"THAT DISORDER": AN INTRODUCTION

> For I the Lord your God am an impassioned God, visiting the guilt of the fathers upon the children, upon the third and upon the fourth generations.
>
> —EXODUS 20:5

First there is the grandfather who has died of "nervous trouble" on the back ward of a state hospital, the uncle who attracts whispers and stares from the neighbors as he staggers down the street, the doctor who says, "Women do not get it." Rumors of hereditary insanity linger about the family in question, along with a certain atmosphere of secrecy and suspicion. Divorce, arrests, abandonment, suicide punctuate the action. There is always a moment of discovery, when the protagonists finally learn the truth, usually after having had several children. In the end, the characters all come to resemble one another, and the action winds down to a predictably gruesome close, with no resolution or release and always the promise of more performances to come. This is the drama of families with Huntington's disease (formerly called Huntington's chorea), played out with minor variations on stages around the world.

In the summer of 1968, my sister and I discovered that this drama was also our own, when our fifty-three-year-old mother was diagnosed with Huntington's disease. In our mid-twenties,

we learned for the first time the hidden history of our family, summed up in the awful word "chorea." We learned that our maternal grandfather and all our uncles had died of this disease and that our mother would repeat their fate. Nancy and I each faced a fifty-fifty chance of inheriting her disease ourselves.

Back in 1872, the physician George Huntington wrote the classic account of the disease that would become associated with his name. He had learned about it from his father and grandfather, both physicians, who had seen it among their patients on Long Island, New York. It was "confined to certain and fortunately a *few* families, and has been transmitted to them, an heirloom from generations away back in the dim past." It was spoken of by those "in whose veins the seeds of the disease are known to exist, with a kind of horror, and not at all alluded to except through dire necessity, when it is mentioned as *'that disorder.'* "

The symptoms began extremely gradually, "by the irregular and spasmodic action of certain muscles, as of the face, arms, etc." The movements grew progressively worse over a period of years "until the hapless sufferer is but a quivering wreck of his former self." In the end, every muscle in the body was affected "(excepting the involuntary ones), and the poor patient presents a spectacle which is anything but pleasing to witness." Nor could the patient hope for remission. "I have never known a recovery or even an amelioration of symptoms in this form of chorea; when once it begins it clings to the bitter end."

Huntington described three notable peculiarities of the disease. One was a marked tendency to insanity and sometimes to suicide. "As the disease progresses the mind becomes more or less impaired, in many amounting to insanity, while in others mind and body both gradually fail until death relieves them of their sufferings." Another was that of late onset: rarely before the age of thirty or forty, "while those who pass the fortieth year

without symptoms of the disease, are seldom attacked." (In fact, onset may occur both earlier and later.)

The pattern of hereditary transmission was perhaps the most striking aspect of the disorder. If a parent was afflicted, "one or more of the offspring almost invariably suffer from the disease, if they live to adult age. But if by any chance these children go through life *without* it, the thread is broken and the grandchildren and great-grandchildren of the original shakers may rest assured that they are free from the disease." This illness never skipped a generation to reappear in another. For those who were stricken, however, no treatment helped, "and indeed nowadays its end is so well known to the sufferer and his friends, that medical advice is seldom sought. It seems at least to be one of the incurables."[1]

Our mother's diagnosis in 1968 prompted my father to organize the Hereditary Disease Foundation to support research, and my sister to become a researcher herself. The research in which she participated led to a breakthrough in 1983. That summer, scientists localized the Huntington's gene on the short arm of chromosome 4 by identifying a genetic marker for the disease—a neighboring stretch of DNA indicating the proximity of the Huntington's gene. This event marked the first significant advance in Huntington's research, since the marker would make possible the identification of the gene and, it was hoped, lead to an understanding of how that gene caused brain cells to die. The marker also enabled researchers to identify who would develop the illness years, even decades, in advance of any symptoms. The dream of prediction long cherished by geneticists and counselors and even by affected families became a reality.

The marker discovery reverberated throughout the biomedical community. It demonstrated for the first time the power of a controversial new technology for mapping genes and opened

the way to accelerated advances in many areas of human genetics. Never before had this technique been used to locate a disease gene that could have been anywhere on any one of the twenty-three pairs of human chromosomes. Moreover, the localization of the Huntington's gene marked a significant step in the union of human genetics, which had been largely clinical and descriptive, and molecular biology, which had been highly reductionist and focused on mechanics. The coming together of these two worlds fundamentally transformed each of them.[2]

Second, the extensive dialogue that developed around presymptomatic testing for Huntington's has served as a model for thinking about all kinds of genetic testing. The cautions that scientists, doctors, health professionals, and HD activists have built into the procedures for testing have served as examples for those developing ways of testing for other illnesses as well. In a world in which growing numbers of disorders may be diagnosed before symptoms appear, even though there may be no effective therapy for them, the response of the Huntington's disease community has been carefully watched throughout the biomedical world.

Third, the way in which the research on Huntington's has unfolded—through interdisciplinary workshops, collaborative efforts, and a high degree of cooperation between families and investigators—has also served as a model for other research ventures. Although scientists collaborate on all sorts of projects, molecular biology and biomedical research have been arenas of especially fierce competition. The Huntington's Disease Collaborative Research Group, organized under the auspices of the Hereditary Disease Foundation, has been considered by many to be a model of a successful, large-scale collaborative effort in biomedical science.

Fourth, many have seen the status of being at risk for Huntington's as an extreme example of what it means to be at risk for a wide range of other conditions, including AIDS.

Huntington's poses stark questions about the meanings of certainty and uncertainty and what it means to occupy a "third space" outside the categories of either-or that we conventionally use to organize experience.[3]

Finally, Huntington's is also about nonscientists playing an active role in science, not only through fund-raising and lobbying but by participating in decision making about which research to support and working with scientists to organize research efforts. By intervening directly in the scientific world, these Huntington's activists have significantly influenced the priorities and practices of biomedical research.

My family's involvement began at a moment when biomedical interest in this disease had already started to revive, on the eve of the recombinant DNA revolution and the blossoming of neurobiology in the late 1960s. The scientific milieu was highly favorable to the intensification of interest in a disease like Huntington's, with its combined neurological and genetic dimensions. At the same time, its late onset made it peculiarly difficult to study, since it was hard to distinguish those who were unaffected from those who might develop the disease later on.

The decade of the 1960s, with its blossoming of social activism, also helped foster a political atmosphere favorable to mobililizing families directly affected by the illness. Civil rights activism, the feminist health movement, and patients' rights movements of the sixties and seventies all created an environment that encouraged families with Huntington's to act on their own behalf. Moreover, Woody Guthrie, the great poet and songwriter of the Dust Bowl, had died of Huntington's in 1967, the year before our mother was diagnosed. His long illness had inspired Marjorie Guthrie, his ex-wife, to start an organization of people with Huntington's in their families. Founded in 1967, the Committee to Combat Huntington's Disease, or CCHD, grew into a national, grassroots organization—later called the

Huntington's Disease Society of America, or HDSA—which lobbied Congress, developed services for families, and organized educational campaigns for the public and for health professionals.

Although inspired at the beginning by Marjorie Guthrie and CCHD, my father's deepest commitment was basic research. Imbued with a profound faith in science, he wanted to find a cure. At a time when the disease was of interest primarily to a few neurologists and geneticists, my father and sister helped create a support system—seed money, tissue banks, pedigrees, and workshops—that enticed many basic scientists to study Huntington's. In doing so, they pioneered imaginative ways of working with scientists, and of fostering dialogue among scientists, which would have implications far beyond the Huntington's research community, a community they helped to create.

This book began as a project of documentation in that heady summer of 1983, when we thought Huntington's might soon come to an end, like polio after the invention of the Salk and Sabin vaccines. As a historian, I wanted to record this first major turning point in the history of an obscure, seemingly hopeless illness and in the development of human neurogenetics. That summer, I began to interview the scientists who had been involved in the marker discovery, as well as others who had been associated with the Hereditary Disease Foundation. I hoped to collect memories before they became too encumbered by myth and before all the publicity in the press began to feed back into the scientists' recollections.

I soon realized that, as a member of a family that had been deeply involved with this effort and had helped to fund it, I could not write as an outsider. Although my own role has been primarily that of an observer, I was too close to the participants to write about their efforts with much critical distance. And, as

a person at risk for Huntington's, I was too emotionally involved in the outcome of this research to regard it with much detachment.

I realized further that I did not want to write as an outsider, nor did I wish simply to document an exciting moment in the history of biomedical science. In my early forties and approaching the age at which my mother had begun showing symptoms, I wanted to explore the emotional meanings of being at risk, for my mother as well as for myself. Although my sister and others had studied the psychology of being at risk for Huntington's, few people actually in that position had written personal accounts of their experience outside the context of psychiatric testing, genetic counseling, the neurological exam, or the journalistic interview. As a feminist I particularly wanted to examine the relations between genetics and gender in our family, since I knew it somehow mattered to my own experience of growing up female that my mother—my same-sexed parent—was the parent at risk and that she was the one who had developed the disease. I wanted to see how our lives intersected, the rhythms of her hopes and anxieties informing those of my sister and me. Huntington's, I thought, could even be seen as a metaphor for the fear of many daughters of my 1950s generation—that we would somehow turn into our mothers, that our mothers were mirrors of our future selves—and for that common guilt of our mothers, that they had inflicted suffering on their children. What was the mother-daughter relationship, when viewed through the lens of Huntington's disease? *If the mirror, whose precursor is the mother's face, offers an illusion of wholeness to the child's body of bits and pieces, what then of the daughter who sees the mother imagining herself and imagining her daughter as the fragmented body she fears to become? What psychic map of the body is projected by a mother who recalls her own parent's choreic body? What map of the body is taken in by the daughter who sees chorea memories written on her mother's face?* [4]

Finally, I wanted to explore the meanings of secrecy and silence within our family, the ways in which what could not be said reverberated as loudly as the words that were spoken. Feelings cannot be buried as easily as facts. Denial creates its own emotional force fields, even if the relevant information remains hidden. Secrets, moreover, especially so dramatic a secret as Huntington's, may form part of a family's emotional inheritance, a psychological legacy handed down along with the family Bible, affecting every aspect of family life for generations. Since Huntington's had been a secret in our family, long hidden from my sister and me, our mother's diagnosis had implications far beyond the medical. Learning of our mother's failure for many years to tell our father about Huntington's disease in her family, and discovering our parents' decision not to tell my sister and me of our mother's risk, and therefore our own, meant recasting my entire understanding of our family history.

Our situation, then, had much in common with that of other families whose secrets differed in their content. Certainly any stigmatized condition may be surrounded by webs of secrecy, whose maintenance requires hard work and active effort. As the historian Michel Foucault argued, "Silence itself—the things one declines to say, or is forbidden to name, the discretion that is required between different speakers—is . . . an element that functions alongside the things said, with them and in relation to them . . . we must try to determine the different ways of not saying such things, how those who can and those who cannot speak of them are distributed, which type of discourse is authorized, or which form of discretion is required in either case. There is not one but many silences."[5]

In exploring the impact of secrecy in our family, I also wanted to consider the ways in which our silences were gendered. In recent decades, much feminist writing has addressed the costs for women of socially imposed silence, especially the

silencing of our own deepest thoughts and emotions. Feminist historians in particular have described how the female body became the arena in which forbidden speech was acted out through physical symptoms. In our family, much remained unspoken and unspeakable until that day in 1968 when our mother's body spoke that (death) sentence. This book, in part, is my translation.

My decision to write about HD in our family raised many questions for all of us as to what kind of book this would be, particularly because there is still no effective treatment for Huntington's and the research and fund-raising efforts continue. Would this book affect those efforts? Would I write an "official story," celebrating the successes of the Hereditary Disease Foundation? a family romance to inspire others? a publicity piece for fund-raising purposes? What about those family members who did not want their personal lives publicly disclosed, particularly by someone who might not represent them as they wished to be shown? The fact that my father is a psychoanalyst, and a most unorthodox one at that, further complicated the project because of the involvement of some patients and former patients with the Hereditary Disease Foundation. There was also the fact that I was challenging Dad on his turf by interpreting our family, often differently than he did. As the book grew more autobiographical, my anxiety deepened. I realized I was no longer writing a "family story" but rather a memoir of my own.

The book I have written is far more personal than the one I intended when I started, partly because I began to see just how profoundly Huntington's had colored our family's history and, conversely, how our family history, in all its particularity, has shaped the ways in which we have responded to the illness. Unresolved angers and resentments from the past often got displaced onto arguments about the disease, while the disease, in

shadow on multiple aspects of the family history, even before we knew of its existence. In short, we made emotional and metaphorical meanings out of the disease that were not determined solely by its biomedical character. The disease was often the vehicle for expressing feelings only tangentially related to it. As the medical writer Arthur Kleinman put it, "Acting like a sponge, illness soaks up personal and social significance from the world of the sick person."[6] To tell the story of the disease, then, I would have to address these other dimensions.

This wider aspect of the story has proven to be the most difficult to write, since each of us brought a different set of associations to bear on the problem of Huntington's and distinct conceptions of what counted as important in telling the story; indeed, different constructions of "the story." Certainly, in any family history, questions of privacy and confidentiality emerge in relation not only to the past but also to the present; what one person considers as central to his or her narrative of identity may involve information that others in the family consider private and "no one else's business." All members of a family, in some sense, compete for control over the family narrative, at least when it comes to public speech, and perhaps even in private as well. Whose voice may be heard, whose speech is legitimate, who can tell their own story when it also involves the stories of others—these are questions every writer who ventures out onto the thin ice of autobiography must face. When inherited illness confronts a family, such questions become especially charged, as feelings of anger, guilt, blame, and loss inevitably come into play. To erase those parts of our history that were traumatic or embarrassing, to others as well as to myself, in the hope of presenting a more heroic image, seemed to me not only false but unfair to those who are currently struggling with similar dilemmas. I wanted to show how even a family with all kinds of advantages, like ours, can still struggle mightily with such a disease. Yet I have continually wrestled with the

knowledge that my speech was capable of causing others considerable pain.

Another set of questions concerned the audience to whom this book was addressed. For whom was I writing this book? For the "general public" interested in medicine and science? For medical students or genetic counselors? For scientists who wanted to learn about the emotional aspects of a disease they know only in the lab? For other people with Huntington's in their families? Was it possible to draw on some of the recent feminist critiques and cultural studies of science while still writing in a way accessible to these groups?

In the course of writing this book I have grappled continuously with these questions. At the very least, I have tried to be aware of my position as a white, Jewish, upper-middle-class woman who writes from within an academic community but outside the structure of an academic institution. Even while maintaining a critical perspective, I have tried above all to speak in ways accessible to those most affected by this story—people in the Huntington's community—in the hope that this story may encourage others to tell their stories as well.

In weaving together a personal narrative of a family confronting Huntington's disease with a more detached account of biomedical research, I have utilized several approaches. On the one hand, I have drawn on the traditional resources of the historian and journalist in investigating the past—interviews with many scientists, archival materials, and reports in the scientific, medical, and popular press, as well as personal observation at meetings and workshops. At the same time, I have pored over family papers and photographs, old letters, newspaper clippings, scrapbooks, transcripts, conversations over many years, even dreams and memories, diaries and journals. I hoped my doubled perspective as insider and outsider, as participant and observer, might be a useful position for approaching a topic that is both scientifically significant and emotionally volatile.

While my story shares some of the elements of the illness narratives written about cancer and AIDS, it is really less about an illness than about the possiblity of an illness, less about the medical dilemma of living with disease than about the existential dilemma of living at risk.

In writing this book, however, I have come to realize how privileged my sister and I have been in relation to other families with Huntington's. We have had more resources, financial and professional, than most other families with the disease. Moreover, our family is small, so that the illness has not multiplied through the generations—of my mother's family, only she had children, while her three brothers who had the illness had none. Besides my sister and me, there are no other direct descendents of our grandfather, Abraham Sabin. Most of all, our mother developed symptoms late, after my sister and I had left home, so we did not grow up under the frightening shadow of this disease that haunted so many other young people, watching their parents and grandparents, uncles and aunts, siblings and cousins growing ill and dying. A more typical experience is that narrated by a woman who testified before the Congressional Commission for the Control of Huntington's Disease and Its Consequences in 1977: "As we were growing up, all of us felt that our family suffered from hereditary insanity and that we were singled out as a family with this 'crazy streak.' We were taunted with 'Your mother is in the crazy house!' I did not then even know that my mother was alive, and if she was alive and in the crazy house, I did not know why. My mother, two uncles, two brothers and one sister have been diagnosed with HD. My brothers' and sister's spouses all divorced them immediately after committing them to the state mental hospital." Or, as another woman testified, "Now this insidious killer has a possibility of forty-five victims in our family alone."[7]

Huntington's is a devastating disease. Yet not all of its misery

comes from the illness. People with Huntington's can and sometimes do live active lives for a number of years after the diagnosis, if they have the necessary supports and services. The suffering associated with the disease and with living at risk is intensified by the lack of resources available in our privatized, for-profit medical system. Nearly all of the people from families with HD who testified before the 1977 Congressional Commission spoke of the limitations of health insurance and lack of access to services. That people with Huntington's who are still relatively intact intellectually and emotionally often end up in state mental hospitals merely underlines the failures of our current health care system and the need for a national program that addresses the needs of chronic, long-term illness.

Human genetics in the 1990s inhabits a volatile space at the intersection of medicine, biology, corporate profits, law, government funding of science, state health programs, private insurance companies, genetic counseling services, schools, courts, and popular culture. Issues of race, gender, and class figure in the discourse of the new genetics, reviving old debates about the distribution of traits such as intelligence and aggression, dominance and disease, within different groups of the population. Genetic engineering is a multibillion-dollar industry, with companies competing for control over diagnostic tests for newly discovered genes or markers of lethal illnesses, whose discoverers are often shareholders and members of the board of directors of the companies that will market the tests. Clearly many groups of people have strong stakes in the technologies that are revolutionizing all of biology.

The new genetics has already opened a vast arena for contests of power over what it means to be human, who has the power to define what is normal, who has access to what resources and when. Who will control the knowledge of our bodies after the Human Genome Project has mapped and sequenced all human genes? How can we ensure that this will

not be another project for enforcing narrow norms of "human nature," as the historian of science Donna Haraway has put it, for legislating "genetic destiny"? How can we respect the diversity and difference that the Human Genome Project also establishes as "normal"?

Although Huntington's affects a limited population—some seven to ten people per 100,000, or about 30,000 in the United States, with another 150,000 at risk—it has usefully been considered a prototype for biomedical research since it destroys such a wide range of functions. Understanding Huntington's may shed light on more common inherited, neurological and psychiatric disorders, such as Parkinson's, schizophrenia, and sickle-cell disease.[8] Because it is caused by one gene, however, some have argued that Huntington's may be an inappropriate model for thinking about disease, since most diseases are caused by complex interactions of genes and environment, or by a combination of genes. As the historian of science Evelyn Fox Keller has written, only for very exceptional diseases can "genetic components be considered apart from the environment. For such cases—e.g., cystic fibrosis and Huntington's disease—there is no question that molecular genetics is providing powerful and unambivalently welcome tools. But most diseases are not so simple." Indeed, the ever-expanding category of genetic disease has recently threatened to claim such social conditions as homelessness, while what Keller calls "the geneticization of health and disease" threatens to move discussions of disease from individuals to their DNA.[9]

But even Huntington's, with its straightforward genetics, may not be entirely reducible to DNA, since the cellular environment and even social milieu may also influence its expression. My hope is that the Huntington's story may suggest the ways in which even this obviously pathological, genetically determined killer may acquire distinct meanings for different individuals, families, and cultures. Biology itself, in this view, is

are partially shaped by its social, political, and cultural contexts. Moreover, as recent social studies of science have argued, what counts as biological "fact" may be partly a product of cultural struggles over power.[10] Part of the fascination of the new genetics concerns the questions it raises about the construction of knowledge—how, for whom, and for what is this knowledge being constructed? In this context, then, Huntington's disease may serve as a space where many discourses collide and therefore help make visible the hidden stakes in this contest for human survival and identity in which all of us are at risk.

Huntington's is, above all, a disease of endless replication, reducing the wonderful multiplicity of human lives to a dreary, deadening sameness, repeating over and over again the same awful saga. In this connection, I have often thought of a story by Jorge Luis Borges, in which a modern-day gaucho dies in Buenos Aires in order that a scene from the life of Caesar may be reenacted. In the story I have written, the gaucho lives to tell her own tale.

—Santa Monica, California
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