experience in a variety of ways (Hill 1994, 160). Instead of focusing on how they lacked control or felt stigmatized by the diagnostic process, parents spoke about the advantages of receiving news of CF at whatever time it came.

Those who got a prenatal diagnosis – though they did not actively seek the testing -- were glad they knew about CF before the baby was born and therefore had time both to mourn and to prepare. For Crystal, the birth of her son did nothing to alleviate "this feeling of… dread that I had [that] I thought once he was born [would] feel better." Nonetheless, she says,

> I would rather have found out the way I did than find out later. Because it takes me a long time to process things and I would rather be prepared. And I was, you know. Before he was born, I already [had gone to]… a cystic fibrosis clinic, I visited with them and talked to a pediatrician to see if they were gonna be good for him… and find out what we might need to prepare for. I'd rather have done that, I'm a planner and I like to plan… And I mean I still have bad days where I'm upset about all of this, but I went through a lot of that when I was pregnant, and I'd rather have done that than just coming home with a newborn, and having to go through, you know, you're happy and then all of a sudden everything's not happy…or having him… home with us for a while and not knowing what's wrong with him. And I feel good knowing that we could do preventive things to keep him from getting sicker.

Kate too feels it was critical to have had the chance to respond to and become accustomed to the diagnosis before the baby arrived, and that her suffering was therefore less than it would have been if she had gotten the diagnosis later.

> I heard such awful horror stories of people that were misdiagnosed or had to go through all kinds of testing for months and years, to figure out what was wrong with their child, and [I'm so grateful] to not have to go through that, and to go into her birth really armed with information… like at day 3 we started her on enzymes and at day 5 we started her chest therapy, and I was quote unquote completely prepared for it. I didn't know what it was going to feel like to do it, but I knew what to expect and I felt more than anything else, and I think this is a huge deal, I did all my mourning. Because I do think this diagnosis has to be mourned. I did my mourning when I was pregnant and so by the time she came, I'm just happy she's here and I'm just loving my baby and I'm just having a great ole time. A lot of the people who [get the diagnosis after birth]… I think they didn't have the time to mourn that diagnosis. [They are] so busy taking care of their child, they still haven't had time to truly mourn it.

Parents who got a diagnosis at birth are also grateful to have found out when they did. They did not know prenatally, and they're glad –with perhaps one exception – that they did not since they imagine knowledge of the disease would have destroyed their enjoyment of the pregnancy without any compensatory benefits. Lilly notes that fetal diagnosis would have just been "more worrying and what if," and Francesca says "I just think it would have taken away from my pregnancy with her… I was so happy pregnant… I wouldn't want that to [be] taken away…." Not having been faced with an explicit decision about whether to continue the pregnancy, and with the ensuing negotiation with the baby's father over this issue, was also a relief for a number of mothers. "Do I wish that I would have had a prenatal diagnosis?" asks Paige.

> Absolutely not. I think if I would have gotten a prenatal diagnosis, although I didn't believe in abortion, I think I would have been incredibly overwhelmed. I think my husband would have wanted an abortion. I think it probably would have ended up an abortion, and something I would have regretted for the rest of my life, so I'm glad I did not have a prenatal diagnosis.

Like those who found out about CF soon after birth, most with a later diagnosis (nine out of eleven)[48] are relieved not to have been faced with a prenatal diagnosis, for all the same reasons. As Nancy put it,

> I'm definitely glad I didn't have the prenatal. I had a great pregnancy. I was extremely happy, you know, and just excited, and I think if I would have had the diagnosis prenatally it just would have been horrible for me…. I mean we'd rather've had the blissful pregnancy and then find out you know that something's wrong because we would have continued with the pregnancy no matter what.

[48] One of the two mothers with a later diagnosis who said she did wish she had found out about CF while pregnant was under the impression – erroneous as far as I can tell, though understandable given how fast technology is evolving and how complex these issues are – that if she had been tested prenatally "they can actually start treatment in utero… I could [have] start[ed] treatment before he was born."

And from Anthony, "unless there was treatment that could occur beforehand, no [we would not want prenatal diagnosis]…. [Since] there's no course of treatment that could happen in utero, then I don't think that I would need to know beforehand in any way."

As I discussed in Chapter Two, parents who got a later diagnosis have complex, sometimes contradictory feelings about what was gained and what was lost. Those whose child's health was not compromised by a period of "blissful ignorance" treasure the time they had without CF. These are the parents who feel fortunate about their personal experience, though they believe in the abstract that the earliest possible identification of the disorder leads to the greatest control over its course.

Parents whose children were symptomatic for a prolonged period without a diagnosis, and who suffered temporary and/or permanent physical harm as a result, are the only subgroup in my study who don't significantly normalize their experience. Of course they don't: the consequences for their children were too grave. As I discuss in more depth in Chapter Five, their children's well-being was at stake, and the health care system failed them. Kim says,

> Definitely I wish I had known at birth. She'd be stronger, healthy, bigger. We wouldn't have had to suffer during all the prolapses. When they'd happened we could have looked it up to see if it's a sign and known instead of months of agony wondering what this was, all the different tests she had to see why she was having it. The doctor, she says no it wouldn't be CF, she's been pretty healthy with her lungs. She did not recommend a sweat test. If we had known at birth definitely she would have been healthier.

And from Roberta,

> If we would have gotten [the diagnosis] at birth, I think it would have been a lot easier to know right away… to be treating it right away…. I think [that] would have been preferable as far as just not having this huge stretch in time where he's very sick, and we're convinced that the human race wouldn't survive….

These parents wish with all their hearts that CF had been recognized earlier so that they could have been spared the stress of not knowing what was wrong, and so that their children could have gotten prompt treatment for their symptoms as well as overall preventive care. Some blame late diagnosis on uneducated clinicians, and advocate broader education about CF.

> More information needs to be said about the symptoms of cystic fibrosis, not that's it's just a sticky mucus that affects the lungs, you hear that everywhere. It needs to be said there are different symptoms of cystic fibrosis. You know because so many children could be so much healthier if they knew okay your lungs could be fine but you could be malnourished because you have cystic fibrosis. You know things like that. You could have these sinus problems because you have CF. Okay. Let's take care of it. You know so many kids are undiagnosed.

Most parents, however, see newborn screening as the primary – or perhaps unique – solution to the problem of the "diagnostic odyssey." Whether they experienced the trauma directly, or just heard about it from others, all agree that this confusing, damaging experience is to be avoided at all costs. As Kim says flat out, "I think that kids should be checked for it at birth, and if that could be put into every state that would be so helpful." Andrea agrees that the policy "would save parents a lot of heartache," save them from going through wondering what's wrong and having "doctors look at you and go 'well there's nothing wrong, you're just imagining it.'" And from Joan,

> Well I definitely think that newborn screening should be mandatory, should be the way to go, because so many parents have to have their kids sick and in and out of the hospital and they've been tested for everything else. And then one doctor will think 'hmm we haven't tested for CF,' and then these parents have just been through the wringer when they finally found out what's wrong.

By asking questions about their own diagnostic experiences and whether they would have wanted things another way, I encouraged the parents I interviewed to think more explicitly about what happened to them, and about their preferences and decisions, than they might otherwise be likely to do. This is a challenging exercise. In interview after interview, I was moved by the adaptability and optimism that parents showed in coping with their child's diagnosis, however it arrived. It is possible that good luck gave those with prenatal diagnoses, those with diagnoses at birth, and those with later diagnoses and no "odyssey" the diagnostic process that really was best suited to them before the fact. But this seems highly unlikely. Much more plausible, given the minimal amount of choice parents in my study actually exercised around diagnostic information, is the hypothesis that parents find useful ways to make sense of the experience that was dealt them. They look for the bright side, and – unless something positively damaging occurred – they focus on what Frank describes in his typology of illness narratives as the "belief that something is to be gained" (Frank 1995, 115) through the experiences they have had.

When we enter the realm of hypothetical behavior, of asking parents to suppose what they might do or have done in a situation they have not yet faced, the terrain becomes trickier. Would more parents have in fact opted for CF carrier screening if it had been offered to them? Would more have sought out immediate testing after birth if it was a truly voluntary rather than a mandatory program? If it were possible, would parents prefer to wait until their child shows that first sign of illness before getting a (prompt and efficient!) diagnosis?

Other researchers have documented the common discrepancy that exists between hypothetical and actual "uptake" of predictive tests. For example, about three quarters of surveyed families at known risk for Huntington's Disease indicated they would want to be tested for the condition despite the fact that no effective treatment exists (Kessler et al. 1987). However, only about 10% or fewer actually come forward to be tested, and of these, a significant number may be actually using the test to confirm their own suspicion that they are beginning to have symptoms or that they are free of the gene because they are past the age of usual first onset (Richards 1993). Macintyre summarizes the issue this way: "… asking a global hypothetical question about whether people want screening may be neither helpful nor predictive of what people actually do, and… what people actually do is extremely context-dependent" (1995, 227).

Complicated predictions of behavior at the population level are not appropriate for the size and scope of my study, and making them is not what I have set out to do. However, my interviews made apparent that attitudes and decisions about diagnostic testing for CF are complex, and that what parents think they may want in theory and what they do when faced with actual situations are not always the same. The decision-making process is complicated by a number of things. How explicit is the "choice" of testing (i.e., does it need to be sought out, or is it just done as a matter of course)? How many alternative routes to testing are available? How problematic are the alternatives to any given test? How significant are the "opportunity costs" of testing at any given point?

Explicit support by parents for newborn screening is enthusiastic. Al-Jader et al. (1990) found 83% of parents of screened infants were in support of the policy, as were 91% of parents of unscreened babies. My data suggest similar verbal endorsement of a

universal testing policy: almost all parents in my study noted they believe newborn screening is a good thing, although some did so with specific reservations (see Chapter Six). My data also suggest, however, that parents' underlying attitudes towards such tests are complex and ambivalent. Some wish they had had more time before the diagnosis, as I have already shown. Others say they think newborn screening is a good and important thing, yet they themselves choose – when choice is an option – to wait a while before pursuing testing for a subsequent child, one born after the diagnosis of CF in an older sibling.

Six mothers in my study went on to have another child after CF was a known genetic risk in the family. None of them had any intention to abort. Three got fetal diagnoses (one positive, two negative). One opted for carrier testing for a new partner (not the same man who fathered the older child with CF). When the carrier screen failed to identify any of the more common CF mutations, the couple did not proceed with prenatal diagnosis. Rather, they found out the new baby's CF status by waiting for return of the newborn screening test that is mandatory in their state. The remaining two mothers did not do prenatal testing, *nor did they test the infant immediately at birth.* Both were anxious, very anxious, about whether the child was affected. But they waited to do the actual test until they knew the child for a number of months (four in one case, and as many as eight in the other). For both, by the time the test was done, they had a clear sense based on observation and experience of what the result would be.

Meredith and her partner refused prenatal testing for baby Jim, whose older brother Steve was diagnosed with CF at birth because he had MI, reasoning that it would not "have made any difference on whether we continued through the pregnancy or not."

Once Jim was born, though, Meredith watched him nervously, looking for any first indication of a problem or symptom.

> I don't know how many times [my boyfriend] said 'Meredith, will you just relax, he's fine.' [But] its just that whole not knowing. I have to say that did just totally encompass me, the not knowing, the wondering 'ok, so could this be a sign of that, or could this be a sign of that?'

For Meredith, those early months were full of stress, of "watching every move he made and then wondering." Yet she held off on getting him tested until he was 8 months old or so.[49] "I think it was fear of possibly getting the diagnosis was why we didn't," she says. "With Jim, it was just more of a wanting to know but being afraid to, I think is the best way to put it." By the time the test was actually done, it served mostly to confirm her sense that he did not have CF, yet it was still nerve-wracking.

> It was more, I really don't think he does, but let's be positive. Let's not wait till he is two years old and begins having symptoms and then say 'oh yeah, he had CF too'…. Its just that reassurance, that proof…. So we were pretty positive that it was gonna be negative but still that fear was there of 'ok, what happens if he's just a very moderate [case], you know, very very mild?'

Erica had originally thought she might test her third child, born after diagnosis of the older two, at birth, but her doctor suggested delaying a bit. Even though she didn't feel like this physician "was someone who had my best interest at heart" she decided she "wanted to wait." Rather than labeling the baby right away – something she is wary about given issues with labeling she had encountered with an older child in school -- she decided to just watch for and treat the symptoms, to "treat how he presents." As it turned out, he was symptomatic for CF very soon. Like Meredith, Erica used the test to confirm

[49] Meredith could not recall exactly how old Jim was when the CF test was done, but thinks he was 8 months old or younger.

what she already knew, what she had learned little by little from parenting her child. And she was very glad that she got the confirmatory news later rather than right after birth.

Many people embrace diagnostic technology and use it to gather information, to make decisions, to prepare psychologically for what will be coming next. But some people choose to find things out in other ways. They learn by observation and experience, and want to protect themselves from the blunt reality of a test. A black-and-white indicator of "yes" or "no" may be too harsh for some. Perhaps if newborn testing was mandated, these parents would feel grateful that the decision had been made for them. But as a matter of volition, immediate testing even for those children *known* to be at risk for CF may not be the norm.

Chapter Four

The Ghost in the Room: Parenting in the Shadow of Cystic Fibrosis

As we have seen, getting the CF diagnosis is unquestionably a shock and a sorrow, a critical moment in the lives of parents. They remember it vividly even if many years have passed since its occurrence. They relive it often, recalling every detail of who was kind and what was hurtful as it unfolded. In addition to the significance of this event in and of itself, however, getting the diagnosis is important because it is the moment that ends the parents' pre-diagnosis existence; it marks the beginning of their careers as parents of children with CF. Once on the post-diagnosis side of the divide, parents can never go back. This chapter explores what the parents I interviewed had to say about going forward: the impact a CF diagnosis has had on how they connect with and view their children; and how their parenting practices and styles unfold in the context of CF.

Living with the Ghost of CF

The CF diagnosis is unquestionably in the forefront of mothers' lives when it is new. Most are utterly preoccupied, at first, with understanding and responding to the news. But over time, as with any seminal event, there is a process of acclimatization; CF begins to take its place within the family context, no longer occupying every nook and cranny of available space. However, it is never forgotten, either, even in times when the child is healthy or when other major life events – the birth of other children, divorce, career changes – are in process. As one mother with an at-birth diagnosis puts it, CF is "constantly a part of our life." Another describes it as "… just a burden, there's just a

weight… there's just a little shadow lurking around… it's almost like there, there's a ghost in the room you need to live with…and that ghost is cystic fibrosis…."

"That ghost" transforms, for many parents, their fundamental perspectives on their child, and on what the future will hold. The context in which their parenting unfolds suddenly feels like a bounded one, one that can no longer hold grand dreams or the lavish hopes that come with a sense that you are caring for the next generation, the one that will outlive you. Suzanne, whose son was also diagnosed at birth, describes the change this way:

> I think beforehand you sort of think about, like you have certain dreams and expectations of what life with your child is going to be like and… that they are gonna live a normal life and be happy and healthy and everything else. But afterwards it's all different. Like you sort of think about well, no, it's not always gonna be healthy and maybe they're not going to live for as long as you think that they would've before knowing about it and I mean… I think it just changes your perspective on things….

Paige's dreams for her baby were also radically altered when she received the newborn screen diagnosis, her taken-for-granted optimism also suddenly shattered.

> I was going to do everything to make, to make sure this child would grow up to be a, a great individual, who, you know, with a great childhood… that thrived so well. I was going to do everything in my power to make sure everything went well for her, and all of her dreams were coming true. When I found out she had cystic fibrosis…, a lot of my dreams for her, I felt like they very well could die, because CF might take her before she can attain these dreams.

This changed perspective alters, in turn, the way parents bond with their child, and the way they organize, practice and understand their own care-taking.

This Isn't the Baby I Thought I Had: Bonding After the Diagnosis

In my study, parents who received an early diagnosis described the impact of CF on the process of bonding with their baby as more significant than did those who got a later

diagnosis. This finding is consistent with what Al-Jader et al. (1990) discovered. These researchers conducted highly structured interviews with twenty-nine parents of babies diagnosed with CF either through newborn screening or after the development of symptoms. Of the eleven families they identified as "most severely affected" with respect to bonding, nine had received a diagnosis from screening rather than clinically (464).[50] One of the mothers in that study described rejecting her child during the diagnostic period and beyond, stating that learning her baby had CF "'destroyed the bond and natural feelings that I had at first and I had to start building a new relationship all over again'" (Ibid., 462).

For parents in my study, profound gratitude for information that may result in better health and development for the child often co-existed with a sense of loss and sadness about how the process of connecting with her was irrevocably altered by an unexpected diagnosis during the first few weeks or months of parenthood. Joan, for example, like the mother quoted above, experienced the newborn screening diagnosis as a clear interruption of the bonding process she had begun with her baby.

> [I]t seemed like it was 6 months to a year before I felt, okay, I know what I am doing now and I really feel close to this baby… I think I had to mourn the loss of the healthy child I had. You know, I had to go through that mourning process of, okay, I don't have a healthy child. This isn't the baby I thought I had.

Francesca also found the first year of her daughter's life heavily dominated by CF; for her, too, it was only when Tess turned one that she began to regain the confidence and ease she had felt that first month of Tess' life, before the call from the pediatrician with

[50] The authors attribute this difference largely to a longer delay between mention of CF and confirmation of the diagnosis for parents in the group that received newborn screening. However, they fail to offer any evidence demonstrating why they conclude the relationship between these two variables is causal.

the positive screen. “It took me from when [the positive newborn screen was confirmed by follow-up tests at] two and a half months [until] her first year birthday where I actually felt like ‘okay, we’re not dealing with CF, the CF is gonna have to live with us.’” These are radical transformations Francesca experienced – from not suspecting anything at all could be wrong with her apparently healthy baby, to having Tess’ identity dominated by an unknown and invisible disorder, to coming out the other side with a synthesis of the two, a clear sense at last that the lives of the family and the child are primary and the diagnosis secondary rather than the other way around.

Betty’s daughter was not only diagnosed right away, but was also symptomatic. Like Shannon (see Chapter Two) whose healthy baby had been diagnosed by newborn screening, Betty found the diagnosis made her self-protective, afraid to love a daughter who might die young.

> It was kind of like, how can I love this baby? Because I tried so hard at the very beginning not to love her very much, because I didn’t know if I was going to lose her. And it was really easy to say this just, this is the baby and she’s got problems… so let’s just keep our distance. I mean that was really easy to do. And um so after that meeting [with the doctor when we got the diagnosis], to go and just say here’s my baby and she has CF. It was, it was just hard.

Reflecting further on what it was like to connect with Rose in those early days, Betty adds “we had to get to know her as somebody with CF, not just to get to know her for her and then figure, oh, she’s got CF, you know?” Like Francesca, she feels keenly the effect of never having gotten to really know her child before the diagnosis.

Jennifer Rosner’s observation that the “excitement of having a new baby was entirely eclipsed” by the early diagnosis (2004, 20) was thus keenly echoed by the mothers in my study. They too felt grief over every experience their child might not have, over the

struggles the child was already encountering or might encounter as she got older, over the loss of the "baby they thought they had." Some of them were unavoidably confronted with the immediate diagnosis because the child manifested illness, but others lost the ability to get to know their baby first as a baby *not because she was sick, but because of the early test.*

Only one parent who received a *later* diagnosis spoke explicitly about the significance of bonding issues in the post-diagnosis period. This is the mother who discovered her daughter had CF around her first birthday, at the close of a relatively healthy year with no identifiable symptoms except in retrospect. Although the child in this family was older than those diagnosed at or immediately after birth, the transition from a "well" to a "sick" identity was similarly abrupt since the diagnosis followed immediately on the heels of the first really recognizable sign of a problem. Here is what Nancy says about her response.

> At first my initial reaction, which is a horrible feeling to have as a parent… while we were even still in the hospital, was that I didn't want to be close to her. I mean I didn't want to bond anymore with her. Because I was afraid of losing her. You know I just had this like guard put up and it lasted for probably a week or so and I hated it, I knew that this is wrong. This isn't how I should be feeling but I couldn't remove the feeling and then I started to feel like I needed to spoil her.

It is customary for research looking at the impact of early diagnosis on bonding to discount shorter-range effects, seeking instead to determine whether there are long-term outcomes in terms of behavior or attitude. The underlying question is often this: "Were difficulties with attachment temporary and ultimately resolved, or were they sustained and significant enough to cause lasting harm to the child or to the parent/child relationship?" The narratives recounted to me by the women I interviewed highlight a different aspect of the issue – i.e., the residual sorrow, guilt, and regret that comes to

mothers as a result of having *ever* kept at a remove from children to whom they are now so passionately committed.

Each and every mother I spoke to, no matter how hard that initial period was, found a way to get through it, a way to profoundly connect with her child. Some of the most moving moments in these emotion-filled interviews came when mothers described this process. Listen again, for example, to Betty, who began right away the work of bridging the distance she felt from Rose after the diagnosis. Imagine you can hear the tears in her voice as she speaks.

> I went up there [to her room]… with my camera and I sat there and I held her, and I dressed her up and we took pictures and I just talked to her for a while and it was, it was, it helped. It helped a lot just to tell her that you know I'm her mom, and its ok and we're gonna take care of her and, um we're gonna have a lot of fun, and not to worry about whatever's gonna happen cause we'll take care of it. And um I just kinda sat there and explained it all to her, and it kinda put a perspective [on] how I felt about her, cause obviously I cared.

Of course she cared. Each parent I spoke with cares, and each also tends her child with tenderness, competency, and abundant love. But this does not mean that the bonding issues raised by an early diagnosis weren't both devastating in the shortrun, and also resonant over a much longer period of time. In fact, it's precisely the contrast between what they felt at first and what they came to feel later that remains an open wound for these mothers.

Nicole communicated the pain of this contradiction in all its fullness. She told me in detail, weeping, about how right after birth, with the pending diagnosis and a host of medical issues to attend to, the work of parenting was numbing, devoid of the emotional connection she grew to feel once the child got older and she came to really know her. " I

think in the beginning it was such a, it was a job," she recalls. "I loved her but I was so busy caring for her that it was almost kind of routine almost at the beginning. And [then] she grew and she became stronger and um I guess you just have to know her but she is so happy and loving." Having at first experienced caring for baby Jenna as numbing and technical rather than joyful and heartfelt causes enormous regret and guilt for Nicole: how could she ever have failed to love this baby as she deserves to be loved, seen her as co-extensive with her diagnosis rather than as the full human being she now knows her to be? None of the mothers I spoke to could recount their version of this without similar emotions, even if many years had elapsed since initial estrangement melted into absolute devotion.

More than a decade ago, Ellen Wright Clayton observed that "[n]o one has determined… whether the benefits to the child of avoiding parental uncertainty exceed the impact on the child's bonding with the parents when serious disease is diagnosed in the newborn period" (1992a, 642). My study is not sufficient in size or scope to answer that question, but my data do suggest that it remains an important one to examine if we are to grapple fully with the implications of newborn screening.

Whatever's Gonna Happen, We'll Take Care of It: Adaptive Parenting Practices

A majority of parents I interviewed, regardless of when they got the diagnosis, described CF as having a substantial impact on their parenting practices. For some, the influence was greater in the months or years right after the diagnosis, and for others it was more permanent. The range of adaptations parents made in their parenting was fairly broad, but clear themes emerged over and over.

Building a Bubble: Vigilance in the Post-Diagnosis Period

One major change many parents experience after CF diagnosis is increased protectiveness towards the child. As they learn about and/or experience the pulmonary and nutritional vulnerability CF can produce, parents – particularly parents of younger children -- find themselves trying to control their child's exposure to germs of every kind. This can take the form of becoming a "hand-washing Nazi" in the home, of sprinkling sanitizer over every surface and feverishly scrubbing every toy and appliance. As Kim describes it,

> [we] got rid of our bird feeder, got rid of our little pool in the back yard, you know. Constantly dumping water on her swing set in the back yard every time it rained in case it's got pseudomonas. Scrubbing the house constantly, disinfecting this, disinfecting that, until we were driving us both crazy.

Paige too found herself "very protective" after the newborn-screen diagnosis.

> I was like the mother bear. I mean, don't come near her without washing your hands, don't take her to the mall during the dead of winter, you know protecting her in every possible way. I just became so intent and almost worried and obsessive about some things, pouring bleach down my drains every other day because pseudomonas could live in my drains, cleaning like crazy, almost to the point where I was driving myself nuts… I mean, since then I have calmed down a

> lot, and I've learned a lot more about cystic fibrosis, but it was just very intense [in the beginning].

Vigilance in the post-diagnosis period can also result in social isolation, as parents follow medical advice to minimize exposure to germs by keeping their affected child away from other people, especially other children. "[You become] very protective, very over-protective," says Meredith, wanting to guard your child "…from anything…be it the insane world we live in, or the bugs that some kid next to him sneezes and wipes his snot on his arm." And from Kim again, "You [are] afraid to go to the store. Afraid to go to church. Afraid to shake hands…. You can't take her to pools because of the chlorine levels, what if they're not high enough to kill the germs?" Some parents do create a sort of bubble, keeping their children out of child care settings and many public places, especially in winter; keeping them away even from family gatherings if others attending might have an infection of any kind. There is satisfaction in guarding the baby's health so thoroughly, and relief from the anxiety of watching over her in public can be huge. But the costs for the mother herself can also be significant. A parent interviewed by Taner-Leff and Walizer summarizes succinctly what mothers in my study described about this experience:

> Laura's pediatrician wanted her kept away from everyone with their wintertime germs – 'don't take her shopping with you, don't take her near other kids,' – and of course both the season and the presence of a new baby made it harder for me to get out anyhow. I had been used to working closely with adults and teenagers every day – literally having a couple thousand interactions – and suddenly I was locked up in the house with a… newborn and a cat. The telephone helped some, but my physical isolation echoed the isolating effect of Laura's birth defects. (1992, 139).

Many parents in my study were well aware that there is a danger in becoming *too* protective, both in terms of the child's healthy all-around development, and in terms of their own need to "have some sort of social life." However, arriving at that balance between minimizing the risk of infection and getting out into the world a bit is not always easy. As Andrea puts it:

> I was kind of torn. I wanted to make him really healthy, wanted to put him in a bubble.... People used to joke that they would almost have to go in the bathroom and shower before [touching] the baby…. We cleaned out the house, we scrubbed it top to bottom and just set everything up. No carpeting in his bedroom cause I wanted to make sure, I mean I knew that there were respiratory things so I wanted to make sure that there was no chance of asthma-related things… I wanted to wrap in him in a bubble but I still was logical enough to know that that would not keep him healthy, what I needed to do was make him active…. The whole thing is keeping his lungs strong… keeping him active, to let his lungs grow strong and his heart grow strong.

Nancy and Paul, too, strive to find that middle ground with their daughter Alexandra. "We've gone through cycles," says Nancy, "where we feel like just keeping her home, because it is easier, safer…. I do definitely worry about public places… But then we realized you know she wants to get out. I mean she's really active, really social, she doesn't wanna be stuck in the house."

Parents whose children were older at the time of diagnosis are generally more sanguine when it comes to issues of vigilance. They protect their children, and a number of them mentioned specifically and with regret how connecting in person to other families with CF is impossible because of the high risk of cross-infection between kids. At the same time, they see a substantial difference between their own parenting practices regarding infection control and the practices of those diagnosed early. As Erica puts it:

> Well one thing I have noticed [is that parents with early diagnoses]… become more like lunatics. A lot of these people they will share different stories. They are putting like plastic on the shopping carts. I guess they are so young when they are diagnosed that they are so concerned about bacteria and stuff. They become more over-protective, I guess that would be a good word. And I guess I'm more relaxed with things. I mean some people are, "I can't come into contact with other people." One girl, I wonder what happened to her. She had two kids with CF, and her sister had it. She wouldn't let her sister see her kids.

The only parent I interviewed who seems to have entirely avoided a tendency towards increased vigilance is Lorraine, the mother whose older child was a completely healthy two-and-a-half-year-old when both he and his little sister were diagnosed. '[I]f I had known when he was a brand newborn I might have been a little bit more alarmist [and] concern[ed] than I was knowing he had made it to two-and-a-half without any real misses and had CF," she says. "I honestly think [if I had known earlier] I would have been way more protective than I ended up being." However, with the diagnosis coming later as it did, she is emphatic that CF didn't change how protective she was, or really much at all of her feeling about who Luke is.

> My son had done so well…. I was just determined that he was going to be fine and I really didn't become over-protective. I mean at that point we were in, we had a nanny and she was watching my son and another boy and they went to the park, and he went to preschool and you know little art classes and gym classes and that kind of thing.

This experience is strikingly different from those of the parents in my study with *early diagnoses* and no or only mild symptoms. Crystal, whose third child, Martin, was diagnosed prenatally but had had no symptoms to date, describes herself as feeling exceedingly protective towards him, notably more so than with her first children, a set of twins with whom she was relatively relaxed and comfortable. With Martin, though, she

found herself sanitizing everything and not "tak[ing] him out as much as I would if he… didn't have cystic fibrosis." The fact that his CF mutation has been identified as a mild one, and that his health-care providers have said he is unlikely to have any symptoms for quite a number of years, if ever, does not prevent Crystal from guarding his health zealously, nor does it relieve her of a vigilant watch over his development and body functions. "I think that if we had not had this testing," she says, "I don't think he would be diagnosed right now…. He doesn't have symptoms, I mean he's a baby, he's gaining weight…." Nonetheless,

> there's things about him that I just pay much more attention to… I mean all this stuff about poop, it's kind of funny -- what do babies do? They eat and poop. And the twins… they were just healthy, whatever they did, they did. But with him, I'm like calling, you know, every time I visit a doctor… it's like talking about how often he poops and et cetera, and I'm concerned about it…. [And] he's really gassy, and… a lot of times, I'm like I don't know if his gassy problems are just from being a baby or [from] cystic fibrosis… I'm worried. I just want, I just want him to be normal.

Francesca describes how her experience of parenting was changed by the early diagnosis – transformed from an assumption that all was well to a preoccupation with what might be wrong -- this way:

> When I just had her those first four weeks [before the diagnosis], I would have never have thought she was different than anybody else. But then I don't know if it made me more paranoid after the sweat chloride test but you know then you would hear her breathing heavier or like her chest sounded like… more kind-of gunky and just you know like it didn't sound clear, it always sounded like it had a rattle to it, and I don't know if some of that is just paranoia that set in or if it was truly that she you know had these symptoms and just being a new mom, that Tess was my first one, I just didn't know different.

Judy is a mother of three. She had long worried about her oldest child, who suffered a variety of health problems from birth. At age six, this child was finally tested, and CF was added to her other already long-established diagnoses. The other two children were then also tested: the middle child was negative, and the youngest positive. Before the diagnosis, says Judy, "I worried about [the two youngest] the same," though both were relatively healthy. But afterwards, she became a "little more paranoid" about the youngest, wondering "okay, what does this mean, is it gonna be turning into something more" every time there was a little sneeze or cough, every time anything potentially signifying the formerly-unsuspected CF came up.

Kate's baby, like Crystal's, was diagnosed prenatally. At the time of our interview, Kate's baby was four months old and, like Crystal's son, still symptom-free. But Kate too describes hyper-vigilance as a prominent aspect of her parenting.

> I analyze everything. It drives my husband nuts. When I come home and the babysitter's there, 'When did she poop, what time, what did it look like?'… Cause I feel like I have to stay on top of it, like if we need to make an adjustment for her enzymes, I want to do it immediately…

Kate has not yet found anything truly worrisome to report. Neither has Shannon, whose child was two-and-a-half and completely a-symptomatic at the time of our interview. Shannon doesn't feel she treats Margo differently than her older sister, in particular, but at the same time, she does describe her own version of the monitoring, the vigilance, and the uncertainty voiced by others who had an early diagnosis without symptoms. "There's never been anything to show, or anything scary or tarry or mucousy looking or foul nasty smelling," she says. "I mean they said there's so many ways on that

end that you would notice something was up, but you know**,** it's hard, cause I've always been like, am I missing something?"

"Am I missing something? Is there a symptom here I might fail or have failed to catch early enough? Is what I'm seeing normal, or is it a sign of CF?" These questions are most common for women with early diagnoses and relatively healthy babies. Children who get a later diagnosis are already known: symptomatic or not, their mothers have developed some sense of their children's bodies, their patterns, how they work. Although some mothers of children with a later diagnosis certainly also become more protective after they know CF is to blame, for most of them the change is markedly less dramatic. They may be vigilant about germs and contagion for a time, their hearts may skip a beat when they hear their child cough or see her clutch her stomach, but anxiety over the unending hunt for not-yet-visible symptoms is one heartache they're spared.

Boland and Thompson, in a study looking at effects of newborn screening for cystic fibrosis on maternal behavior, found that mothers of screened children who were asymptomatic scored significantly higher than mothers of screened symptomatic children on an "intrusiveness" scale. They also found that mothers of asymptomatic children scored higher on a "fostering dependency" scale (1990,1242). Similarly, Southern, in a 2004 analysis of the implications of newborn screening for CF, speculates that parents of asymptomatic children identified through newborn screening may find their situations "stressful," since "in some ways it is more difficult to be living with the anticipation of future deterioration in condition" (2004, 59). I don't know if such anticipation is *more* difficult than coping with manifest symptoms; I'm not sure that framing of the hypothesis as a comparison is even useful or appropriate. But as a matter of empirical verification, I

can say it is clearly true that parents caring for diagnosed children wait for that first symptom nervously, look for it vigilantly, and sometimes doubt their own competency to detect it early enough. And even while they wait for it, they also, as I will show below, try with all their might to avert it.

Is My Sick Child Healthy?: Measuring Parental Success by the Child's Health

Protectiveness and vigilance are, of course, means to an end. They are tools parents use to keep their child healthy, to keep CF at bay. Parents work both to prevent illness and suffering in the moment, and to forestall long-term damage to the lungs and other organ systems. Monitoring stools, listening for subtle changes in breathing, communicating regularly with health-care providers, administering aggressive proactive treatment at the first sign of a cough or sneeze – these things, and more, rapidly become a critical part of what it is to parent a child with CF. For most of the parents I spoke with (with the exception of one or two in the group whose children were asymptomatic), doing chest percussions and/or administering digestive enzymes also become routine aspects of parenting immediately after the diagnosis.

Although learning about CF always feels daunting at first, generally those whose children were diagnosed later not only experience relief at finally knowing what is wrong, but also take great satisfaction in learning and embracing whatever interventions are recommended. As already noted in Chapter Two, after the helplessness of tending a sick child with no diagnosis, there is enormous comfort in having something concrete, at last, to *do*. Erica talks about the introduction of CF-related home treatments and preventive therapy for her children, ages six and four at diagnosis, like this:

> With my [six-year-old] daughter, I always knew something was wrong. And I think sometimes… finding out something, it gives you… not peace, but at least you have a name and you know how to treat it. As opposed to non-stop coughing, that worry, that uncertainty or worrying. At least… you feel like you're doing something, you're treating them, you're doing something.

Many of these parents – particularly those whose children were quite ill at diagnosis -- see their new regime of treatment and infection control rewarded by marked improvement in their children's health. Administering enzymes to children who have pancreatic insufficiency, and have become malnourished as a result, helps them begin to gain much-needed weight at last. Treating pulmonary symptoms with a combination of medication and chest percussions often brings them under control: lung function and thus overall health begin to improve. Some children who had been in and out of doctors' offices and hospitals before the diagnosis stabilize, and these visits become a rarity rather than a routine. "She was terribly malnourished and underweight without us realizing it," says one mother. But after she began getting treatment, "she became this totally different little girl."

While parents' gratitude for being able to "do something," and for improved health and quality of life, is enormous, it can also be bittersweet These parents had already established routines and caretaking practices with their children before the diagnosis, had already developed their parenting habits and styles. Just as a CF diagnosis creates a clear "before" and "after" in the parent's perception of her child and in her dreams for the future (as seen above), so also it creates a clear "before" and "after" in her career as caretaker, and the adjustment can be substantial. Here is Erica's description of the transition:

> One impact of [the diagnosis] was, like with the kids, everything is before and after. I mean, I really truly view things as before they were diagnosed and after they were diagnosed. *In terms of?* Everything. How our social lives changed…. `cause CF varies, treatment and medicine. It's all involved. At times we'd have up to 3-4 hours a day doing treatment. So before that we never had that. So it's a lot different. Like I would say I was more carefree. I always remember that it wasn't until about two, three years ago was the first time we had gone out at night, since the kids were diagnosed. But just for a walk round the block or something because their treatments… take from 7:00 to 8:30, then they go to bed…. When [kids] are born with it they don't know any better and I think for me as a parent you fight it more when you find out later because you knew what normal life was…. You deal with it, but I think later on you fight it. You just remember how things were. That sort of before and after thing. I mean like every picture in the photo album I can look at it and see whether it was before or after…. Your concerns change and it's a very isolating feeling because there is nobody who can really relate to you.

Nancy too described the enormous divide marked by the diagnosis. For her, too, the shift was both very emotional and very practical.

> [O]ur whole focus, our whole perspective, everything has changed since we had the diagnosis… [W]e don't say 'this won't happen' or 'that's not gonna happen.' We don't have that kind of thinking anymore because we just, we don't feel so secure like we used to, or so immortal I guess. Yeah, I definitely think there's a division… It [also] take[s] a lot of time, just [feeding her]. I mean normally you'd just hand them the sandwich. And now we're like, 'okay… we need to get the enzymes. How many enzymes does she need for this meal? How much is she gonna eat? Is it a high-fat meal, or a regular meal?... Does she need her vitamins at this time or does she not?'…. And just cleaning the nebulizer cups and sterilizing those and you know everything is just, it's very time-consuming. When we were on the month without Tobi[51] its like 'ahhh [we're so relaxed],' and then the month of Tobi is 'urgghh. You do the Tobi and I'll clean, I'll boil the neb cups and hang them out to dry and then I'll get them ready for the morning treatment….' So it's definitely very time consuming, it's extremely life-changing just in all those ways, everything that I took for granted.

All the parents whose children were diagnosed later worked diligently to keep them as healthy as possible. For all of them, their new parenting practices in the service of health are a prominent aspect of the "after" side of the diagnostic divide. And most of them

[51] A specific medication for CF.

recounted to me, with pride, their successes: stories of children able to participate in sports and other regular childhood activities; stories of children who have been hospitalized only minimally or not at all since diagnosis; stories of children who keep up with or surpass their unaffected siblings or friends.

These narratives are powerful; parents' investment in improving and maintaining their children's health cannot be overestimated. But this aspect of their parenting takes place within a broader context. For some parents, this context includes the "normal life" that came before, with its as-yet-unaltered parenting styles and priorities and its as-yet-unaltered expectation that the child would live an average life span.[52] For others, it may also include the very abnormal health problems their child suffered before the diagnosis, and the feeling that things have now begun to get better. Parents feel enormous anger at the delayed diagnosis (see Chapter Five), and sometimes guilt as well. But the physical and developmental signs they monitor with their diagnosis-inspired vigilance most often mark an improvement rather than a deterioration in health, at least in the short run. They aim to influence the course of the disease as much as is humanly possible, but they have generally already learned, through the difficult experience of seeing their child show symptoms, that they cannot mitigate its effects entirely. Furthermore, they have some degree of confidence that they'll recognize their enemy CF when it manifests: the disease is after all an old foe already, even if it only recently got its name.

Attitudes about and experience with controlling the child's health are quite different for parents who got an early diagnosis, and whose parenting has therefore always focused – at least for the affected child or children -- on maintaining good health despite CF. For

[52] For a couple of parents in my study, it should be noted, fear about longevity preceded the diagnosis because the child's health had deteriorated so very far before CF was identified as the problem.

these parents, vigilance, protectiveness, and preventive treatments *are* the norm that substantially defines what it means to parent that child -- without any sort of "before" to act as outside referent.[53]

The normalization of their focus on maintaining good health was described by parents of children with early diagnoses in a variety of ways. Some talked in straightforward terms about routines of care. For example Andrea, whose son Bobby was diagnosed via newborn screen, describes (with a chuckle) how very taken-for-granted these parenting practices become after a time. "I really don't even remember how to feed a normal baby anymore," she says. "I was watching my nephew when he was a newborn and caught myself getting ready to give him enzymes…. It's automatic; you see it's a baby, you give 'em enzymes!" For Andrea, there is no parenting of Bobby without these practices.

> Now, 6 years later there's no remembering a time without out it. There's no remembering a time before the treatments, and before the chest percussions and before the enzymes and the antibiotic rounds every few months. It's just normal to us now and we try to make it as normal as possible for Bobby cause then he doesn't feel like he's different.

Other parents spoke about how the early diagnosis transformed an amorphous parental intention to "do everything in my power to make sure everything is good" for the child into a much more specific commitment to keep her physically well. Lilly states that after the newborn screen diagnosis, it became her explicit goal to "make sure [Mia] stayed healthy." She measures her success in this most critical of parenting arenas carefully.

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[53] It would be useful to explore whether and how parents with an at-birth diagnosis experienced the period of pregnancy as some sort of "before" with respect to parenting, and if their perceptions on this issue differ from those of women with a fetal diagnosis. However, I don't feel my sample size or interview guide led me to any substantial insight on this question. Therefore, for the purposes of this discussion, I am combining prenatal and at-birth diagnoses, with the caveat that it is likely there are meaningful distinctions between the two that should be taken up in future research.

"She never has been hospitalized, she's never had intravenous antibiotics, or anything." Furthermore, Mia rides horses, plays the saxophone, and is extremely physically active. In fact, "she's healthier, my sick child is healthier than some healthy kids."

For Anthony, the major change after baby Denise's newborn screen diagnosis at age two months centered on keeping after every potential sign or symptom. It became essential to communicate with health-care providers frequently about every suspected problem, just to be sure.

> I have no doubt that… we do probably treat her differently, or will sort of treat her differently, just knowing that she does have a genetic disorder and that her health and you know all that stuff may be compromised… if we don't give her [prompt medical attention]. I guess our biggest concern is that she goes to the doctor often….

Kate also describes her success as a parent in terms of keeping her child – diagnosed in utero -- healthy. "I feel I'm succeeding if she's doing well," she says. "If she doesn't get sick, I'm doing the right thing as a parent. If I can tell you every time you call me, 'She's doing great! The enzymes are working!'… then I'm doing the right thing." Clearly, though, there's a problematic flip side of this strategy for defining parental success: the heavy weight of guilt that descends when something goes wrong, even something as banal as the onset of a mild cold. When a primary intention and course of action as a parent is to protect the child's health, there's an awful lot riding on every fluctuation in physical well-being. As Kate says,

> When she caught the cold, I felt awful…I should have kept her away from people, `cause I, I took her to my sister's baby shower… and then she got sick… And so I shouldn't have brought her to the baby shower. Well, of course I should have brought her to the baby shower, you know what I mean…. But I felt so guilty and awful, you know, like that I was failing her and that I was failing as a parent.

One Step Ahead of Trouble: CF and the Illusion of Control

Kate's situation illustrates a critical issue for all the parents I interviewed: the possibility and limitation of control in the face of CF. Nothing can be done to alter the genetic make-up of the child. Despite continued research in the troubled field of gene therapy, the chromosomal mutations themselves are a reality parents must accept for the foreseeable future. The clinical expression of CF, though, is immensely variable, as noted in Chapter One. It remains unclear – both at the population level, and of course in each individual case -- how much of this variability is attributable to the specific nature and combination of the CF mutations a given person has, and how much can be influenced by active medical, behavioral and "environmental" interventions (Moskowitz et al. 2005; WHO 2000). But since such interventions offer the only hope for parents, their only opportunity to exert any control whatsoever over a disease they know is capable of ravaging and cutting short their child's life, it is not surprising that they channel so much energy towards seeking out and implementing them.

It is also unsurprising that parents struggling with the meaning of their child's diagnosis – particularly when that diagnosis is early and/or the child is not symptomatic -- are extraordinarily susceptible to the public health establishment's strong emphasis on the advantages of early diagnosis. When refracted through the lens of newborn-screening programs, these messages represent the disorders being screened for as imminent dangers to the child, and the knowledge gained through testing as a means of minimizing that danger. The New York State Newborn Screening Program pamphlets, for example, tell parents that the screen is done to "help ensure that your baby will be as healthy as possible," and that "early treatment is very important" since if left undetected

and untreated, these diseases can be life-threatening, or may "slow down the baby's physical development or cause mental retardation." The pamphlet notes that "none of these disorders can be cured," but goes on to emphasize repeatedly that "the serious effects can be lessened – and often be prevented completely – if a special diet or other medical treatment is started early" (http://www.wadsworth.org/newborn/babhealth.htm). Diagnostic testing of newborns is, like all such testing, clearly "conceptualized as offering control, [as] being a way of 'doing something' in the face of the incipient disorder created by the presence or potential of disease" (Lupton 1995, 78).

On a conscious level, nearly all the parents I interviewed accept this view of early diagnosis. They believe it has given or would have given them the much-coveted capacity to protect the child and substantially mitigate the effects of CF. Again and again, they talk about how important it is to be proactive, to have known about CF before symptoms began so as to forestall them, or soon after they've begun so as to reduce their effect. Anthony, for example, says he's "definitely grateful that we found out before she was symptomatic, and we could start treating this beforehand rather than taking the child into the hospital." Francesca agrees: "I just think to be proactive on things is important so finding out when we did [via newborn screen] was just a good time for us." Lilly, who got confirmation of CF at four weeks after a positive newborn screen, regrets those several weeks of prophylactic treatment that were missed before the diagnosis, even though her daughter Mia has grown and developed just fine. "Mia could have started taking enzymes sooner," she says, "so she could have grown better right away from the beginning rather than waiting." Even Leslie, whose son was quite sick when he was diagnosed first with MI and then with CF right after birth, finds herself oddly grateful for

his early illness, since it led them to such a rapid diagnosis. The seriousness of his immediate manifest symptoms pales, for her, in comparison to the potential harm that might have come to John had his disease remained undetected. "I mean we were fortunate that he did have the meconium ileus," she says.

> Even though he had to have major surgery we were able to find out immediately and we were able to start his treatment…. I mean it's still a horrible diagnosis, but… I feel fortunate that we found out immediately… rather than risk lung damage and not have treatment.

These testimonies are just a few of many that illustrate parents' internalization of the assumption "that individuals must have 'knowledge' of their hidden disease, or its precursors, to be able to act to protect themselves against it" (Lupton 1995, 78).

But there are problems, too, that come with knowledge of the hidden disease – complexities troubling to parents even though they say they support newborn screening as a matter of policy. Despite efforts to identify and meaningfully analyze the CFTR gene's multiple mutations, the "clinical course" of cystic fibrosis remains very difficult to predict, even in those cases where both of a particular person's mutations are clearly identified (WHO 2000; Moskowitz et al 2004). It is therefore a case (as suggested in Chapter One) where correlation between the biomedical definition of the "disease" (i.e., abnormalities of structure or function) and the lived experience of "illness" (i.e., the person's perceptions and experiences of the disease) is remarkably poor (Hill 1994, 12; Conrad and Schneider 1992, 30; Kleinman and Seeman 2000, 231-2).

Newborn screen and follow-up diagnostic tests give parents information about the disease, the fact of CF. But the course of the illness, if there is in fact to be one, remains unknown and – at least for the time being -- unknowable. However, once the parent

knows the genetic mutation lies within her child's body, if becomes impossible for the identified disease not to impinge on the life of child and family, even if no symptoms are manifest. As Conrad and Schneider point out, drawing on the classic work of medical sociologist Eliot Freidson, "calling something an illness in human society has consequences *independent* of the effects on the biological condition of the organism…. Medical diagnosis affects people's behavior, attitudes they take toward themselves, and attitudes others take toward them" (1992, 31).

For parents whose child had an early diagnosis but ends up being unaffected, only mildly affected, or asymptomatic for a long period of time, it is illness, not disease, that shapes their parenting practices from the beginning. Yet the illness is not one of physical symptoms. In the absence of manifest signs of the disease, the only "illness" affecting child and family is one of preventive routines, vigilance, and the construction of a CF identity in response to the diagnosis. Parenting is defined by knowledge that the invisible disorder exists, and by an understanding of the disease, and of how it can be mitigated, that is acquired through a combination of medical visits and self-education. Just as a CF diagnosis is very different for those who receive it as an abstract test result than for those who receive it in response to manifest symptoms, so too are "perceptions and experiences of the disease" greatly influenced by the child's actual state of health.

As discussed above, parents embrace the promise of control offered by the early diagnosis with tremendous energy, significantly shaping their early parenting around prophylactic regimes and defining their successes and failures based on the child's state of health. Boland and Thompson's conclusions based on their research with newborn-screened and clinically-identified children with CF and their parents summarize aptly

what parents of asymptomatic or late-onset children in my study experienced. "The period of prophylactic treatment," they write, "was one in which the ethos of 'always being one step ahead of trouble'… developed to a greater extent" than it did among the group of parents whose children have symptoms. This is because, as I have shown, mothers of healthy children often feel that "the child's continued symptom free health (and hence his longevity) [is] dependent upon her competence" (Boland and Thompson 1990, 1243). The experiences parents in my study are having with asymptomatic CF also resemble in certain ways those of parents whose child is diagnosed with PKU. As summarized by Schild, for these parents "the early diagnosis takes on an illusive, somewhat nebulous quality, as [they]… watch the treated newborn PKU child grow and thrive without apparent pathology." But the watching can be fraught with tension since "the parents operate under a constant threat that if they fail in maintaining the diet, their child will become brain damaged. They tend to respond with excessive monitoring…" (Schild, as quoted in Clayton 1992b, 112) Although the extent of the damage that can be caused by a break in prophylactic vigilance for CF is less dramatic than in the case of PKU, the focus of parental energy on preventive care is nonetheless intense.

Mothers' competence is surely formidable, and many of those I spoke to felt certain that their care and vigilance was resulting in better health for the child. At the same time, at least one mother found herself practicing all that vigilance for a child who is unlikely to get sick at all. Reflecting on this aspect of her own experience with CF diagnosis, Shannon said this. "With the information that we have [now], I don't think she'll ever have any health issues with cystic fibrosis, and pray to God she doesn't. But the thing is, is yeah, I don't know if she's ever going to have anything to deal with with it, so

therefore it's like, okay, the tears and the worrying, what have I gotten for it?" In her case, the only illness present was the one manufactured by the diagnosis. And her vigilance, clinic visits, and preventive routines likely have nothing whatsoever to do with the blessing of Margo's good health.

Other parents, like Kate, may not know for some time if their child will ever develop symptoms or not. These parents can only hope their experience will resemble Shannon's, since a child who never gets sick is what they all want. But in the meanwhile they are subject to guilt and self-blame at the very slightest sign of a physical or developmental problem, since they are practicing active early intervention with such fervent hopes of forestalling each potential effect of CF.

For parents who got an early diagnosis and are now beginning to see undeniable signs of CF despite their every effort, the discourse of control presents a different kind of challenge and heartache. Parents' devastation at the diagnosis is heavily mediated, if the diagnosis is made early, by the discourse of prevention and risk reduction: repeated assertions by health-care providers that it is best to know early because useful interventions are at hand breed an inevitable hopefulness. For many parents, though, the hard truth is that the degree to which any real "protection" from the disease can be offered through early intervention is minimal, and that their own power to protect the child is woefully insufficient in the face of deteriorating health.

Paige feels the anguish of this helplessness in every fiber. On the one hand, she is grateful for the early diagnosis, and enthusiastically invested in early intervention.

> Kaya's lucky that since day one she's been getting treatment, we've been doing everything since day one.… We've always been able to know that she has it and I, we've been very aggressive in her care.… Rarely there goes a day where we

> don't have her do her chest therapy, or her medications. We're very diligent, very aggressive. At the first sign of cough or illness, I call the physician after I've kind-of examined her myself to say okay this is what I think is going on, and what's helpful… I'm happy that [the newborn screen] was done, that it was caught early, that we could start treatment early….

At the same time, however, she articulates how impotent she ultimately feels when it comes to protecting her child from CF.

> The hardest thing is as a mom, you're supposed to fix everything, you know. You're supposed to fix the owwies and the booboos, and you just can't fix this, there's nothing you can do. And I think that has been so hard for me, `cause I can't control it at all. I have no say in how this disease ravages on her body. I can do what I can with treatments and, and medications, but other than that I have no say in what it does… [it's] like [CF is] taking away my chance to give her everything I wanted to give her.

Expression of two conflicting emotions on this topic, two realities, is not uncommon. Parents feel, at once, powerful determination to protect the child from the disease, and powerful despair in the face of what sometimes proves an unconquerable opponent. Both sentiments undoubtedly reflect an important truth about the complicated matter of parenting a child diagnosed at birth with a genetic disorder. But the latter part of the story, the part about how hard it is to have knowledge of the disease and not be able to "fix everything," is often drowned out -- in the popular press as well as in the construction of most research in this area -- by the dominant narrative of progress and improvement, of extended life-spans and enhanced quality of life. Even for parents who find out early, and who have access to high-quality health care and adequate financial resources, control over the disease has definite limits. In most cases,[54] substantial

[54] As noted in Chapter One, there do not appear to be any reliable data estimating what percentage of CF mutations are considered severe, what percentage moderate, and what percentage mild. Among the reasons

suffering and a substantially shortened life span will be their children's reality no matter what. Paige captures here how very painful, how really excruciating, this knowledge can be.

> I could flash, like flash in my head, seeing my daughter, my daughter coughing like [others I have seen with CF], my daughter struggling to breathe, um… and how, how [am] I gonna to handle that? I struggled with questions in my head of how are you gonna handle this when it's Kaya…. It's so scary, and you read things too, I just can't imagine, um, how it feels, you know. I wish I could be her. I think right now she has very normal, um, she doesn't have normal lungs, but she has normal lung capacity, and I think she… does not struggle to breathe, I think she feels that she gets enough air in. But I think, when she gets older, how is that gonna be? What's it like? Is it, you know I've heard people describe it's like suffocating, it's like you can't get enough oxygen, you can't get enough air, like you're starving for it. And how awful that has to be…you know, how awful, it's just, I'm just so sad, and so scared for her.

It's a Way of Life: Children's Adjustment to the Preventive-Care Regime

Children as well as parents must adapt to the routines of preventive care at home and to the frequent clinic visits that follow after CF diagnosis. Parents spoke often about the phases children seem to go through as they grow, noting that their willingness to do what needs to be done can change over time. However, parents of children diagnosed early see the immediate establishment of care practices as a distinct advantage. As Joan put it,

> when they are diagnosed that young, it's nice because it's a way of life for them, they don't know anything else. Versus if you have an eighteen-month-old and all of a sudden you have to do all these treatments and medicines and it's all new to them. So it's kind of nice that she's never known any thing else.

Andrea has a similar sense that things are easier for her son because treating CF has always been part of his life. At age six, "he doesn't ask a lot of questions," she says.

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for this is that persons with mutations leading to very mild symptoms or no symptoms at all were never identified in a systematic way before newborn screening began in selected states.

"He's so used to it he doesn't even notice that his classmates [don't] have to go to the office for medications before lunch, and he doesn't even notice that his classmates don't have to eat extra food or don't get practically a meal for snack in the morning." Leslie says her son just assumes "this is what everyone does." He has been getting the chest physiotherapy daily since he was two weeks old, and "it doesn't bother him."

As children grow older, parents must begin explaining to them directly why care routines and precautions are necessary. Preventive health practices thus become an explicit as well as implicit aspect of parenting, and part of the parent's work is to teach the child herself to participate in the never-ending routines of illness prevention. Francesca describes here her efforts to keep her daughter and her nephew, who also has CF, apart when one of them may pose a danger to the other.

> When one of the kids gets pseudomonas it's hard because you have to try to keep them apart, and you know it's just it's trying at some points because they're close…. It's hard to tell Tess that [she can't]… go play with C. [I say], 'Well you can't.' 'Why?' I'm like, 'You're sick.' [She says] 'Well, I don't feel sick… I'm not sick' [I say] 'Honey, but you're always kind-of sick, you just don't realize it.'

Parents with early diagnoses are relieved that they don't have to suffer through a transition with their children, going from "normal" life before the diagnosis to an existence full of treatments and precautions afterwards. They feel the child has been spared the pain of losing prized freedoms, and they themselves have been spared the frustration of introducing new routines into a life already built without them. Indeed, some mothers who got a later diagnosis describe their experiences with just such problems, as did Erica in the quote above. But, as Francesca's observations illustrate, the formation of precautionary habits and schedules of care at birth also signifies the

development of an "illness identity" right from the start. Children are "always kind of sick," even when they "don't realize it." Parents, as I quoted Betty saying above (see page 136), "get to know [their child] as somebody with CF, not just get to know her for her and then figure 'oh, she's got CF.'"" Or, as Kate put it:

> I know people say she's a baby first and then she has CF, [but] I can't divorce the two. It's like, my baby has CF. I'm at the gym giving her enzymes, so people see it and they ask me questions about it and 'Oh my God, you're giving your baby applesauce, it's too early.' It's so funny, because people start to go berserk, and it's like 'She's been doing this since she was three days old, I think she's fine, thanks!' And so it's, it's so tied to me as who she is….

And it seems likely that's how baby Erin will think of herself, as well.

What if Tomorrow Don't Come?: The Preciousness of Time

Knowledge of CF makes parents more vigilant, more watchful. It opens them up to the grief of possible suffering and death for their child. It thoroughly reshapes their parenting routines if they already had them before the diagnosis, and shapes them from scratch it they did not. But many of the parents I interviewed made sure to let me know that it can also serve to highlight how precious time is, and how important it is to live well in the moment instead of squandering it. Like those who Frank describes as adopting a "quest narrative" in order to make sense of their illness (see Chapter Three), parents in my study often talked about how they have *used* their child's CF, making it "… the occasion of a journey that becomes a quest… [and] the quest is defined by the… belief that something is to be gained through the experience" (1995, 115).

Regardless of when the diagnosis comes, it can inspire a sense that the present should be lived to the fullest. Plans for trips and for engaging in favorite activities should be

acted on right away; postponement suggests the luxury of an indefinite future, and parents with CF know better than to assume this. As Francesca puts it,

> If we plan on doing stuff we kind-of make sure we follow through on stuff like that, just knowing that our days aren't always guaranteed. And not even the worst-case scenario that she would pass on, but just sickness-wise we don't know what's gonna come. So let's just take what we have now and do what we can with it. Maybe if she didn't have her CF… we wouldn't put such a priority on doing some things, `cause it just makes you think like what if tomorrow don't come? What if we can't put it off for five years? `Cause we don't know where she'll be at with things.

This profound appreciation for the preciousness of time, and for the wisdom to cherish the child each day, is felt by some parents to be the compensatory gift CF has to give. Paul describes it this way:

> When the diagnosis was made I definitely feel that in me I gained an additional sense of appreciation and attention, specific attention to just you know everything that made her her. You know everything [that] made [Alexandra] who she was…. I definitely appreciated the time that we had with her [before] but I think I appreciate the time more now when I look back in retrospect… because of the fact that we now know she has cystic fibrosis. It's also changed the way I spend time with her as well. I spend more time with her.

For some parents, the affirmative desire to get the most from every day with their diagnosed child becomes entangled with a temptation to become too permissive and lavish. This urge to "spoil" the child is fleeting in some cases, a phase parents go through on their way to settling into a more normal routine and a steadier state. This was the case for Nancy, who quickly reined in her impulse to spoil the child because she felt so keenly the truth of her health-care provider's admonition that "you don't want to spoil CF kids because, and basically this sounds horrible, but nobody is going to like a child who is sick a lot and coughs a lot and then acts like a brat." Annie had the same experience, and feels

the best advice the doctor ever gave her was not to treat her son any differently the day after she learned about his diagnosis than she had every day for the past seven years. "If you do you're going to ruin him as a human being," the doctor told her. "If he's bad you crack him on the butt, and when he does something good, you hug him and kiss him."

For other parents, over-indulgence just becomes an indisputable fact of life. As Sheri says of her son Jasper, reflecting on what changes the diagnosis occasioned, "If anything, he's more spoilt than he would have been." Francesca too wants to give her child "extra treats, or I wanna let her do extra things."

Spoiling, however, does not seem to be the most crucial issue even for those who raise it. Far more important is the transformation of priorities that can occur. "I really cherish my son," says Roberta,

> and I do wonder how much it has to do with the diagnosis, through thinking you know the time really is precious…. He's really healthy he's doing really well, he's bigger than most kids his age, he seems healthier. But… I think we decided right after he was diagnosed that because we didn't know what could happen, we still don't know what could happen, we decided that we were just going to make his life the best life that we could make it. And… that's still, that's still how we live…. If he hadn't been diagnosed, if he had just kind-of been normal, I mean maybe there wouldn't have been that….Maybe there wouldn't be a lot of the attention.

For Judy, priorities changed in a different, but equally poignant, way. Inspired by her daughter, she became very active in the Cystic Fibrosis Foundation. As she tells it,

> [O]ne day my oldest one, it was within the first year she was diagnosed, she came downstairs and… she asked me if the doctors had found a cure yet for CF. And I said 'Oh, not yet, but I'm sure we'd be the first to know if they did.' And she says, 'But Mom, you're not even looking!' And I thought, 'You know what, you're right, I haven't done anything to help the doctors try to find a cure….' [A]t least [now]… I'm trying to raise money so that they can get all that they need to try to find a cure.

These responses to the CF diagnosis show a remarkably resilient side of parents – their ability to learn something important, to create something worthwhile, even while experiencing, on other levels, grief and devastation. These heartfelt descriptions of personal transformation also illustrate – along with the other data presented in this chapter – how very profoundly the diagnosis influences parenting and family life. Clearly, as we move towards a system where more and more diagnoses are made at birth, there is a great deal at stake for parents and children and for the relationship between them.

Chapter Five

CF Odysseys: Parents and Professionals

Parents do not encounter CF -- at least not for the first time -- on their own. Unlike the discovery that one's baby likes to nurse on the left better than on the right, or that she has a pointy chin like Uncle Joe, CF is not usually identified by parental observation alone. Rather, as I noted in Chapter Two, a major feature of finding out about CF is the immediate and pervasive contact it brings with health-care providers - first those who are involved in one way or another with the diagnosis, and then the "army" of professionals who make up the treatment team. Getting the diagnosis, and then learning to live with it, are not private family matters. They are public ones, heavily mediated by those who test the baby, give the news, impart instructions, provide education and treatment, answer questions, make referrals, and generally interpret the meaning of the condition. We have seen already how the diagnostic process – its timing, structure, and delivery – has an impact on intra-familial relationships and practices. This chapter examines how that same process affects the development and assertion of parental know-how, as well as power dynamics in the relationships between parents and professional service providers.

"You Guys Are the Doctors": The Power of Professionals After an Early Diagnosis

It is a truism that all parents, no matter who their child is and what the state of her health, gain confidence as caregivers over time. Certainly some of us start out with more confidence than others when tending a newborn, but none of us are as sure of ourselves at the beginning as we are later on, after trial and error and the sheer number of hours we spend parenting have taught us what we need to know. The same trajectory of less to

more confidence holds true with respect to caring for a child with a genetic condition. As one parent put it, "as you deal with a disease you become educated. You become educated about the disease process, [and] you become educated about your particular child." In what follows, I will call this knowledge, which is based on experience, observation, and experimentation -- rather than on what is codified in books and generalized professional systems -- "parental knowledge" instead of "lay knowledge." In so doing, I follow Stacey, who favors the term "people knowledge" and writes that the term "'lay'… tends to be what is used for those people who do not belong to a specific profession. In referring to people who lack particular qualifications," she goes on, "'lay' suggests the absence of something valuable or prestigious, and may imply less competence, or even less moral worth." (Stacey 1994, 90) Like Stacey, I assume we *all* develop and deploy useful knowledge, and that the technical expertise of professionals – while certainly invaluable in many situations – is not "superior" *per se* to other kinds of specialized capacity.

Time is certainly one important factor in the development of parental knowledge and confidence. But the diagnostic process, which permanently connects most parents to the health-care system[55] -- complete with its array of professional definitions and norms -- also has tremendous influence over when and how parents mature into a sense of their own competence and the need to assert it. As Taner-Leff and Walizer note in their book

[55] I was not able to find, in all my research on CF, any data about what percentage of families with a diagnosed child are not connected to consistent medical care for CF. Researchers estimate that 80% of diagnosed children are seen at one of 115 accredited CF Specialty Centers nation-wide, but it is not clear what proportion of the remaining 20% are treated in other settings, and what proportion are not receiving medical care of any kind. I have also been unable to find any data about what proportion of families might be using some form of "alternative" or "complementary" health care instead of or in addition to allopathic medicine. I did not ask specifically about this issue during my interviews, but none of the parents in my study noted making use of any form of care outside the allopathic norm during our extensive conversations about the professionals and treatment routines in their lives.

about the relationship between parents, professionals, and children with chronic illnesses/disabilities, the diagnostic process "sets the stage for all future parent-professional encounters" (1992, 69). Further, "[P]arents NEVER forget what is said at the time of diagnosis… It is at this intensely painful point in the parent-professional relationship that issues of trust, caring, and mutual respect are emblazoned in bold relief in the family's memory and consciousness." (Ibid., 96 and 69) My research corroborates this finding. I also found that the timing and structure of the diagnostic process affects the confidence parents have in their own power, instincts, and knowledge, as well as the amount of dependence they feel on health professionals.

When the diagnosis comes early, before the parents know the infant themselves, no substantial parental expertise has yet been developed. The baby is brand new, and as I discussed in Chapter Two, even learning how to care for her in the absence of any identified health issues -- particularly if she is a first child -- can feel overwhelming. This is a time of vulnerability, of trial and error, of just trying to learn “how to be a parent.” Professional advice of all kinds therefore exerts a particularly powerful influence during this time.

There is, of course, no shortage of such advice directed at all new parents, whether their child has any "special needs" or not. As noted in Chapter One, the process of child-rearing in the twenty-first century is heavily mediated by professionals of all kinds. As Rose puts it:

> Childhood is the most intensively governed sector of personal existence…. The modern child has become the focus of innumerable projects that purport to safeguard it from physical, sexual, or moral danger, to ensure its ‘normal’ development, to actively promote certain capacities or attributes such as intelligence, educability and emotional stability (1989, 123).

"Expert" parenting advice in the present era focuses on an ever-broadening array of themes. These include, among others: the need for bonding versus the need for discipline as the parent's guiding principle (Hulbert, 2003); the importance of cognitive development (Wrigley 1989); and -- most relevant to my study -- the imperative to identify and protect against multiple intellectual, developmental, moral, and physical threats (Stearns 2003).

I would argue that early diagnosis of genetic disease via newborn screening as a technique for identifying potential physical threats has now become an accepted part of newborn medical care for well infants, and moreover that its influence will only continue to grow as testing panels continue to expand. With the mapping of the human genome and the subsequent emergence of tests capable of identifying mutations of all kinds, the baby's genetic endowment has become a map (Katz Rothman 1998) that experts try to read and interpret The capacity to identify genetic conditions is far more sophisticated than the capacity to usefully treat them (Hubbard and Lewontin 1996; Shakespeare 1995; Nelkin and Tancredi 1994). But once the possibility of these conditions is mapped, professionals – and, in their wake, parents -- feel bound to treat the child according to the markers on her genetic map.

In the best-case scenario, the tests promise greater control over health outcomes for the child in the long run. When parents want to know if a disorder exists, it is usually because they want to control what happens to their child as much as possible, because they want to protect her. In the case of CF, as I discussed in Chapter Four, parents certainly make every effort to use diagnostic information in that way. But when CF is

first added into the newborn picture, parents themselves feel even *less* qualified to be in charge of the infant than they did before. At that crucial and vulnerable moment, parents see professionals -- the ones who have made the diagnosis – as the ones best positioned to understand the situation and chart a course of action. As Joan describes it:

> Well, I would say in the very beginning [when we got the newborn screen diagnosis] we were kind of like, me and my husband, we were both kind of like what do you [doctors] want to do, what do you think we should do, what do you think is best? You guys are the doctors, you know.

A diagnosis by newborn screen creates an unusual dynamic between parents and providers. The medicalization of childbirth in the United States[56] has created a situation where birth is considered a medical problem that must be managed by health care professionals in the institutional setting of the hospital. Indeed, 99% of babies are born in hospitals (Curtin and Park 1999).[57] In this environment, obstetrical specialists structure the birth experience itself, and then pediatric specialists step in to structure the care provided to newborns. Mothers may have more access to their babies while still in the hospital than used to be the case, since some hospitals now "allow" the baby to stay in the room with her mother most of the time if that is what the mother requests. Even in such an instance, however, the infant is still in a liminal state. She officially belongs to her parents, yet she remains predominantly in the care of the hospital's medical staff. As such, she is subject to their rules, schedules, routines, and diagnostics – including newborn screening tests to determine genetic abnormalities that might, according to the medical orthodoxy, require additional medical interventions.

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[56] See for example Katz Rothman, 1989; Ehrenreich and English, 1978.

[57] Even babies born at home are subject to mandatory newborn screening, however, since the medical professional attending the birth is required to "arrange for collection and submission of a specimen from the newborn between the third and fifth day of life" (NYSDOH 2003, p. 2-2).

Newborn screening is, as we have seen, certainly not a parent-driven process. Often, parents don't even realize it is happening, or if they do, they understand it only vaguely. And often, the child with a positive screen for CF is not actively ill in those first days or weeks. She may be entirely without symptoms, or she may be showing some early signs of illness, usually digestive in nature – but in any case, she's not in a medical crisis, and thus is relinquished from the hospital into the care of her parents, who begin learning what it means truly to be in charge of their new baby. Yet when the infant, already at home, is found to have a positive screen, the health-care establishment once again asserts its dominance over the fragile new family dynamic by remanding parents to a role of simple compliance with instructions coming from the professionals. In the words of New York State's newborn screening pamphlet, their job is to "make it easier for the doctor to help the baby."

What parents confront when there is a positive screen, then, is a specific form of medicalization, of that process (so thoroughly documented by sociologists) by which "medical interpretations of conditions… acquire cultural legitimacy… [and] become the dominant frame for understanding a large range of… human problems." (Litt 2000, 4-5) The very fact of the newborn screening test declares the importance professionals attach to finding out about and addressing even unexpressed genetic disorders as early as possible in infancy. But from the perspective of the parents, they receive this diagnosis at a time when they are just finding out for *themselves* how to care for their baby. Now they must learn from their *doctors* what to look for and see in the child -- how to recognize, forestall and address symptoms; how to minimize the impact of CF to the extent possible; how, in essence, to "parent in the genomic age," when part of having a new baby is

knowing about and responding to her genetic endowment. And they must learn all this while continuing to care for the child at home, in an environment with restricted access to those very professionals who have just constituted themselves as indispensable to the health and safety of the infant.

Once the CF diagnosis is confirmed, parents generally have a first clinic visit where they meet an interdisciplinary team of professionals: nurses, nutritionists, genetic counselors, pulmonologists, gastroentorologists, social workers, and more. They receive large amounts of information and instruction (see below), and then they go back home, where they alone are responsible for carrying out the daily preventive care regime. Parents describe these first weeks and months with the diagnosed infant as a time when they struggle mightily to understand what CF is and what it means for this newborn whose already-inherent fragility -- perceived automatically to be considerable just by virtue of her small size and recent delivery -- now feels like actual peril. In order to manage this stressful situation, many parents rely extensively on contact with professionals via frequent office visits, or via the telephone.

These forms of contact are experienced by parents as a vital connection to expertise, to advice, to reassurance, to instructions. Unsure now about whether the infant is okay, about whether their own care-giving techniques are adequate, they contact professionals often, looking for help. Anthony describes how things changed after the newborn-screen diagnosis like this:

> I guess our biggest concern is that she goes to the doctor often and my big, my concern and I think my wife's as well is [that] the day that we kind of are relaxed and go, 'well it will pass, let's give it a few days' is the day that something major happens. So we always err on the side of caution. You know we'd rather be a pain in their side.

For Franscesca, the fear after the positive screen and subsequent confirmatory test was intense, unremitting. "Once I found out that she possibly could have the CF, I called so many times in the middle of the night. I'm like 'Oh my god, she's breathing really heavy, I don't know if this is right.'" Fortunately, Francesca's health-care providers, particularly the nurses, were compassionate, and comfortable with making themselves available to her as a resource. "The nurses that were on call," she remembers, were

> my biggest help because you could call these people up anytime and even if it would've been a dumb question to them they were very kind about answering them and just you know, like assuring me that I was doing everything okay and everything was fine…. There was just a lot of follow-up that came from the hospital that helped.

Paige had a similar need for help from professionals, a similar reliance on them for guidance in caring for a baby that the diagnosis had made suddenly strange and vulnerable – a baby whom she could no longer know based on observation alone. But for her, the process of getting assistance was complicated by her sense that her health-care providers regarded her as overly needy.

> What was hard I think in the beginning [was] being new as a parent for one and not knowing what was normal for children… and then dealing with the disease, the health care…. The people in health care were somewhat hard to deal with, because I sometimes would get the feeling that I'm a little pain in their ass, excuse my language…because I would call a lot because I didn't know, because I was so scared, because there was such a fear…. I would call, I would call the nurse a lot and say 'I don't know if this normal or not, this doesn't seem right.' And a lot of times, you just felt like you were a real pain or like you were kind-of a nuisance and that was hard.

It is clear that during this period parents perceive the locus of control over their child to lie with professionals. As Joan says, "I remember thinking, 'I can't believe they are

going to let her come home with us, at the clinic. We have no idea how to take care of her. I can't believe they are going to let us walk out of the door with her.'" She had, of course, already walked out the door with her newborn when she was discharged from the hospital after birth, had taken her home and cared for her just fine. But once they got the diagnosis,

> it just seemed like everything changed…. It was like there is so much more now to taking care of her, and are we really fit to do that?… [I]t was just so overwhelming. I mean the first time we went to the clinic they were like well, you have to do this and this. And we met with nutritionists, respiratory therapists and pulmonologists and socials workers and you know it was just all so overwhelming, all this stuff we were going to have to do. I remember leaving there thinking 'how am I going to do all this stuff in one day?'

The dramatic decline in her self-confidence as a parent, in her sense of the adequacy and value of her own parental knowledge, couldn't be more clearly expressed.

It is scary and difficult to be immediately responsible for an infant's complex new care regimes at home, but it may be even harder to get a diagnosis for a non-symptomatic or minimally-symptomatic child while still in the hospital. Only one parent in my study -- Suzanne, the mother discussed earlier whose baby was at known "high risk" for CF because both parents had carrier-tested positive, and who was delayed in the hospital because of iatrogenic problems -- had this experience. For her, "everything changed" in the most literal sense after the test result came back. The hospital staff, who "were not used to dealing with babies with anything wrong," took charge of baby Quinn, no longer allowing Suzanne to hold or to nurse him. "They just got a little bit over the top," she says, "cause he was having feeding problems so they sort of attributed everything to him

having CF. Which we later found out wasn't the case at all. So they, they were 'he's too tired for you to hold him,' or whatever, so that was their decision." Suzanne says that "a lot of Quinn's symptoms, like he didn't appear to be sick when he was born," yet the hospital treated his diagnosis almost like a medical emergency, controlling every aspect of his care minutely and completely marginalizing Suzanne. "I got really depressed," she recalls. Yet at the same time, she was grateful for the early diagnosis. Like other parents in her place, as a young, first-time mother she was in no position yet to counter or question medical authorities when they recommended immediate testing.

> How it had been explained to me, the sooner you find out about it the better the outcome for the child. Which I could understand once he was born. That it was important to know cause it meant *they* could start treatment straight away instead of having to wait for two months to find out whether he had it or not [via the regular newborn screen] (emphasis added).

Quinn's at-risk status before birth, and his quick diagnosis afterward, firmly situated him as a medicalized infant from the beginning. Suzanne is unhappy with the way events unfolded at the hospital where he was born. It was emotionally devastating to lose control over him when the test result came in at age four days. Yet on balance she is still glad to have gotten the diagnosis immediately. She fervently wants what is best for Quinn. How could she help but believe that "they" who had advocated an early diagnosis were also in the best position, ultimately, to assure his well being by "starting treatment right away?"

"You Have to Be an Advocate for Your Child": Diverse Pathways to Parental Empowerment

Parents who have lived with and cared for their CF-diagnosed child over a significant period of time understand clearly that they themselves are not only the child's central caregivers; they are also her best advocates. They need their health-care providers, but experience has taught them that "as a parent, you have to coordinate everything," and that "you have to be a good medical consumer… you have to be an advocate for your child because [the doctors] don't know it all." As Taner-Leff and Walizer put it, parents learn that "in the bureaucratic health care system of changing shifts, multiple personnel, and inconsistent caregivers, the parents are the child's constant protectors" (1992,152). Parents enact their advocacy work in different styles. They develop many different ways of working with, relying on, and using health care professionals. But all find over time that it is not really the professional "they" who are keeping the child as healthy as possible, but rather the parental "we."

For parents who get an early diagnosis, or a rapid diagnosis for an otherwise-relatively-healthy child who suddenly develops a tell-tale symptom (e.g., rectal prolapse), the diagnostic process itself may be the first occasion that brings them into sustained contact with health-care professionals outside routine preventive care. For these parents, the sheer number and diversity of providers is bewildering at first, and it takes time to sort out who does what: who does primary care, and who provides which specialty services related to CF.[58] As Nancy put "I didn't know what my rights were as a parent or

[58] The relationships of parents to each of these groups of professionals, as well as the connections among providers themselves, are complex and multi-dimensional. Parents did talk in my interviews about some issues relating to differences between primary and specialty care, and what was helpful or problematic for them in each arena. In future research, it would be useful to gather data on this issue more systematically.

a patient. Like can we switch doctors? I didn't know anything; I had never been through anything like this. I mean I was unfamiliar with doctors in general… I didn't know anything about the medical field." Their crash course, their on-the-job-training in advocacy thus starts with the diagnosis, and they have little prior knowledge to draw on at the beginning.

Joan, who is quoted above, is a clear example of someone who felt completely reliant on professionals at the beginning. "When she was diagnosed… we were just so blind going into it," she says.

> But the more we lived it every day, the more experience we got, and now we tell the doctors 'no we don't agree with that, we think you should do it this way.' So we are more vocal and more advocates for our children, because we know our children and we believe that we are part of the CF team and their doctors are pretty much in agreement with that [now] too.

Like Joan, Betty -- whose daughter had MI and was diagnosed shortly after birth -- learned as she went.

> In the beginning, everything that any doctor ever had to say was gospel. You took that to be the truth, you took that to be the absolute answer. That it was correct, everything and anything that they said you agreed with. You did not question, that's just the way it was. It took a long time… to get an education within myself… that I needed to question some of the stuff that was going on with Rose. That I needed to make sure that I was the one who knew everything; everyone else didn't know everything about Rose…. *I* needed to advocate for Rose.

With experience, Betty did come to know "everything about Rose," and she certainly no longer takes anyone else's word as gospel when it comes to coordinating, overseeing, and managing Rose's care.

However, for the purpose of my overview-style analysis in this chapter, I have lumped providers together rather crudely instead of attempting a finely nuanced examination I don't feel I yet have the data to support.

> Now I'm not above calling and telling anyone what I think about them or their assessment of my child. And I try to do so as respectfully as possible, but when I obviously disagree with someone, I'm certainly not going to hide it. And if I think that they're not listening to me, I don't, I don't stop…. I'm not afraid to ask for what I think Rose needs, and I think that I have certainly evolved in that respect.

Leslie also began with much more reliance on her providers and grew to be assertive, to make her own judgments and speak up about them, over time.

> I think you know we're probably more, we take more care of John's health [than we did in the beginning]… we're more proactive….If something doesn't sound right or if we know there's something that's not quite right with him we'll push for it and say 'this is the way it's going to be.' I mean we try to we try to play nice but if there's something that we just aren't in agreement with we'll speak up… [if] there's something that's not quite right we'll push a little more…. You know most people, their doctor tells 'em something and they don't question it, and I guess we, we'll question things more.

For Paul, the process of becoming an advocate had a lot to do with getting educated about CF on his own, taking charge of the research process himself rather than simply trusting that the doctors were already experts. Over time he developed multiple sources of information and ideas instead of relying solely on Alexandra's doctors, who may have had general medical expertise but didn't share his own passion for every detail about the specifics of his daughter's condition.

> From the beginning you of course… you're skeptical, but then you put your trust in [the doctors]. Then we [did] our research, and you feel that you get educated well on some things, and when you ask a doctor maybe about that topic… and they just kinda brush that off… you maybe have a negative opinion of the fact that they're not keeping up, maybe they don't know what's going on, maybe *you* know [more than they do] …. [W]e can never know how much they truly know, and we also have a different vested interest.

Kate got a prenatal diagnosis. Her baby was still only four months old at the time of our interview. She described herself as already "proactive in analyzing everything," but understood that she was still in the process of gaining the degree of confidence she might have later on.

> I'm probably bugging the hell out of the CF clinics, but until I truly learn all the signs… I call… the doctor immediately…. Somebody had said to me, you know, some mothers just increase the enzymes on their own when they think it's time.… I haven't done that yet, but I probably don't see that out of the realm of something I would do.

Though she's not ready to alter enzymes without medical advice just yet, Kate educated herself thoroughly about CF before the baby was born and feels she therefore started off with a bit of a leg up on the whole medical process. "When I was pregnant," she says, "a lot of stuff was coming in but I wasn't fully understanding it. And then when she was born, I started applying some of the things that were in my head."

Both Kate and the other mother in my study who had gotten a prenatal diagnosis for a first child had experiences during pregnancy that showed them how central their own role in protecting the new baby would be. Those long months of prenatal care and preparation for the birth of a child with a known diagnosis taught them quite a bit about the health-care system and what it will take to parent a child who is anticipated to have special needs within it. In both cases, skepticism about professional knowledge and the formation of an advocacy identity came very early. This may be partly because their obstetrical providers had no experience with pregnant women carrying a baby with a CF diagnosis to term. Rather than being confronted immediately with a cadre of experts who seemed to know a good deal more about their baby than they did, Kate and Crystal found

themselves instead in a situation where their providers knew little or nothing. As Kate put it:

> Every single one of them [in my obstetrical group], I would ask them, 'Have you ever had this situation?' They were always like 'No, you're the first….' And they get together as a group, you know, every week, and so they all knew about me before they'd even met me because I was just this interesting case. I'm like, 'I don't wanna be an interesting case!'

And from Crystal:

> [My prenatal care] was like stressing me out because my OB just treated it like a normal pregnancy. And I didn't know, it was all so new to me…. I had seen other people writing online about that, about doing ultrasounds to check the intestines, I think…. I finally got out of my OB that they had never been in this situation before… we asked them finally, because my first question for them was 'Do you feel okay being my doctor? Should I find another doctor?' And… by the time I really started to get uptight about all of this it was almost the end of my pregnancy, and I was like I can't switch doctors now…. [But I wondered], should they be doing something else, should they be doing more ultrasounds to check… it's hard to coordinate.

For Crystal, the lack of experience on the part of her obstetricians presaged a lack of experience on the part of her pediatricians, and also the fight she would have to wage about insurance issues.[59] When she was pregnant, the obstetricians could not or would not answer her questions because "they would say, 'Well, that's the pediatrician's [area].'" Then, once the baby was born, the pediatrician "didn't even know that Martin had CF…. and that pissed me off, because he should know that. I-I called their office so many times, he didn't know he needed an RSV shot `cause he didn't know he had cystic

[59] Crystal was just one of six parents – five of whom received the CF diagnosis by newborn screen -- who spoke extensively about their difficult experiences with and/or worries about insurance issues, despite the fact that I did not ask any specific questions in this area. These data, preliminary as they are, suggest that commentators' worries about the impact of newborn screening on insurance coverage are shared by parents.

fibrosis and I had to tell him…." By the time the baby was a few months old, Crystal had come to the terrible conclusion that "nobody, nobody cares about Martin, I mean, that's just the way the world is." If she didn't look out for him, nobody would. Thus her training for advocacy began with the prenatal diagnosis and propelled her into full swing earlier than those who started with the diagnosis at birth.

Parents whose child has symptoms before the diagnosis stand in a significantly different position with respect to the advocacy process than do their counterparts quoted above. These parents have already spent a significant amount of time both caring for an ill child, and seeking out medical assistance of one kind or another. They have cut their teeth as advocates for their child before the diagnosis, and thus enter the post-CF landscape ready and able to assert the value of their own experience, interpretations, and beliefs.

Catherine, who received the CF diagnosis when her child was nine months old and already symptomatic, asserted her own knowledge of Joseph's condition and needs from the outset of her post-diagnosis interactions with professionals. When medical staff told her they planned to hospitalize her baby after witnessing first-hand a symptom she herself had encountered and managed at home for months, she had no difficulty challenging medical authority and insisting that the baby be tested instead on an out-patient basis.

> The one thing I have learned [from the delayed diagnosis] is that I now fight harder for what I want. I don't stand there and let doctors look at me and think 'oh she's crying, she's a mess, she's a first-time mom.' After his diagnosis… we were at [name of clinic] and he had some severe reflux… he threw up all over me, like all over my dress, all over, and it just didn't stop, it kept coming and coming. And they thought he had intersussception, which is when your bowel twists inside your body. And they wanted to hospitalize him and I stood there and said 'no way… I'll come back tomorrow for any tests you want… I'll come back, but I'm

> not staying here. He does this all the time; he's been doing it since he was born…. I'll be here at 5:00 A.M. if you want, but you're not making me stay here.' I was just like 'no way, you've got to be kidding me. I've been complaining about this since birth and now finally someone's going to take notice and you're going to admit me? I don't think so!' And he didn't have it; he didn't have intersussception, he just had severe reflux. Which I had known all along.

Another mother articulated her confidence in her own superior knowledge of her child and his needs – gained over time, and radically different from her earlier reliance on leadership from her health-care providers -- this way:

> You can't sit back and let them misdiagnose something when you know it's gonna be wrong. You know when they're telling me to give a cough suppressant to him and I know it's gonna be wrong, I can't sit there and go 'okay.' I have to tell them 'no, you can't give this child a cough suppressant. You need to call his pulmonologist… and you get back to me with the correct answer.'

Erica, who has three children with CF, two of whom were not diagnosed until ages 6 and 4, also felt empowered as an advocate from the beginning. She had a lot to learn and found the adjustment very difficult, but she evaluated her providers on their merits from the start, changing doctors and pushing for specific kinds of treatment as needed. A third mother asserts that getting the diagnosis later, after symptoms, "makes you realize that medical professionals are… not always right, and they don't always have all the answers. And probably I should trust in myself a little bit more." It's not that the new routines of care and the dynamics of the disease aren't overwhelming; they still are, even for the seasoned parent. The difference is that these parents know right away that when it comes to caretaking and really understanding what's happening with the child, "it's more the parents" than the health-care providers. Diagnosis of a genetic disorder may be no less devastating for parents who find out later, but it is certainly less intimidating. The child

has already been in their care for months or years, and they have already established their own competence. Knowing the child has CF changes many things for them, but it doesn't change their own baseline of parental experience.

For some mothers, having parented one or more children before the child with CF comes along also provided a baseline for advocacy and assertiveness during and after the diagnostic process. Lorraine, the mother whose infant daughter's diagnosis precipitated the diagnosis of a healthy older brother, is a case in point.

> Well it was very interesting because I was fortunate in that I was a very well established nursing mother when my daughter was born and I was very comfortable with nursing and I knew I had enough milk output because my son had done so well. So I pretty much immediately sensed that something [else] was going on [when my daughter failed to thrive]. We were regularly seeing a pediatrician and you know I brought her in and he would look concerned but it's really interesting because the immediate thought was there was something wrong with my nursing or my milk rather than something going awry with her digestion. And even in fact my doctor asked us to stop nursing and feed her formula, which for most nursing women would have been the end of their milk production. But I knew enough to and had enough confidence to continue pumping and freezing the milk that I had pumped for basically ten days. She didn't do as well, she lost more weight on the formula than she did on the breast milk. And you know then there were all kinds of theories that my milk didn't have enough fat in it, or that we perhaps weren't actually feeding her. There's all kinds of things that get thrown around in that kind of situation. But we persisted and in the end a friend of mine who's a pediatrician said to me you've gotta get a second opinion.

My data suggest that many complex factors play into the balance of professional and parental expertise as care for genetically-diagnosed children progresses. One factor is the *timing* of when the diagnosis is made. Is the child not yet born? A newborn? An older child? An eldest child? A younger sibling? Another is the *health* of the child. At the time of the diagnosis, is she asymptomatic, or has she already been showing signs of the disease? If she is symptomatic, has she been very ill or just bothered by one or two

problems? How long has she had symptoms? A third factor is how involved and knowledgeable parents are with respect to the diagnostic process. Did they already know something might be wrong and agree that testing was needed to find out what, or was the screen a surprise to them, a message from the professional world that they may not even know what dangers lurk for their newborn? Although more in-depth study than what I have done here would be required to tease these multiple strands apart authoritatively, parents I spoke to appeared more reliant on professional authority during the period after diagnosis when that diagnosis was made early, and/or for a first child, and/or without their knowledge or consent. They described themselves as less reliant on providers in the post-testing period if their child was older at the time of diagnosis, and/or if the child was a younger sibling, and/or if the diagnosis was the answer to an empirical question about the child's health that the parent herself had raised.

That parents in my study with a later diagnosis were more comfortable acting as advocates for their children from the start of their CF saga by no means implies that these parents had a qualitatively *better* diagnostic experience. In fact, the majority of those with a later diagnosis (seven out of eleven) illustrate in vivid, sometimes horrific detail how painful the "diagnostic odyssey" can be. Parents described to me -- with all their rage and frustration at having been belittled and disbelieved -- how infuriating it was to have their fears dismissed by professionals, to be told that they are hysterical, uptight, inappropriately "worried about every little thing." Other researchers have concluded that later diagnosis causes significant parental anxiety; increases the chances that parents will consult a lawyer regarding health-care concerns; and leads to "a growing sense of fear that their child's deteriorating health [is] the result of their own incompetence" (Boland

and Thompson 1990, 1243; Clayton 1992a; Waisbren et al. 2003; CDC 2004). My research certainly corroborates the finding that late diagnosis causes substantial distress, but my data suggest a somewhat different picture of the longer-range impact of distressing experiences with health professionals. At least in retrospect, parents in my study with a later diagnosis felt primarily a combination of outrage at how they were treated by professionals, and regret that they were not able to assert themselves more effectively on behalf of their child. As Annie put it:

> I am angry about [the delayed diagnosis] to this day, like in my, in myself, that nobody would listen to me. Doing all these things to him, [other tests and treatments] that were totally unnecessary. I mean, I've carried that anger inside of me for a long time because I was like, I knew from the day he was born. You know how frustrating that is? So flipping frustrating. Because I knew from the day he was born and for seven years, and I was intimidated by doctors, you know, and it was like okay the doctor said no [to testing for CF], so we're not gonna do it…. That day when we found out, I called his pediatrician and I went off on him on the telephone…. I had him on that phone for about an hour and a half. I called him every name in the book, and I said, 'You know you didn't listen to me. I told you. Mother's instinct knows.'

Other mothers also describe how demeaning and infuriating it was to be dismissed and pathologized by the very professionals to whom they were looking for help.

> My sister is the head nurse at a hospital and she had gotten us an appointment with this very supposedly renowned pulmonologist. And he examined us and we felt like we were being blown off. [He] jammed a letter in her file and… I was reading it through, which I like to do. I was reading the letter and it said 'Doctor observed child, parents are hypochondriacs.' As in, it's all the parents, there's nothing wrong with the child. I just remember reading that and you know it's something. [Later, my sister] said to him 'you know, you were quick to judge and tell them that they were hypochondriacs…. Well let me tell you something, thanks to you they have two other children [with CF].'

> I'd been trying to tell them so long that there's something else [besides what was already diagnosed] that is wrong. And… I'm not just a paranoid mom, a first-

> time mom, you know it felt like that was the way I was treated, especially with my oldest one…. That I'm over-reactive, and not to worry and that kind of thing.
>
> At the doctor's office I… would cry every time because he wasn't gaining [weight]. I think they kind of looked at me like this hysterical first-time mother, and the doctor whom I kept going to see kept saying 'Oh he'll kick in, some babies take a while to kick in.'... That was really hard, being so powerless…. Like if I had to do it over again I would stand on my head in the doctor's office until they did a test. Even if I didn't know what it was called. I'd be like 'you have to do something. I can not bring this child in here anymore.' Like I would have ranted, I would have raved. I would have done something or changed doctors.
>
> I think there were notes all over our chart that we were just overly worried. [The doctor] was convinced it was all reflux and all reflux-related…. I had actually found a site where a mom had said you should get your child tested for cystic fibrosis if they have smelly, abnormal diapers, which I thought he did. But again, we had shown them to the doctor at times when we were in the office, and she said they were normal. But they didn't seem normal to me, because I didn't know any other parents who said that their infants had smelly diapers before starting solid food…. We had been in and out of our doctor's office three times a week with congestion and kept being told it was normal baby congestion and it just didn't seem right to us….

So in some cases, the confidence and advocacy skills parents have developed by the time of diagnosis were forged in the course of earlier traumas with medical professionals. Yes, these parents have already learned to be advocates, but some of them have done so under the worst of circumstances, i.e., because their child's health and their own poor treatment at the hands of professionals demanded it. I do not mean to assert that "empowerment" by this route is *preferable* to ceding substantial control to providers after an early diagnosis, to feeling that CF puts professionals in the front seat and parents in the back when it comes to knowing what's best for the child. The point of my analysis here, as in the dissertation as a whole, is *not* to compare these two models and determine which is the lesser of two evils. Nor, as I suggested in Chapter Three, should debates

about newborn screening be defined by an apparent choice between the “diagnostic odyssey” and mandatory newborn screening as currently organized and delivered.

What is useful about the two kinds of parents’ narratives explored above may in fact have more to do with what they have in common than with how they differ. Each demonstrates the problematic nature of power dynamics between parents and professionals as diagnoses are negotiated and produced. In both scenarios, the experience of parents is delegitimized or de-emphasized as a guide for action once a medical definition of the situation -- whether of genetic abnormality in the child, or hysteria and incompetence in the mother -- has become dominant. In one case, it is the mother's observation of manifest illness that is devalued and pathologized in the absence of any scientific verification of "real" disease. In the other, a system of uniform testing is used in a way that devalues parents’ nascent knowledge and authority by standardizing care regimens for genetically-diagnosed children -- regardless of the true genotype/phenotype correlation for that specific child; regardless of the real health benefits of knowing about the genetic disorder in the period immediately following birth; and regardless of whether medical treatments or recommended care routines actually safeguard health. Once the genotype is revealed, parental knowledge recedes, replaced with a medically-prescribed regime of watching, vigilance and preventive care.

The dangers and costs of the first case – i.e., of delayed diagnosis -- are significant, as the mothers quoted above demonstrate. They are also comparatively well documented (see for example Southern 2004; Waisbren et al. 2003; CDC 2004). But the impact of mandatory genetic testing of newborns on parents’ sense of competence, and on their relationship to professional knowledge, is also significant, and is much less documented.

As Nelkin and Tancredi write in their book on the dangers of diagnostics, "testing can transform doctor-patient relationships." My data suggest that in the case of newborn screening, one effect is more reliance by parents on professionals. Nelkin and Tancredi's conclusion that increased focus on diagnostics means "medical professionals are relying more on test results than on the symptoms of the individual" also appears to hold for many of the families with early diagnoses whom I interviewed. Identification of CF sets off an immediate sequence of care practices and early interventions that are overwhelming to the parent. They leave wondering, as Joan did, how they can possibly "do all this stuff in one day." And yet just as there are standardized procedures for giving the newborn screen result in each state (see Chapter Three), so too are there standardized processes for handling a newly-diagnosed infant and getting her "into care," regardless of whether she is symptomatic and in need of immediate intervention or not. Thus, as Nelkin and Tancredi observed with respect to diagnostic testing more generally, "the more physicians rely on tests, the less involved they become with the patient as a person, and the more they distance themselves from the person, the less they are able to assess the implications of professional advice for the life of the individual involved" (1994, 66). Further study of the implications of professional advice for the families of newly-diagnosed newborns certainly seems warranted.

Looking for the "Positive Stuff "or the "Dirty Truth?": Learning About CF After the Diagnosis

How information about CF flows between parents and providers is a critical aspect of the post-diagnosis period. Professionals are presumed to possess, or to have facilitated access to, medical and scientific knowledge about the disease.[60] Parents view providers as a crucial resource in this regard, and -- as I discussed in Chapter Three -- the difference between their own knowledge base and that of their doctors can cause many kinds of distress and confusion. At the same time, parents have a detailed sense of their own coping mechanisms, their own psychological balance and how it can best be maintained in the post-diagnosis period. This is knowledge providers don't have, and when they try to apply a generic approach to communicating information instead of taking cues from parents about what they want to know and when they want to know it, there is trouble.

Parents do not all want the same amount and kind of information about CF during and immediately after the diagnosis. For some, no matter how they discover their child has CF, it is critical to know absolutely everything there is to know, every detail, fact and statistic, no matter how sobering. "Give me it all; I want to know every possible [detail]," said one parent. Or, as another put it, "I wanted to know everything, which I'm still that way. I'm like a sponge when it comes to CF stuff.… I was reading all this literature which was pretty heavy to read when you have a brand new baby." Information about average life expectancy for CF was particularly "hard to swallow" for this mother, as for most parents in my study, yet she wanted to know even that from the get-go. A third parent described her thirst for medical details and statistical information like this.

[60] As I note in Chapter Three, it is not always actually the case that professionals – especially primary care practitioners -- have substantial knowledge about genetic disorders. However, even a poorly-informed provider likely has more information about CF than a parent who has just received an unexpected diagnosis.

> I'm the kind of person, I'm really proactive, so if I find out about a problem or an issue I want to dive into it and figure out what's the best way to do this, or what should we do? So I want all the information I can get…. I don't just want to be clueless and think 'Oh she'll be fine, she'll beat the odds.' I want to know the dirty truth. I want to know what these people [with CF] go through so that I know how I can prepare myself and how I can prepare Alexandra.

For these parents, it is infuriating when professionals try to "shield" or "protect" them by withholding information or meting it out slowly. As Andrea put it, "One problem I have with some doctors is that they talk down to you and don't explain things thoroughly." This form of condescension breeds mistrust of providers, as illustrated by Kim's description of her communication with professionals at Sarah's CF clinic not long after the diagnosis. When Sarah cultured positive for pseudomonas, a bacterium that can attack the lungs of people with CF because of chronic airway inflammation and lowered pulmonary defenses (Moskowitz et al. 2005, 9), her providers tried to "manage" her reaction in the most infuriating way.

> I'm like 'what is this pseudomonas?' 'I will have Frances call you back in a couple of days,' the nurse said. And I'm like 'What?' Well, apparently they needed to give me time to let this sink into my head. So of course I call for two days straight and then Frances called to see how I was handling it and to tell me all about it, this thing that could kill my child. If you don't catch it soon enough it colonizes and causes lung damage and gets worse and worse and pretty soon you're needing a [lung] transplant. Or the child is needing a transplant because you know all her lungs collapse…. Well thankfully they got the pseudomonas in time so it didn't colonize. But this is one of the things where they wait to tell you about…. and you're just horrified that this is happening to your child and they never even warned you. So of course, in my moment of, I don't know if I should say the word grief, but whatever, I kind of scolded this lady. Saying, 'What else could I expect? Tell me so I can prepare, that this might happen to my child. Because I need to know. So that I can prepare myself, so that I can prepare myself so that I'm not devastated like I am right now.'

For Lorraine, medical paternalism manifested most grievously as she was trying to get her healthy older child tested for CF immediately after her baby daughter was diagnosed. At first, the doctor wouldn't do the test.

> [H]e was entirely dismissive of whether or not my son would be affected. He actually refused to test him. He said, 'We're gonna want to focus on your daughter, you can't handle having two right now. So we're not even going to look at him.' And my husband and I were very concerned that if my son did have it then we wanted him to start treatment immediately `cause it had been two-and-a-half years and he hadn't gotten anything. We were just very concerned and he took one look at him and said 'I can look at him and see how big he is and know he doesn't have CF. We're not worrying about him.'

Lorraine and her husband persisted, and the test was finally ordered. When the positive result came in, the doctor said "'Oh, I knew all along that he had it, but I didn't [think] you could handle it…'" Lorraine continues:

> My husband and I are just not accustomed to being treated like we're stupid so that was just infuriating beyond belief…. [W]hen parents are being confronted with a genetic disease and they have a sibling, they are concerned about both kids. And so I think that to try to shield them from the diagnosis is just really patronizing. If a parent were to say to the doctor, 'I can't handle it. Please don't test the other child, let's deal with them one at a time, let's deal with the sick child first,' that's fine. But we weren't saying that. Everybody's different. I mean we're people who just wanta, let's get this straight so we can start working on it.

Lorraine echoes here what one of the parents interviewed by Taner-Leff and Walizer said when providers attempted to shield him from information about his children. "'In keeping the 'bad news' from us they are also exercising power over us and the circumstances that we find ourselves in. This is unacceptable to us" (1992, 77).

Lorraine is absolutely correct when she imagines that other parents may pace themselves differently than she does when it comes to learning about CF and what it

means for one's own family. A "one day at a time" approach is much more comfortable for some, particularly for some mothers. Although not all mothers prefer incremental learning to total immersion, parents in my study did suggest that fathers are almost always interested in the "hard facts" if they are involved in the information-gathering process at all.[61] As Anthony put it:

> I know from my wife… she will often want to sort of screen as much information as she needs, and all the other details, the real nitty-gritty details, she doesn't need to know. `Cause maybe its more of a negative than a positive. But for me I prefer to have as many details as I can, [though] it was pretty real stuff.

For these mothers, the phased accumulation of information gives them time to assimilate little by little, to get used to general ideas and feel competent to judge their relevance or irrelevance with respect to one's own child. "They had some handouts and things… as far as treatment and dietary concerns, you know," said one mother. But "there was just too much at that time to absorb, so we [would] look at it a little bit [at a time]… there was just too much at that time to absorb." These mothers, like three out of four in Collins et al.'s study of parents' experiences with genetic counseling after the birth of a baby with CF, found themselves struggling with "information overload," altogether "[too] much information to absorb in a short time as well as coming to terms with the diagnosis" (2001, 59).

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[61] This observation should in no way be understood to imply that fathers are more active than mothers in pursuit of information about CF. In my study, the predominant pattern seemed to be quite the opposite. Many mothers talked about their husband "putting blinders on" and just "saying everything is going to be okay" rather than joining her in her reading, research, and web discussions. As Andrea put it, "There's one of two ways you go: complete denial or immerse yourself in it and learn everything you can. And I went one way and my ex-husband went the other. He said that it can't be happening… and I actually found all the information we could…. So I guess it took me, it took me and him about the same amount of time to sit back and go 'oh my god this is really happening,' me because I was so busy trying to make sure [the child] was healthy and he because he was so busy trying to avoid it." However, if fathers <u>are</u> involved in researching and learning about the disease, they tend to want all the "nitty-gritty" details sooner.

For many mothers, what is hardest to absorb, and what they don't want to be forced to confront as a bald, statistical representation of their own child's possible future, are data about life expectancy and descriptions of the "natural history and progression" of the disease. They know, of course, that this information exists, and they may even have encountered it on the internet or elsewhere. But they don't want it held in front of them, don't want it to serve as some sort of absolute foreknowledge of their own children's fate or "crystal ball predictions of what our children won't be able to do" (parent as quoted in Taner-Leff and Walizer 1992, 93). As Suzanne put it:

> We had one doctor… and we walked into the room and she sat down and we sat down and she said, 'Having CF is not a good prognosis.' And I sort of thought 'I don't really need to hear this. I'm well aware of what it does.' I didn't really think that was very thoughtful to say to someone while holding their new baby…. This one doctor… could be quite callous. And not really think about how you might be feeling as a parent… [What I needed was] the basics for the moment. You can find out everything else as you go along. It's not necessary to know everything right from the start.

And from Judy,

> I think I at first I don't wanna know everything, just wanna deal with things as they come…they threw everything at me and I wasn't ready for that… I didn't want to know the bad things, I just wanted to know as of today what would happen, today…. I don't wanna know you know the lungs are expanding and you know… the organs, all the organs, it's gonna affect all the organs eventually. I didn't want to know all that. I didn't wanna know the life expectancy of it. I just wanted to know what we were dealing with today.

But Judy's CF center -- like many centers -- has a policy of sitting down with parents and giving them an overall education, a comprehensive overview of CF and all they can expect.

> [T]hey gave me pamphlets and and all kinds of stuff you know, a big book about [CF, they] gave me that book and I guess I needed to know those things but I wasn't ready for those things…. It was too much to handle at the time…. I just kind-of sat there and I didn't know what to expect…. I just thought that they were gonna give us medicine and you know go home… I wasn't expecting it. I guess I didn't realize how much there was to it really.

Parents who pace themselves in learning about the disease are not in unhealthy "denial," nor are they looking for a paternalistic model of care which presumes that providers have an obligation to protect parents by deciding what information to give and what to withhold. They agree wholeheartedly with Lorraine that it should be the *parents* who determine how information is shared, not the providers. What they want is for professionals to check in with them about the education process, to *ask* them how much they want to know and when they want to know it rather than adopting a "one-size-fits-all" approach. Francesca's description of her own needs post-diagnosis, and then of how extraordinarily well they were met by her providers, says it all.

> We wanted to know, where's the positive stuff? We don't want to know all the negatives right away we just want to… hear stories about people who are living with this, who are doing good with it…. We try to really focus on some of the positive information about CF… I don't even think at first we even asked about life expectancies and stuff like that, or hospitalizations… we just wanted to know who do you know who is living it, making it just part of their life, it's not controlling them and stuff like that….

Unlike Kim's providers, and Suzanne's, the professionals working with Francesca in the post-diagnosis period knew how to take the lead from her, responding to where she was at each point and remaining both respectful and available.

> We would just ask them, 'What about this?' And they would come back with stuff. Or we would say, 'We're really getting stressed out, there's a lot going on

> right now.' And they'd be like, 'Okay, you know what, you let us know when [you're ready] and we'll follow back up with you… and see how you're doing….' So they were really good at listening to us, and hearing about what we wanted. They just cared about how we wanted to deal with this, they never told us like how we should deal with it or what to do next or anything.

Several other parents also recounted stories of health-care providers' competence and kindness over the course of our interviews. These other anecdotes, however, had more to do with a particular professional going out of his or her way to show concern and empathy than with a group of professionals being so perfectly in sync with how parents want to pace their learning about CF. Catherine, for example, says:

> "The doctor we had seen who took us to the emergency room [the day they first suspected CF was the problem], who is still my doctor today, he left at 11:00 at night after… admitting us and said, 'Mrs. X., you haven't had anything to eat all night, what can I get for you?' At 11:00, and this man had a family. It was 5:00 when I [first called him that day] and he was ready to [go] home. And [he] stayed with me until 11 o'clock. I mean he was wonderful. So I'll never forget that."

Similarly, Shannon notes with deep gratitude how her genetic counselor, who is "just such a sweet, smiley, happy woman who explains everything on a very good level," called her with test results on a Sunday. "That doesn't surprise me of her," she went on, "that she would take time out of her own schedule, `cause… I know she has called me from home…."[62]

In my study, the variation in how parents wanted to become educated about the details of CF did not appear connected to when and how the diagnosis was made. Other

[62] The data I have about these "good matches" between parents and professionals are too partial for any sort of pattern to emerge, or for analysis in this dissertation beyond this footnote and an affirmation of Taner-Leff and Walizer's assertion that "Kind, nurturing acts [by professionals can] become powerful therapeutic tools" (1992, 83). However, more rigorous study of *why* things work well between professionals and parents when they do work well would be an important component of a future study.

researchers who have conducted qualitative interviews with parents of children with disabilities have also documented a range of preferences on this issue, but have not suggested any particular pattern based on the timing of the diagnosis (Hill 1994; Taner-Leff and Walizer 1992). However, the rapid expansion of newborn screening programs that is now underway means more and more parents will be contacted by providers in the period right after birth with a diagnosis of some kind. At the same time as the sheer number of parents having this experience is increasing, so too the range of actual phenotypes that children with identified "disease" will express is continually expanding -- as it already has with testing for the highly variable condition of CF. In order to meet parents' need to control the flow of information, to get as much or as little as is right for them at any given point, newborn-screening systems and the follow-up services with which they articulate would have to become highly individualized. Providers would have to spend substantial time with each family -- including all those who turn out to have had false-positive screening results, since there is no way of knowing at the beginning of the testing process who is in for an unpleasant false alarm and whose life will be radically altered forever by the experience. They would have to learn – as did Francesca's health-care providers -- to "find out how parents actually feel by listening and not project how they would feel onto the parents." (Taner-Leff and Walizer 1992, 43) Given the haphazard nature of newborn screening at the present, and the population-level rather than personal-level orientation of these large, bureaucratic public- health programs, it's difficult to imagine systematic efforts in this direction will be a reality any time soon.

Parenting "Up to Code": Professionals Define the Standards

In Chapter Four, I examined the influence of a CF diagnosis on parenting practices. That discussion showed how knowledge of the CF genotype increased parental vigilance, and led parents -- particularly those with an early diagnosis -- to focus extensive energy on preventing illness in their diagnosed child. Of course the drive to keep the child healthy comes from the parents, from their fierce love, protectiveness, and determination to hold CF at bay. But the tools and strategies for accomplishing this work come to parents largely from the world of professionals, as do (to an appreciable extent) the standards for measuring its success.

The expertise parents of sick children develop is nothing short of awesome. Parents whose children are actively struggling with the disease in one way or another are able to pick up on symptoms immediately and take action: as one mother put it, "I am a firm believer [in]… 'listen to those parents because they know their child better than you do!'" Parents know when the child needs to be seen by a professional, and when it's okay to keep her at home. They suggest to providers what medications should be tried, and adjust levels themselves when necessary. At times, as Andrea describes here, they seem to have a miraculous sixth sense.

> [W]ith Bobby, right before he comes down with the respiratory infection, when he was younger his breath would smell sweet, almost syrupy. Now it smells musty. When I smell that I start right away with the expectorant and I'll call his nurse and say, 'Right, I'm adding a couple more albuturol treatments for the next few days because he's starting to smell like he's coming down with an infection.' When I first told her that… he was about 18 months old. I took him in for one of his regular visits and she said, 'How's he doing?' I said, 'His breath smells sweet, he's coming down with an infection.' And she just, she was taken aback by that. I

> don’t know how many other parents recognize that as a symptom. But I can recognize that now.

Some parents exult in their expertise, and a few even playfully lord it over their health-care providers. Erica, for example, sometimes likes to "test the doctors." "That's always interesting," she says. "Find out what they would prescribe… you know, would it provide coverage against this or that? And they [the doctors] look at you!" Others talk with pride about the providers’ acknowledgment of their skills. "I have a good enough rapport with my pediatrician," describes one mother. "He says… 'What do you think, you know him [your son] better than I do.'" Another mother's doctor told her she "could probably diagnose a child before he could!"

These anecdotes illustrate that on one level, power relations between professionals and parents have fluidity. Providers begin with the stacked hand, no question. Diagnosticians and treatment specialists alike have power over parents because of the authority afforded them by their professional status; because they have formal training and codified knowledge that most parents lack; because it is their role to "see [families] as having problems and needing to be fixed" (Crane 2000, 3; Litt 2000; Conrad and Schneider 1985). Yet the messy, difficult, rewarding work of nurturing sick children day to day brings parents an expertise all their own, and sometimes a good deal of decision-making authority along with it. A few parents described their relationship with providers now, a long while after diagnosis, as a joint venture on behalf of the child, a satisfying collaboration that's an impressively far cry from the "you guys are the doctors" stance they had at the beginning. As Meredith puts it, "I feel very fortunate to have that outstanding of a team, working with me…. And I have to say that… you really have to

work as a team, because you know the feedback they get from us and that we get from them is so vital to his [my son's] success."

Parents also learn over time to rely on one another, to tap into sources of parental expertise that find expression outside the medical context altogether. Although (as mentioned above) it is very difficult for parents whose children have CF to see one another in person, the internet has made it possible for many to connect from their homes across the country. List serves and on-line support groups were described by several mothers in my study as important sources of information and emotional sustenance. Here, parents share tips, experiences, worries, joys, and strategies for coping with medical practitioners. Lorraine notes that she only got accurate information about CF, in the post-diagnosis period, "when I went to an internet web page that hooked me up to other parents," and Paige describes "get[ting] on an internet support group" as one of her main "way[s] of fighting the disease." Others describe getting the "inside scoop" on new CF medications from other parents on line, or sharing the ups and downs of a new pregnancy – with all its attendant worry about whether the baby will be affected – with the internet group.[63]

But with respect to the all-important issue of medical care itself, the sharing of power between parents and professionals goes only so far. Despite the ingenuity, courage and aplomb of parents who take charge of their child's care, and who exert their advocacy and their agency in the face of medical authority, structural problems remain. These were highlighted in my interviews in several ways.

[63] My data on this issue are incomplete, since I did not inquire about parent-to-parent support in a systematic way. This too would be a fruitful area for additional research.

One issue is that parents continue to perceive professionals as retaining substantial authority to define situations and to judge them. Even after time has passed, and all concerned may have arrived at an explicit agreement that both parties need to be "part of the CF team," often the balance of power is still weighted towards the side of the professionals. In many parents' accounts, in the language they use, lurks a semi-conscious assumption that professionals retain an expertise that gives them the right to mete out praise rather than themselves having to earn it. Some parents talk about how much they trust or distrust their health-care providers, but many also talk about how much their providers trust them, about how they have earned the confidence of the professional team. "I am the one who tells them every little thing that goes on," says Kim. And then, laughing a little at the irony, but not objecting outright to its implication of who's really in charge, who gets to judge whom, she adds "they [the doctors and nurses] have told me ***they feel safe*** that Sarah is with me. I guess that's good for me. The nurse is… very confident in my care because I give them so many details" (emphasis added).

Suzanne is pleased that over time the doctors have come to have confidence in her, that "when Quinn's been sick, ***they'll trust me***…" to make certain decisions, "you know, ***trust you*** that you know what's wrong with him and that I live with him to know how he is" (emphasis added). She goes on:

> Once you've been in the system for a while… they sort of learn to trust you and to know that you're gonna do the right thing by your child…. I mean I guess they've had kids where the parents haven't treated their children and what not. But when they know that you do, and that you look after them in the right way, they respect [you] for doing that too I think.

As this quote reveals, once the child has a diagnosis of CF, parents' caretaking practices are evaluated against specific standards set by medical and public health professionals. Of course some of these accord seamlessly with what the "common person" would consider humane: taking a child to the hospital in an emergency; administering medicines that obviously alleviate her suffering; not intentionally exposing her to things known to make her sick. But the boundary between excessive social control and the simple protection of children's rights has never been an easy one to draw (Katz 1986; Donzelot 1977; Parton 1994). With state-mandated diagnosis of CF at birth, and the concomitant immersion of parents in a world of preventive treatment regimes regardless of whether the child is symptomatic or not, I believe there is good reason for concern about the impact on families of professional expectations about "the right way" to care for these children.

Consider Francesca's experience with the professionals at her CF specialty center in the months and years after her newborn-screening diagnosis for baby Tess. Beginning on the first day she brought her (asymptomatic) newborn in to the center, she – like all the parents in my study -- was assigned a social worker as part of her treatment team. This is routine, and though Francesca was not looking to any kind of mental health professional for emotional support, she did not object to the intervention *per se*. "I guess the first time I could understand," she says, "`cause they didn't know where we were going with it, or how we were doing." But the visit with the social worker remains a standard part of every clinic appointment. Now that she's an established mother with substantial experience, Francesca feels second-guessed and criticized both by the social worker's very presence and by her "rude and opinionated" manner.

> To me as a parent… I'm not asking for somebody's advice in that area. I'm not you know seeking that out. And you can see my child's growing healthy, she's doing fine. I'm coming in for all these routine visits and everything. And I-I've really taken her care into consideration, it's kind-of almost like I feel insulted that they're there... she comes in and she tells you 'this is what you're doing, this is what you should do, if you're not doing this you need to do this by next [time]'…. I mean I-I do everything for Tess. I-I would give my own life if I could to make her CF go away. There's probably a need for parents you know that maybe don't get it, or aren't taking as good care, but I've never once missed a clinic. I've never once denied tests for Tess. I always do the best for her. I probably take her in more than I have to sometimes `cause I'm paranoid. And then yet the social worker's telling me that it's just maybe not quite good enough yet,… 'You're not doing it this way, so maybe you should try this or that.' Or like she'll talk to me all the time about my parenting skills, with you know treating [Tess' little brother] Gus equally and stuff. [But] he's not lacking in any means. I mean he's developmentally up to code you know, from a physician's standpoint. He's right on track with kids his age… I don't just neglect Gus and take care of Tess, which is how she kind of makes me feel sometimes….

Francesca holds everything together, working unbelievably hard to take care of both her children. Sometimes, the unwanted intervention of the social worker is just too much. Yet she doesn't know if it's possible to say she doesn't want this person's services, or to switch to a different worker. The social worker may just be an inevitable part of the package.

> We're frustrated. This is hard enough as a mom to deal with being a parent of a child with cystic fibrosis but then now I have somebody telling me how to parent…and step[ping] on my feet. Like I said, I can completely understand if one of my kids wasn't meeting health guidelines or developmentally they just didn't seem to be as sharp as they should be. But my kids are both you know tested and go to the dentist and… I mean the stuff that she's concerned about we're doing so it's like why can't you get off my back a little bit?

Francesca's objections have partly to do with the inappropriate judgments of a professional with poor skills: it is infuriating to be found wanting by someone who has no idea what the real situation is. Further, for her it is insulting that the social worker is

there at all, because it implies that Francesca's parenting needs to be monitored. A number of other parents raised similar issues about social workers and "counselors," noting that they resent the presence of these professionals, or – more commonly -- that they simply don't need their help. To the extent, then, that early diagnosis of CF results in a new kind of state intrusion into general parenting styles and psychosocial issues,[64] parents may have a particular bone to pick. Put in sociological terms, they don't want to be subject to paternalistic forms of professional dominance that presume the ignorance or incapacity of the patient, who in this case is not the diagnosed child but the parent herself.

But in my interviews it was not the *existence* of prescribed rules for parenting that I heard parents objecting to: it was the *generic, indiscriminate application* of systems to monitor and judge compliance with those rules. Suzanne understands that it takes the doctors a while to trust that parents will do the right thing because some parents don't "treat their children" according to medical prescription. Francesca acknowledges that not all parents come faithfully to every clinic, proceed with every test, keep their child or children "up to code." It is difficult for the parents in my study to feel sympathy with those who don't follow these rules, since they have come to define their own participation in prescribed methods of preventive care, monitoring, and ongoing testing as the central way they can physically protect their child in the face of the diagnosis. It is part of how parents in the medicalized system set in play by newborn screening enact their love, and I think I would probably do the same. Despite parents' fervent wishes, it's not possible to make their children's CF go away, even by giving up their own lives. So they do what the medical system has taught them to do since their child was born. They

[64] See Katz 1986, Chapter 5 for an excellent review of historical examples of this phenomenon.

follow the advice of the day – the professionals' rules -- as closely as they can. They go to every clinic, show up for every test, and hope for the best.

But the tacit acceptance of professional codes for parental conduct in the face of an early diagnosis -- and indeed the imperative to get an immediate diagnosis in the first place -- has its dangers. Screening for CF at birth is mandatory, and follow-up testing will be overseen by child-protective services if necessary. Then – regardless of whether the child is symptomatic or not, and regardless of whether available medical care is widely accepted as effective in mitigating the disease or not -- treatment regimes of some sort will be prescribed, with parents again accountable not just for showing up, but for making sure children "meet health guidelines." These policies and practices can be seen as examples of a domineering state, plain and simple: as noted earlier, parents' and citizens' rights to informed consent and freedom from coercion are at stake.

Moreover, mandatory early diagnosis and immediate preventive treatment for genetic conditions that cannot be cured are examples of "a more general deployment of expert knowledge for shaping thoughts and actions of subjects in order to make them more useful and 'governable.'" (Petersen and Lupton 1996, 15) As Petersen and Lupton argue, drawing on the work of Foucault (1973; 1977; 1980), the "new public health" is a little-explored yet powerful arena where the technologies of government work at a distance, an example of how "power operates most effectively when subjects actively participate in the process of governance" (Ibid., 11). I am not suggesting a conspiracy theory of any kind here; nobody is setting out to dupe parents, and of course newborn screening has saved and continues to save much suffering and many lives. Yet mandatory diagnosis of CF at birth, and the subsequent prescribed regime of clinic visits, follow-up tests, and

preventive practices, have also undeniably created a new "code" for parents to follow. A new set of expectations, discourses, and regulations now govern the parenting process in these families. That parents accept the code, embrace it even, is not surprising: it is very difficult to do otherwise once the abnormal screening result is reported and its corollary sequence of events ensues. But the automatic testing of infants at birth does more than "save the babies." It also provides new "norms by which individuals are monitored and classified, and against which individuals may be measured" (Ibid., 12). This result of newborn screening may be an "unintended consequence" (Merton 1936) of a policy intended primarily to improve the health of infants, protect the economic interests of the state, and influence future reproductive decisions. Nonetheless, it is a reality, and one that I predict will exert its influence over a growing number of families as testing rapidly expands for CF and other conditions with late onset and/or poor genotype/phenotype correlations.

In closing here, I want to turn again briefly to a subgroup of parents I talked about in Chapter Four: those whose children tested positive for CF at or before birth and are still asymptomatic. Of course, these are the lucky ones in terms of the most important factor: their children's health. But they may also be the ones who struggle most in their relationships with professionals, and who have the most difficulty developing confidence in their own ability to manage the child's CF "up to code." Parents accumulate their impressive knowledge about CF as they encounter symptoms and learn to cope with them, and they develop their advocacy skills as they assert this expertise in health-care settings. When the child still doesn't have symptoms, parents may instead remain perpetually nervous. As Kate describes the experience of parents she knows, "My

daughter's not showing any symptoms, and I'm wondering if she's okay, and I feel like I should be doing something." These families -- like the parents whose children are symptomatic -- have entered a world of intensified vigilance. They have been "urged to turn the medical gaze" upon their children, and to "engage in such technologies… as monitoring… bodies and health states and taking preventive action in accordance with medical and public health directives." (Lupton 2000, 57). Empirically, the parents observe good health in their child, yet the doctors' report of a genetic abnormality, a lurking disease, can in some ways trump the "mere" evidence of the senses, leaving parents permanently watchful and unsure -- deferring to the knowledge of professionals.

Chapter Six

Conclusion: Putting Newborn Screening to the Test

It is October, 2005, and earlier today I had a conversation about newborn screening with a colleague who works in public health. He was recounting to me what just occurred at the September 2005 national meeting of state newborn-screening directors. "HRSA[65] has decided to do away with the requirement that conditions tested for by newborn screening be treatable," he said. "They will be releasing a Request for Applications from the states shortly, looking for pilot programs that will test for a hundred or a hundred and fifty conditions -- and they don't have to be treatable. Genetically diagnosed children will be put in state registries and followed by researchers over time, and we'll learn that way about their conditions and how to treat them." And then he said, "The parents have spoken, and the government has listened. Parents want to know the diagnosis no matter what."

"Which parents have spoken?" I wanted to ask. It is very unlikely that the "parents" he was referring to so generically are those who are, presumably, in the best position to know whether this policy change is a good thing -- i.e., parents who have received newborn screening results that were unbidden: results that were ambiguous in meaning, or that diagnose conditions with late onset and/or little or no effective treatment. No study of these parents' views on newborn screening exists, to my knowledge. Most likely, my colleague was referring instead to a small number of very vocal individuals who exert enormous influence over the policy process by going public with their story of a sick child and of how that child -- or perhaps that child's younger sibling, conceived

[65] The Health Resources and Services Administration.

before the first child's condition was diagnosed -- could have been saved or spared enormous suffering by the addition of one more condition to the newborn-screening panel.

Many of these tales are indeed heart-wrenching, and the pleas of those who live them should not be dismissed lightly. But, as I have shown in the preceding chapters, such dramatic cases are not the only diagnostic experiences that matter: parents actually have complex feelings about the "cursed blessing of newborn screening" (Rosner 2004, 21). It is true my study corroborates other research that suggests parents support newborn screening as a matter of policy – primarily, as I discuss below, because they think it improves health outcomes for children, and also because they believe that without efficient diagnosis at birth a medical odyssey will inevitably ensue. However, parents I interviewed also spoke eloquently about what is lost when the diagnosis comes so very early, or – in the case of later diagnosis – what would have been lost had the diagnosis come much earlier. They spoke about how sustaining even a brief time of "blissful ignorance" during the child's infancy can be; about how overwhelming it is to care for and get to know a newborn and learn about his or her disease all at the same time; about how impossible it is to separate the diagnosis from the child's identity when knowledge of the disease comes so soon; about how much parental power and control tend to be ceded to health-care providers when the parent is taken unawares by diagnosis of a completely unsuspected genetic disorder. The parents I interviewed spoke also by their actions, by electing *not* to have their second or third babies, who were known to be at risk for CF, diagnosed at birth -- despite their verbal support of newborn-screening policy in the abstract.

I am not, therefore, convinced that "the parents," in all their diversity of experience and opinion, have in fact spoken. I am not convinced that the necessary research has been done to elicit the stories and perspectives of the majority of those who have been and will be affected by screening – people who have not "gone public" with a newsworthy narrative, yet whose lives are radically altered by an unsolicited early diagnosis. I am not convinced that the full implications of testing newborns even for conditions where there *are* potential health benefits for the child have been adequately explored, and I am even less convinced that we should nonetheless continue to extend screening programs to include conditions where there is clearly no such benefit.

"Testing Baby" is, I believe, the first study to ask in-depth questions about how newborn-screening programs "actually feel to those they touch" (Clayton, 1992b, 94). It is almost certainly the first to give substantial narrative voice to certain dimensions of that experience: the impact these programs can have on parenting, on relationships among family members, on relationships between parents and their children's health-care providers. Further, it focuses attention on genetically-diagnosed children and their parents – uneasily awaiting the arrival of predicted symptoms -- as members of Frank's "remission society." The textured knowledge gleaned in my study is, I would argue, valuable in itself as a form of testimony about what it means to parent children in a context increasingly defined by genetic screening, genetic explanations, genetic medicine. However, important as it is to provide a platform for narratives about "parenting in the genomic age," I hope that the initial research I have presented here can do more: I hope it can be used to broaden the terms of debate over newborn-screening policy; to reconsider important issues with respect to *how* newborn screening is

implemented and practiced; and to build a more comprehensive research agenda for the future. I turn now to a brief consideration of each of these, and – finally – of the contribution I hope my study can make to the tradition of critical sociology to which it is so indebted.

Policy Implications of Testing Baby

New technology and new scientific capabilities often drive policy development – most often, in the process, curtailing public dialogue and precluding robust consideration of complicated social issues. Such was the case with prenatal testing a generation ago when amniocentesis became routinely available, and such is the case right now, in the year 2005, for newborn screening. Tandem mass spectrometry, combined with the detailed knowledge about chromosomal abnormalities that was amassed in mapping the human genome, have made it easy and inexpensive to screen for ever-larger numbers of genetic conditions and markers. Expansion of screening programs, which has proceeded steadily but slowly since the 1960s, has suddenly become precipitous. As is so often the case in such situations, the criteria used by decision makers (in this case governors, state legislators, and newborn-screening advisory group members) for adding conditions to screening panels lack the complexity required for a policy issue with such vast implications.

My study is an investigation of important under-examined aspects of newborn screening that it seems to me policy-makers and the public at large should consider with care as decisions are made about screening for more and ever more conditions. The manifest function of newborn-screening policy at this time is still to prevent disease or,

when that's not possible, to lessen its effect. But my interviews with parents highlight many other consequences of testing, including its substantial influence over the entire context of early parenting, over intra-familial relationships, and over the balance of power and the process of collaboration between parents and their health-care providers.

In a 2004 special issue of the Morbidity and Mortality Weekly Report devoted to newborn screening for cystic fibrosis, the authors describe in detail the National Center for Disease Control's (CDC's) criteria for evaluating the potential risks and benefits of proposed newborn-screening tests. In addition to an analytic framework for interpreting evidence of "clinical utility," they name four (only four!) potential psychosocial risks worthy of consideration: false negative test results, false positive test results, carrier reporting, and misinformation (Centers for Disease Control 2004). Formidable as these four issues are, my study suggests that the "genetic rendering" of our children right from birth -- the automatic classification and diagnosis of newborn babies -- has even farther-reaching consequences.

Newborn screening diagnoses can and do profoundly alter the experience of new motherhood. Think of Shannon, afraid to fall in love with her new baby during those first days and weeks, or of Betty, who "tried so hard at the very beginning not to love [the baby] very much." Think of the parents whose relationships with family and friends is significantly altered by the predicament of revealing (or not revealing) the diagnosis of a seemingly healthy child during just those difficult weeks after birth -- when community support is perhaps most important, but when it also feels most risky and problematic to "go public" with a diagnosis. Think of Kim and Paige and Andrea and others, trying to build bubbles around their diagnosed but not-yet-symptomatic babies, "just wanting them

to be normal" but "analyzing everything" as they wait for the first symptom to appear, while simultaneously striving mightily to forestall it. And think how profoundly medical experts influenced the parenting process for all my interviewees who received an early diagnosis -- by carrying out tests without the parents' permission, and often without their knowledge, and by then informing parents about a genetic abnormality in their apparently-healthy newborn. Think of how the medical regimes and monitoring processes instituted after a newborn-screening diagnosis created for Francesca, Suzanne and others a whole new set of expectations – externally imposed, but soon internalized -- about what it means to "parent up to code" and to "get with the program."

Parents in my study were absolutely emphatic in declaring that nothing is more important than their child's physical health; that any other form of suffering or loss pales in comparison. They favor newborn screening, but they do so, as I have said, largely because they believe it is the only realistic alternative to a painful and damaging diagnostic odyssey, and because they have been told that starting a preventive regimen at birth – not at one month, or at three, but *immediately* – is the best way to preserve their child's health. When the question of whether to test or not to test seems to be a choice between getting a diagnosis before symptoms or being dismissed and pathologized by health-care providers over an extended period during which your child is getting sicker, of course the decision will be to support early testing. And when it seems to be a choice between preserving some months or years of blissful ignorance before the diagnosis and giving your child the best, most prompt form of preventive care, again the decision will inevitably be made in favor of more tests.

But there are other ways to frame questions about when and how genetic tests should be offered. Newborn screening is appealing to policy-makers and public health officials, as I discussed in Chapter One, because it is efficient to administer (i.e., newborns are a captive testing population during their post-birth hospitalization); because it yields significant epidemiological data and prospective-research opportunities; and because it allows the state to give information to parents – all parents, even those who did not and would not seek it out – that will "assist" them in future "reproductive planning." Yet there are many conditions now screened for, or proposed to be screened for, on newborn panels for which there are *not* convincing health-related reasons why screening must be mandatory, and why it must be conducted immediately -- why disclosure of this life-altering yet often ultimately ambiguous diagnostic information should shatter the vulnerable, precious, difficult period immediately following the birth of a baby. If the health benefit to the infant is not truly undermined by testing at a later point – be it at four weeks, three months, or even later -- instead of before hospital discharge, then genetic screening tests could be *offered* (rather than mandated) as part of routine pediatric care during infancy rather than mandated at birth.

Thirteen years ago, when newborn screening panels were much more limited and when the health benefits of screening stood much more firmly as a necessary criterion for testing, Clayton noted:

> We simply do not know whether the decisions that are made about newborn screening in a political/administrative system and the potential ability of state-run programs to ensure more uniform testing and follow-up actually lead to better results for children than would occur were newborn screening simply another aspect of routine medical practice. If the answer is no, then it is difficult to justify treating newborn screening so differently from other aspects of medicine (Clayton, 1992a, 644).

Today, the answer to Clayton's question appears to be "no," at least for some or many screened or potentially-screened conditions. And it *is*, I would argue, increasingly "difficult to justify" expansion of the new terrain newborn screening has pioneered in American medicine: state-mandated diagnosis of non-infectious disease.

If a child is symptomatic as a newborn, there can be no question of separating the moment of diagnosis from the post-partum period; the condition announces itself, and must be confronted immediately. But we *do* have the option of separating these two events for asymptomatic children with non-emergent conditions – and these may already be, or may soon become, a majority of those receiving true-positive newborn screens. As I have shown, unexpected and unsolicited genetic information has enormous implications for families. When the diagnosis of an asymptomatic infant is made immediately after birth, that diagnosis – not the disease, but the diagnosis itself -- significantly defines the early relationship between parents and their newborns, coming as it does when they have "no other dimensions of [the baby's] identity to counterbalance this early signifier of disability" (Rosner 2004, 20).

There has been no public outcry about the violence done to the newborn period by imposing an immediate diagnosis on it, and policy-makers have thus not been forced to make it an issue of central concern. But my study suggests that the lack of public discussion on this issue may be largely because nobody has ever *asked* parents to describe in detail the impact newborn screening has had on their lives. Research has focused instead on whether health outcomes are improved, and – less frequently – on the impact of false-positive results or on the identification of genetic carriers. Parents of

babies with true-positive results have been presumed, overall, to be unmitigatedly grateful beneficiaries of this mandatory public-health intervention. And indeed, the parents in my study who got newborn-screening diagnoses *are* grateful. They are grateful to have been spared the infamous diagnostic odyssey, and grateful for the diagnosis at birth because iterative public health and medical messages about the promise of control offered by immediate knowledge of the disorder have made them feel certain that their baby's health would have been at greater risk had the disease gone undetected even a little longer. Remarkably, even Shannon -- though observing (as noted in Chapter Four) that all her "tears and worrying" in the months after the positive newborn screen and diagnosis were for nothing since her daughter Margo appears not to actually have CF -- is grateful that she lives in a state with newborn screening. "Look at all those people who would never have been tested who are finding out, who have a much harsher case," she said. "You know, if you're gonna test, there's always gonna be someone on the low end of the spectrum... somebody's gotta be the guinea pig and it just happens to be us…."

Nonetheless, my interviews revealed that many also feel (or, in cases of later diagnosis, would have felt) robbed of that irreplaceable time when "everything was so perfect" and they remained temporarily "unravaged by the news." As was the case for Jennifer Rosner, these mothers find that "[t]he excitement of having a new baby was entirely eclipsed" by the diagnosis, and that "many parental instincts were stunted" (2004, 20). Without a serious discussion of how optional genetic screening might be incorporated into pediatric care rather than mandated at birth, and without rigorous research about whether later diagnosis for each screened or potentially-screened

condition might compromise an infant's health, we should not accept this loss as an unavoidable consequence of doing well by our children's health. Similarly, the inevitable objection to moving screening tests from the hospital to the primary-care setting -- that such a transition is just not practical -- should not set the terms of the debate. Where there's political will, and where the overall well-being of the family truly drives the development of health policy, there can certainly be a way.

Incorporating genetic screening into pediatric primary care would also create a much more appropriate opportunity for educating parents about the tests, and for obtaining informed consent, than can ever exist in the harum-scarum of the period immediately surrounding childbirth. Of course it might also be possible to integrate education and consent into prenatal-care and newborn-screening programs in the forty-seven states that have not yet done so (see below). But my interview data, combined with other research about the impact of prenatal tests[66], suggest that learning and deciding about such tests is particularly stressful for women during pregnancy and the period immediately following birth. At these times, the child is as yet an enigma, with an identity still "undeclared, unknown, and unknowable" (Ibid.), and the discourse of risk thus exerts a particularly powerful hold. With "little else to go on" in knowing and perceiving the fetus or newborn, the specter of all that might be wrong can loom very large.

But to overcome the difficulty of incorporating meaningful parental decision-making into the newborn-screening process by simply mandating testing *without* parental knowledge or consent is, I believe, unconscionable – a paternalistic, interventionist policy

[66] See for example Katz Rothman, 1986.

that is both legally[67] and morally untenable. Francesca's experience of being told nothing about what the test was or meant, and of consequently blaming *herself* for the positive screen even while wondering if her asymptomatic daughter was in pain, illustrates this point in bold relief. But I find myself reluctant simply to advocate for meaningful informed consent within the existing system as the solution to this problem. The mothers in my study who felt that "too much information" about what was being screened for and why would cause them to be in "a real state" until the results came back must also be respected. The stress caused by being asked to actively choose between taking on this ill-timed burden of worry on the one hand, or "denying" the newborn a potentially useful intervention on the other, would likely be enormous -- just as it has been for many women confronting the "choices" now built in to prenatal care. Moving many or most genetic screening processes to the pediatric setting should at least be considered as one intelligent solution to both problems.

Equally important from a policy perspective is that we step outside the testing paradigm altogether and ask larger questions about how we can best safeguard our children's well-being, and about how we can conduct ourselves sanely in an era characterized by hyper-awareness of personal risks. As I noted in Chapter One, sociologists and other critics have argued persuasively that increased reliance on genetic explanations of human health draws attention away from other significant determinants, particularly those that are "environmental" and therefore more integrally connected to social structure than to biological endowment. "Geneticization," as this process is

[67] See for example Annas, 1982; Downie and Wildeman, 2001. For a larger discussion of ethical considerations in research, see Emanuel et al., 2000.

sometimes called, thus results in the selection of genetic disorders as a critical target for "risk reduction," while at the same time relegating other "risks" – e.g., poverty, racism, social isolation, and poor access to primary health care, to name just a few – to the margins of public consciousness. The risks of failing to know, from birth, what genetic disorders may threaten one's new baby are thus represented within the "new public health" as disastrous, while the risks to the child of, say, living in a family without adequate income or social supports – which have been extensively researched and documented[68] – receive much less attention.

Of course individual health conditions should be diagnosed and treated with the best solutions medicine has to offer. Yet the skewed allocation of resources in 2005 may mean that baby Jill gets a diagnosis at birth for CF, yet, like baby Margo, she will turn out not to have CF-related symptoms of any kind. At the same time, she has no health insurance, no safe outdoor space in which to play and develop, no caregiver with the resources to nurture her well. As we screen for more and more conditions, as technology makes it possible to test for elements of multi-gene conditions and predispositions as well as single-gene mutations, the scenario might easily become even more extreme. We might know that baby Joe has markers that increase the likelihood that he will be an alcoholic, that he will be depressed – and at the same time, we might still have no resources to address the fundamentally social underpinnings of these well-known risks to health, well-being and longevity. Newborn screening programs must be judged, ultimately, not just on their own merits, but also in terms of what we give up when we choose – and it is a choice -- this kind of intervention. It's hard in our historical moment

[68] See for example The David and Lucille Packard Foundation 1997; Brooks-Gunn et al. 1993; Schorr 1997.

to advocate strategies that are low-tech and rely primarily on strengthening social and community contexts, but perhaps it is these strategies, more than the scientific and technological ones we tend to favor, that would ultimately provide greater benefit.

There Must, Must be a Safety Net for Parents: Implications for the *Practice* of Newborn Screening

Above I have discussed some broad perspectives on newborn screening *policy* that emerged from my study. Here, I want to focus more specifically on the implications of my research for newborn-screening *practice*. These programs in their present form, and probably soon in greatly expanded form (despite the cautionary conclusions documented in this dissertation!), will touch the lives of tens of thousands of families each year for the foreseeable future. Because parents gave so generously of their time, and entrusted me with their confidence, I was able to learn a good deal about the experiential dimension of newborn screening. Here, in summary form, are key insights for practice that I took from those conversations. Following the lead of the parents I interviewed, who often addressed themselves to a real or hypothetical health-care provider or administrator when describing what happened to them and what they wished had happened, I have written this section in the form of direct instructions for health-care professionals and administrators who implement prenatal and newborn-screening programs.

- Take the need for **education about newborn screening** seriously, and do this work during prenatal care rather than in the period immediately surrounding birth.

- **Implement the "opt out" clause** in existing state newborn-screening laws conscientiously. Find out what the law is in your state by looking it up on the National Newborn Screening and Genetics Resource Center website (http://genes-r-us.uthscsa.edu/index.htm), and let parents know – as appropriate -- that they have a legal right to refuse screening for any and all conditions.

- **Implement informed-consent protocols, or develop them** in your state. If you practice in one of the three states that requires informed consent prior to newborn screening, make sure you are conscientious about implementing this part of the program, and that the process is a meaningful rather than a *pro forma* one. If you live in one of the other forty-seven states, lobby to develop a pilot informed-consent program where you practice. Remember that the data from Maryland, where informed-consent laws have been in place for more than twenty years, indicate that the process is not overly time-consuming for professionals, but that it is nonetheless helpful for parents in a number of ways.

- **Give parents newborn screening results in person** rather than over the telephone. Being face to face makes it easier for parents to ask more questions, to get more information, and to receive appropriate reassurance. If you do have to tell parents about a positive screening result by telephone, call at a time when you do not need to rush off the line or otherwise make yourself unavailable for questions and follow-up communication. As one mother put it, when parents are told their baby screened positive for a disorder, "… they need to be counseled appropriately… There has to be a person on hand to provide that link [to follow-up testing, information, and services]."

- **Read updated literature** on the genetic condition for which your infant patient has screened positive before you contact parents. They deserve to know what treatments are state-of-the-art, and to be given information and

data that are current. "My pediatrician pulled out… a ten-year-old textbook and started showing me how CF kids die when they are five," said one mother in my study. She added, "That was just wrong, you know?" Don't make this same painful and avoidable mistake.

- **Ask parents how much information they want from you at any given time,** and respect the pace of learning that they set as well as the methods by which they choose to educate themselves.

- **Institute an optional home-visiting program** for families who have just received a newborn-screening diagnosis. Here is one parent's description of why this would be useful, and what the home visitor might do.

> A lot of… parents… find out these results, either at the doctor's office or by phone, and then they go home and they know nothing… They were handed a book that says cystic fibrosis for the patient and family. Now I don't know how they feel, but when my daughter was first diagnosed I wasn't really up to reading a hundred-and-fifty-page book…. I was just a little bit on overload, and what I did read didn't really stick. [What I wanted was] somebody who could, you know, come in my house, sit down with me, look at my baby with me, tell me what would be normal or what would be abnormal… show me, tell me, maybe let me cry, maybe hold my hand, maybe just look in my eyes and hear what I'm saying.

- **Be certain there is a system of care in place for parents receiving the positive screen and diagnosis**. There have to be appropriate resources systematically available to help parents cope with both the long- and short-range implications of the information you are giving them. As one mother in my study implored, "If [newborn] testing is gonna be done, there must, ***must*** be a net in place to catch people…." There was such a net in her state – a system that put her in touch immediately with genetic counselors who held her hand through every step of the process. But she knows that in other states "they don't have anything set up. So if it's positive, what are [they] going to

> tell these people to do?" Another mother emphasized that without good genetic counseling in place as a standard protocol for parents being told about a positive newborn screen, the results "could be devastating."

> The counseling provide[s] information for parents about a particular disorder, but it also provides an avenue for where you go from… the screening results, to the diagnostic testing and results, to connection to whoever they need to be connected to…. You can't just get a letter in the mail saying your kid's screened positive for CF or whatever… That is the scary part. What are you [as a parent] going to do with this information? …. There has to be some protocol… there has to be established [a] qualified person who can explain.

Future Research

The data I have presented in this dissertation clearly point, I believe, to the importance and utility of qualitative research about the unintended consequences of newborn- screening programs. The next step is a larger study interviewing a subset of the population I have included in this first effort – i.e. parents who, like Shannon, Crystal, Lorraine and others, have a child who remains asymptomatic for a period of months or years after the diagnosis was made. This group would include both parents of children who may never display a phenotypic corollary to their genotypic "diagnosis," and those whose phenotype may result in late onset of the diagnosed condition. Research with such parents will hone in more tightly than mine has on what newborn screening means for families whose lives are affected not by a manifest disorder, but by the processes of testing and diagnosis. The World Health Organization reports that, in the case of CF, a recent approach "has been to regard individuals with a… genotype but no discernible clinical disease (e.g. some infants detected by neonatal screening) as 'pre-CF' using the analogy of premalignant conditions" (2000, 3). What does it mean to have a child who is "pre" some genetic disorder diagnosed at birth? What impact does this new form of

medicalization have, over time, on the diagnosed children themselves? In what new ways are we inscribed by the "risk society" as babies' genetic codes are examined with increasing scrutiny, and new labels are invented to describe their genetic status? Since decoupling the actual experience of symptoms from the knowledge of genetic "disease" will no doubt become even more frequent if recent policy trends continue, targeted study of this group of parents could not be more timely.

It would also be fruitful to pursue systematic research about how the voice of parents has been mobilized in newborn-screening policy development. As I have suggested above, one distinct subset of consumer experiences has come to stand for "the parental perspective" as if this perspective were monolithic. A study exploring how this has come about might begin to answer a number of salient questions: How are parents' narratives leveraged on an *ad hoc* basis to influence policy, and how (if at all) do these *ad hoc* processes connect with legislated mandates to include "consumer representatives" in formal policy-making bodies? What role do the media play in representing the interests of parents in policy discussions or debates about newborn screening? How does public-health discourse about newborn screening, as evidenced in health-education materials and public-awareness campaigns, influence the way the public and policy-makers perceive the costs and benefits of newborn screening? What opportunities exist for including a broader and more diverse range of parental perspectives on newborn screening in policy-making arenas?

A third promising area of study would build on what Maynard has described as the sociological "literature on bad news." This literature suggests that the "interactional work of forecasting" – i.e., giving the recipient some warning about bad news before

delivering it – is useful in helping recipients of such news to come to terms with it (1996, 109). My study suggests that parents are significantly affected by *how* they learn about genetic diagnoses. Ethnomethodological research on the microprocesses surrounding communication about newborn screening and its results would constitute a valuable contribution both to the existing literature in this area and to the applied work of developing practice guidelines for health-care providers who regularly give positive screening results to families.

I hope, in collaboration with two colleagues, to design and implement research in these three areas over the next two years.

Sociological Significance of "Testing Baby"

In a recent interview published in the New York Times, sociologist Troy Duster asserted:

> By looking at what's in the blood, [geneticists] avoid the messy stuff that happens when humans interact with each other. It's easier to look inside the body because genes, proteins and SNP patterns are far more measurable than the complex dynamics of society…. But few of these basic processes happen outside a social context…. [Y]ou can't… [do] biomedical research… without also doing some sociology (as quoted in Dreifus 2005).

Duster was referring here to genetic research based on "race," but what I have tried to show in this dissertation is that he might just as well have been talking about newborn screening. When we mandate testing of a newborn's blood, we certainly do so in a social context. Furthermore, the implications of the act are far messier than most people realize: newborn screening is not just an isolated laboratory practice with concomitant life-saving interventions for the lucky few who are diagnosed with a treatable disorder. It is, rather,

a particular form of social practice that emerged at a historical moment when several converging factors favored it: technological innovation; state power as enacted through the modern public-health apparatus; the rise of the genetic paradigm; and a pervasive discourse of risk reduction and early intervention. We cannot continue to evaluate newborn screening, then, only as a medical intervention with quantitatively measured effects on health and longevity. We must also investigate it as a sociological phenomenon that both reflects and influences how individuals interact with each other, how they relate to social institutions, and how they conduct themselves – consciously, and also unconsciously -- within a specific historical and cultural context.

It has been nearly forty years since Berger and Luckmann (1966) challenged sociologists to examine various forms of knowledge as social and historical products, thereby revealing how phenomena that are in fact socially constructed come to be understood as immutable facts: we have by now had considerable practice deconstructing the processes of institutionalization; reification; and of values internalization that coalesce to produce our shared reality. However, using this lens to study social phenomena that have not yet been subjected to it remains an important and valuable activity. The devil – as the saying goes -- is indeed in the details of each specific "social fact" (Durkheim 1938; 1951). I have tried here to study, understand, and explicate some of the "details" inherent in the social fact of testing babies, and I have also outlined further steps for mounting a larger research agenda in this area. I hope that what I have written will interest other sociologists in the topic – perhaps, in particular, those who work on the sociology of health and illness, or on the sociology of families -- and inspire them to focus additional attention on this heretofore under-examined facet of social life.

I hope too that "Testing Baby" can contribute to the ongoing project of building a "public" sociology, dedicated to engaging not just other academics but also the public at large in a rigorous consideration of social issues. This aim was fulfilled in some small way through the research process itself. The narratives that parents gave me of their experiences were not, of course, handed to me ready-made. Rather, their accounts emerged over the course of our interactions during the interviews. In some cases, parents noted that I was asking them questions they had never yet asked themselves. One mother, for example, observed at the end of our hour-and-a-half together that she had never really considered what it meant that her baby had been tested – without her knowledge or consent -- for a condition that had still not resulted in any symptoms. "I appreciate this [interview]," she said, "because it makes me realize we need to see it on the other end" – that is, to see it in terms of the general social implications of mandatory testing policies. Meditating further on her situation, she wondered where mandatory genetic testing will stop. "When we hit forty, are they gonna instantly test us to see if we're genetically disposed for breast cancer?" she asked. "You're gonna force that on everybody?" Although I by no means set out to alter the consciousness of parents who gave their time for my study, it was gratifying to see that in many cases I was not the only one learning: parents were intrigued by the sociological formulation of my research questions (as written in my study flier and informed consent form), and by what I asked them to think about and respond to during our interviews.

I believe that what I have written may pique the interest of others as well. By focusing on qualitative aspects of parents' experience with newborn screening, and by connecting my research directly to pressing health-policy and -practice issues, I have

tried to maximize this study's potential to bring sociology out into the broader public sphere where academics can be joined by a diverse array of interested persons in a discussion of what newborn screening means for our collective future. Perhaps "Testing Baby" can move us one step closer to that day when a lead article in the New York Times will feature a sociologist advocating that we can't understand what expanded newborn screening means without also "doing some sociology," and when at least some readers will nod their heads in knowing agreement. If so, it will have satisfied my most heartfelt ambitions for it.

In Closing

Almost twenty years ago, Katz Rothman concluded her study about the impact of amniocentesis on mothers by observing that with prenatal testing and selective abortion, "we ask mothers to decide just what kind of child they choose to mother" (1986, p. 243). In so doing, we alter motherhood, putting at risk what Sandel, in a recent article entitled "The Case Against Perfection," describes as the ability to "appreciate children as gifts… [and] accept them as they come, not as objects of our design or products of our will or instruments of our ambition." Parenthood is and must remain, Sandel asserts, "a school for humility… in a world that prizes mastery and control." The "openness to the unbidden" we embrace by having children in the first place "… invites us to abide the unexpected, to live with dissonance, to rein in the impulse to control." It teaches us to keep in balance what, according to Sandel, the theologian William F. May describes as "accepting love [that] affirms the being of the child," and "transforming love [that] seeks

the well-being of the child," assuring that "each aspect corrects the excesses of the other" (2004, 55; 57; 60).

With ever-expanded newborn screening, a different but related change in parenting looms large. In addition to asking mothers to decide what kind of child they want to mother, we are now beginning to tell mothers right from birth what kind of child they have in fact given birth to. Of course, as parents discover over time, the diagnosed abnormality is just one aspect of who that baby will become. But because the newborn is still so little known at the time of diagnosis, it is difficult -- as it is during pregnancy -- to separate the disorder from the child's overall identity. As we screen for more and more conditions, and as awareness grows that such testing is a routine aspect of newborn care, I worry that parents might become hesitant to announce the happy birth of their baby until the results come back. I worry that the "tentative pregnancy" created by prenatal tests might be replicated in the form of "tentative parenting" while the next diagnostic battery – the mandatory one we do at birth -- runs its course. I worry, ultimately, that tests designed to rule out or diagnose genetic disorders will become instead a test that babies must pass, a bar they must meet, before they gain full entry into their families and communities. Perhaps this is too much to ask of them at such a young age.

Appendix A

Parenting in the Genomic Age Project

Are you interested in sharing your experiences as the parent of a child with a genetic condition?

I want to learn about the things parents of children with a genetic condition experience during and after receiving their child's diagnosis.

I am a researcher from the Sociology Department of the Graduate Center of the City University of New York working on my graduate degree. I am interested in this topic because I believe the health care system might work better if the experiences parents had were better understood. The person who gave you this flier is helping me find people who might want to be part of this project, but the project itself is run by me.

I am looking for parents of children with cystic fibrosis or with other genetic diseases which do not necessarily result in symptoms beginning at birth, who are 18 years or older, and who would be willing to talk with me for about an hour. I am looking for parents whose child was diagnosed either before birth, via newborn screening, or once symptoms appeared.

If you would like to know more about the Parenting in the Genomic Age Project, please contact me, RACHEL GROB, at (914) 395-2371 (day time) or (845) 358-0022 (evenings and weekends), or by email at rgrob@slc.edu. If you are interested in participating after we speak, we will make a date for an interview by phone or in person that is convenient for you. I will:

- **Make sure all the information you give me is kept confidential**
- **Pay transportation costs for you if we meet somewhere you have to travel to get to**
- **Share the results of my research with you**

I can explain more about the project when you call. It is perfectly fine if you decide you don't want to participate after you learn more.

Appendix B

Face Sheet – Parenting in the Genomic Age Project

1. ID: __ __

2. Site of Interview: _____

3. Date of Interview: __ __ / __ __ / __ __ __ __

Notes:[Location, privacy issues]

READ: First I would like to ask you a few questions about yourself. How old are you?

4. Age: __ __

5. (Observed) Gender: 1. Male 2. Female

6. There are many different ways people identify themselves. What racial/ethnic group do you consider yourself to be? _____

7. Who lives in your house with you?

	Relationship	Age	Gender
a	_____	____	_____
b	_____	____	_____
c	_____	____	_____
d	_____	____	_____
e	_____	____	_____
f	_____	____	_____
g	_____	____	_____
h	_____	____	_____

8. (If no mention of child's other parent). Do you have a relationship with the baby's other parent?

9. Which of your children has received a diagnosis of cystic fibrosis?_____

9a. Have others in your family been identified as carriers of the cystic fibrosis gene?

10. Occupation (or former occupation) and partner's occupation if applicable.

INTERVIEW GUIDE

1. Did you want/seek/receive genetic testing during pregnancy?

PROBES:
If so, what were you tested for?

Were there tests you refused?

2. Do you remember giving informed consent for these tests? If so, was this process helpful to you in thinking through what it meant to be tested and whether you wanted to go ahead?

3. Do you remember receiving genetic counseling before getting these tests?

If so, was this process helpful to you in thinking through what it meant to be tested and whether you wanted to go ahead?

4. Were any genetic issues for your newborn identified during prenatal testing?

If so, what impact did those test results have on your experience of pregnancy? On parenting?

5. Did your newborn receive screening for genetic conditions via a heel stick? (If no or unsure, skip to question 6 below.)

PROBES:

Were you aware this testing would occur?

Were you asked to give consent for it to occur? If so, did you consent?

What happened when your baby screened positive?

How and by whom were you contacted?

What was your response at receiving the news?

What sort of follow-up was recommended?

What were the next steps you took?

6. Did your child receive his/her diagnosis after the development of symptoms or emergence of developmental delays? If yes, what was that process like?

PROBE:

What sort of process did you go through when trying to discover what was wrong?

Did it take a long time to find out what was going on?

7. What sorts of service providers are involved with you and your family and your child now?

8. Have these providers changed over time?

9. With which of your doctors and other providers would you say you have the best relationship?

PROBES:

What makes that relationship strong?

Have these relationships changed over time?

How was your relationship with your doctors and other care providers around the time you got your child's diagnosis?

Are those relationships different now? If so, how? Why do you think they are different?

10. Do you share with your providers your own observations and ideas about what is happening with your child, your knowledge of how he/she is doing, what is changing in their lives and with their health, etc.?

11. If so, how do they respond?

12. Are some more responsive to this than others? If so, which ones?

13. What kinds of skills do you need to advocate on behalf of your child?

14. How would you say receiving a genetic diagnosis for your child has affected the way you look at him/her?

15. How has the diagnosis affected your relationship with other family members?

16. Would you have wanted the diagnosis to be made at a different time, earlier or later?

17. Would you have wanted the diagnosis to be made in a different way?

    PROBES:

    Would you have wanted to know more about the testing ahead of time?

    Would you have wanted more tests earlier?

    Would you have wanted more decision-making power? More direction from health-care providers?

18. Do you know parents of other children with the same diagnosis who received the diagnosis differently than you did (e.g., prenatally, by newborn screening, after symptoms appeared)?

    PROBE: How would you say your experience compares with theirs?

Appendix C

Concepts from Interview Data Inviting Further Exploration

- Kindness of health-care providers
- Relationship between newborn screening and research -- being a "guinea pig"
- Being pitied after the diagnosis
- Difference in response to a disorder that causes physical problems rather than mental ones
- The grueling care routine
- The impact of the internet on peer support for parents of children with CF
- Ethnicity and CF: hurtful and inaccurate stereotyping/assumptions by health-care providers
- Insurance issues
- The role of faith in coping with diagnosis and illness
- Siblings, both sick and well
- The sick child and school
- New genetic technologies
- Being "chosen" to care for a child with CF
- Gender dynamics between husbands and wives
    - Differences between how mothers and fathers cope with parenting, diagnosis, disease
    - Mothers in charge of CF
    - Fathers as "helpful"

- Gender differences with respect to decisions about second/subsequent pregnancies, amniocentesis, abortion
- Working as a team for the child
- Diagnosis and disease as added stress in marital relationships

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ACMG: see American College of Medical Genetics.

ACOG: see American College of Obstetricians and Gynecologists

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