

GENETIC "RUSSIAN ROULETTE"

NANCY S. WEXLER

Genetic "Russian Roulette"

The Experience of Being "At Risk" for Huntington's Disease

Genetic Counseling: Psychological Dimensions 1979 by Academic Press, 199-220

Huntington's disease (HD) is an hereditary disorder of the central nervous system. It is transmitted through a single autosomal dominant gene with complete penetrance. Symptoms usually appear in adult life between the ages of 35 and 45. However, persons as young as 2 or as old as 80 have been known to develop the disorder. The disease is most often characterized by chronic progressive chorea and dementia, without remissions. The childhood form of HD, which affects 10% of cases and is inherited from the father 75% of the time, and some adult variants, are marked by rigidity rather than chorea. Psychoses and affective disorders as well as milder emotional disturbances are frequent either prior to or following the appearance of abnormal movements. Life expectancy after the onset of symptoms is on the order of 10 to 20 years for adults, 8 to 12 years for children. Death comes as a result of secondary infections, heart failure, or aspiration. There is no predictive test available and treatment is only marginally effective. For the HD victim, the fatal determination is fixed at the moment of conception. Unknowing, the individual moves through life; the marked gene lies quiescent or its slow catabolic progress is masked by the forces of new cell birth. Then, at the peak of productivity, things go awry. At the stage when most individuals must face the transition from youth to middle age, the HD victim prepares for the passage from youth to death.

If the individual knows he or she is at risk, the years in anticipation of this change can be years of dread, of silent apprehension, of noisy emotional disarray, or of intense productivity. As is frequently the case with a late-onset genetic disorder for which there is neither a screening test nor treatment, individuals at risk are disease wise and doctor shy. They know the odds from an early age and often regard the genetic counselor with the same wary contempt the battlefield soldier shows for the journalist -- ticking off in round numbers the daily toll of lives lost, reducing the human struggle to arid statistics. Nor is the psychological counselor seen as a source of refuge. Mental health workers are considered to be only for the mentally deranged while those at risk perceive themselves to be suffering from a physical threat.

As a clinical psychologist concerned with genetic diseases, I was interested in exploring how the two disciplines of psychology and genetics could pool their expertise to render a more comprehensive service to the client. If the genetic counselor is seen only as a purveyor of information, and the individual already is in possession of that information, he or she will not seek counseling. In order for counseling to be useful for well informed clients, both client and counselor must redefine their major task to be learning to cope with what is known. The genetic counselor or a psychotherapist familiar with genetics should be able to help the individual "work through" (in the psychoanalytic sense) the relevant facts. In doing so, the counselor must be sensitized to detect the subtle psychological effects that the state of genetic risk is likely to produce. It is my firm conviction that although individuals vary widely in their reactions to threat, there are still communalities of concern which cut across individual differences and these can be taught to counselors. In knowing approximately what to expect, the counselor is better able to listen, anticipate, probe, assure, and console.

In order to learn more about the inner world of an individual at risk for a serious genetic illness, I interviewed in-depth 35 persons at risk for HD. My respondents were 12 men and 23 women between the ages of 20 and 36. All had one parent, living or deceased, affected with HD. None had been diagnosed with HD or any other neurological disorder. All socioeconomic classes were represented,

although most were in the middle or lower middle class. The mean level of education achieved was 14 years. The interviews, some conducted in my office, some in the respondents' homes, were open-ended and exploratory. What follows is a distillation of the main themes of these interviews as they emerged -- the fears, griefs, and hopes of persons coping with an unusual life situation. They are the stories which would be told in the genetic counselor's office, if the door were open to them.

Reactions to HD Symptomatology

Every disease calls forth particular images and fears in its victims and potential victims. Cancer evokes the threat of pain and suffering; unpredictability is the hallmark of multiple sclerosis. For the person at risk for HD, the relevant metaphor is the time bomb. To be affected by HD had specific meanings for most of the at-risk individuals I spoke with, because of the nature of its symptomatology. The primary concerns for these people were the intellectual deterioration and personality changes wrought by the disease, the socially embarrassing choreiform movements, regressive problems such as incontinence, and, especially, the extreme dependency involved in becoming chronically ill.

All the at-risk individuals interviewed had known their affected parent and had watched that parent change and decline from a familiar, healthy person to someone somewhat unrecognizable, with bizarre movements, uncontrollable behavior, and slurred speech. For many of the interviewees these changes took place during the child's formative years, often leaving the child with a distorted understanding of the peculiar transformations that had claimed an often beloved parent. In their adult years, these at-risk individuals still retained an image of the illness which they had conceptualized as children. Those who had been particularly frightened as children had especially sinister visions of the disorder as adults. In contrast, in families in which the ill parent was able to remain a functioning member of the family, even marginally, the children accepted the illness with greater equanimity. The nature of the children's early exposure to HD appeared to be critical in determining their adult adjustment to their own genetic risk.

For all these men and women at risk no matter how mature and well-adjusted they were to the presence of the illness in the family, the nature of HD symptomatology seemed to strike at the core of their physical and psychological self-esteem. The peculiarities caused by uncontrollable movements and mental deterioration became translated for many into a vision of a Frankensteinian monster, one who approaches others with affection but from whom others recoil in horror. Subjects spoke repeatedly of how "disgusting," "repulsive," "grotesque," "ugly and horrible" the HD patient becomes. There was a particular dread of losing bladder and bowel control. Some reported feeling nauseated at the sight of their ill parents. One 36-year-old father spoke poignantly of the horror of the anticipated bodily changes.

It's an awful thing to look at your kids and wonder if some day they're going to look at you like some kind of monster . . . just to look in a mirror and see yourself change like that. To look in people's eyes and see how they're afraid . . . and they'd shy away from you and you'd feel hurt. Years ago, you used to read how they'd burn people with HD at the stake and things like that because they were possessed by the devil. I can sure see why they would think so.

Despite the ghastliness of those visions, for all but two at-risk individuals interviewed the most frightening aspect of HD was not the uncontrollable movements but the loss of intellectual capacities. A 35-year-old woman expressed the group's consensus, "You can live with jerkiness but you can't live without your mind." Another woman had a recurrent dream that her head was turning into oatmeal. Nearly all talked of "becoming a vegetable," "stagnating," "going crazy," "having your mind garbled," of not being able to communicate, and of the terror of ending their lives in a mental institution.

Then I saw my mother in the institution with other mental patients. You can imagine what it was like for me to go in there. That's when the severity of it really hit me. I realized it wasn't just like going off to school and playgrounds, fine and everything. Seeing the bars on the windows, I thought, "My God! Why do they need bars and everything" [woman, age 25].

Many at risk had parents who became paranoid and delusional as the disease progressed. One man spoke of his horror and fear of mental illness after seeing his aunt who was affected with HD. All the respondents talked of personality changes and severe personality disturbances in the affected parent, sometimes antedating the diagnosis of the disease by more than 10 years.

Perhaps because most of the respondents were in the prime of their productive years, with newly achieved independence, the threat of extreme dependence on others as well as anticipated abandonment was another anathema associated with the illness.

I worry about being a burden on my family financially. Here's a guy who could've had everything but he's got somebody with HD. It'll take all his money and all his time. I guess mostly I think of how he's going to feel about it. This is why I want to fight; to fight for him. That's horrible for the person who doesn't have it and loves this person [woman, age 23].

The prospect of prolonged dependency and deterioration often made single people despair of marrying while those married considered immediate divorce when they learned of their risk. The single people questioned whether anyone could ever love and value them enough to want to share that risk. A 22-year-old woman felt her anticipated loneliness to be one of the worst aspects of the disease.

I guess, the thing I feel bad about on my part, the part I really dread, is having boyfriends see [my mother], 'cause I am very open about what it is and the fact that it's genetic and everything. And I think, wow, if they see how bad it is, it's just gonna be, "Forget it, baby." . . . It's not being terrified about having it sometime way in the future, like at the age of 35, it's always even when I was in high school, the thought of, oh, I'll never get married and, oh, I'll never be able to, like, it's never bothered me too much the idea of not being able to bear my own children, like I wouldn't mind adopting, but the thing of no one will marry me and the whole thing of being an old maid type of thing. It's really scared me more than just thinking about having the disease.

Those who were married wanted to protect themselves against the trauma of being left if they should become ill. Both groups fantasized taking active control either by not marrying or by divorcing; both felt that so great a burden of responsibility should not be inflicted on someone they loved.

My problem is that all my aunts and uncles have divorced their HD victims. I think it's just awful! The first thing I said was that if you want a divorce, divorce me now because I'm not living alone when I'm sick! [Notice she says "when," not "if."] [woman, age 23]

One of the most frequently voiced fears is of choking and starving to death. Although this is a concern based on reality, it is as if the anticipated emotional abandonment qua starvation is also verbalized in these physical terms.

All of the interviewees were painfully aware that the disease is terminal, but for them termination comes not at the moment of death but at the moment of diagnosis. Most fantasize the period following diagnosis to be a prolonged and unproductive wait on death row. The optimism or pessimism of at-risk persons is directly related to the kind of care they perceive their ill parent to have received, regardless of the severity of the symptoms. When the sick parent is kept at home, or in a good nursing facility, active and a part of the community, the specter of the disease is not nearly as

formidable. None of the at-risk individuals mentioned feeling afraid of death, per se. On the contrary, death is often cited as a welcome relief from life with symptoms.

Q: What is the least frightening symptom of HD?

A: Probably that you die is the part that scares me the least. It's almost more human than the part where you live. I know it is a fatalistic outlook but I can't help it, sometimes. After seeing my aunt for seventeen years -- she couldn't even roll over in bed -- I definitely feel that dying is a relief [man, age 36].

Many subjects did not wish to wait passively as nature took its course. Approximately half the sample felt that they would seriously consider suicide as an option if and when they started to deteriorate. Most of these individuals came from families in which the ill parent had made at least one suicide attempt. (There were no completed suicides among the parents.) Others insisted that they would never consider suicide but they could understand how others might. One man, age 30, had what he called "death insurance." He had vowed to commit suicide when he was no longer able to function. If he was not aware of the severity of his deterioration, or was unable to carry out his plan, he had made a pact with his brother that each should kill the other, should it become necessary. Some subjects who did not consider suicide as a viable option expressed the wish to be institutionalized at the point when they were no longer able to lead useful lives.

Genetic Disease and Family Dynamics

It is impossible to understand fully the impact of HD on families without an appreciation of the complexities which its hereditary nature imposes. Every disease can arouse images of body damage and destruction; each can awaken fears of dependency and rejection. But only if the disorder is hereditary does an individual know the exact probability of contracting it. If the disease is an hereditary disorder of late onset, the individual has most likely witnessed the illness in parents or close relatives. Each individual must take the responsibility for risking passing on the disorder to future generations or refraining from procreation. Such hereditary illness can have repercussions throughout three or four generations simultaneously and have impact on parent-child identifications at each generational level. The history of the disease within the family has crucial ramifications for all those family members who are still genetically vulnerable. In fact, much of the working-through process which an at-risk individual must undergo to accept the illness is often expressed in terms of responses to other family members.

The first reaction which most individuals at risk reported when told of the presence of HD in their immediate family was an overwhelming concern and grief for the afflicted parent. If they already had children of their own, they also felt great sadness, protectiveness, and guilt toward the child. The most common immediate reaction was, "What have I done to my child!" Not only were they genuinely concerned for the welfare of the children, but the at-risk parents' own fears for themselves could be much more acceptably expressed in terms of anticipated calamity for the child.

The fact that hit me the most -- I used to cry whenever I looked at my son. I was afraid. Like, I was scared. I remember a real bad emptiness in my heart [woman, age 23].

Because of the late onset of HD and frequent inaccuracies in diagnosis, many of the people interviewed had not learned of the hereditary nature of their parents' illness until they themselves had had children. Six of the women were pregnant when they learned of the risk to themselves and to the unborn child. (Only two were in the first trimester and both decided to carry the baby to term.) One 28-year-old mother of two small children described her reaction on learning that her mother's disease was hereditary.

I went through a bad depression for about two weeks . . . I'd sit there and cry because I'd think of how I might miss everything my mother missed. She never seen us married or have kids. She knows we have kids. I want to be a grandma . . . I told my husband I wanted a divorce so he could get out of the legal stuff -- let the state take care of you. I'd tell him who I wouldn't want to watch the kids and who I would want. I wouldn't care if he'd get married again and have another woman to raise the kids so long as she loved them; so I would know they had a mother. I want to be home as long as possible but when the day comes, I want to be put in a nursing home . . . It was mainly the kids and how much of their lives I'd miss, depending on how old I was if and when I got it. I'd cry if I thought of any of their graduations or getting married. Anything like that would bring on a tear-jerk. And, of course, me not being there mainly for both of them. Then, after the two weeks I just snapped right out of it like I went into it. I've never given it another thought. I've done said everything like I wanted to.

In most normal families it is common for a child to hear, either in humor or in anger, "You're just like your mother, or father!" For the at-risk individual such a remark has an added impact. Whatever emotions at-risk persons experience toward the ill parent -- tenderness, compassion, pity, disgust, resentment -- may someday be the very feelings their own children experience toward them. As they watch their parents they watch themselves; all emotions rebound.

What was upsetting was that here I'll be watching my Dad and maybe be watching myself. I felt I couldn't feel sorry for myself because I had to feel sorry for him. Yet here I was and nobody was going to feel sorry for me. I'd get mad at myself for being selfish [woman, age 25].

Frequently the person at risk becomes the confidante of the well parent who pours out grievances over the ill parent. The at-risk offspring is sometimes forced to mediate between the two. Often the child seeks to identify with the well parent and feels hostility for the sick parent for causing so much grief. Alternately, he or she may identify with the sick parent and experiences himself or herself as the potential recipient of the well parent's complaints. The child often urges the well parent not to be a "martyr" -- not to sacrifice life and happiness because of the ill parent. Children frequently must listen to suicidal temptations of both parents. And yet while they say aloud that neither they nor their relatives should devote their whole lives to the sick, they usually are well aware that they are encouraging the same dreaded abandonment should they themselves become ill.

If parents with HD were particularly psychotic, violent, or unavailable, it becomes even more difficult for those at risk to cope with the prospect of getting the disease. They experience the possibility of possessing the HD gene as signifying that they would literally turn into their parent -- a kind of cloning after birth. This fear seemed particularly powerful when the same-sexed parent was affected.

I had the impression that HD was like what my mother was. I was ready to pack my bags and leave. If we wouldn't of stayed and talked to that social worker for two or three hours, I probably would have left. I really thought it was like what my mother was. I didn't know there was symptoms and all that stuff. When I found out I wasn't going to beat my kids, be mean and jealous, I calmed down. I didn't want to be like my mother to my kids; I'd sooner left [woman, age 28].

The dilemma of identification in a family affected by genetic disease has many complex ramifications. In the group interviewed, all of the at-risk individuals had had strained and disagreeable relationships with the ill parent for some time prior to the diagnosis of the disease. To most, the medical recognition that the parent was truly ill -- apart from the hereditary nature of the illness -- came as a welcome relief. The rage and disappointment they felt over the parent's inadequacy could thus be directed to something external -- a sickness -- for which neither parent nor child was to blame. The parent was not cruel, ill-tempered, or inconsistent because of a failure on the child's part, nor could the parent be held accountable for performing destructive and hostile acts. The child's guilt over murderous feelings harbored toward the parent was dissipated, but in its stead came fear. They reasoned that if HD

caused behavior in the parent that was beyond control, might it not also cause the same behavior in the child if the child manifested the disease? This concern was especially felt by those individuals who witnessed a radical change in their parents from being loving and affectionate to being irritable and withdrawn or psychotic.

But I think really, the thing that really scares me the most is I know, despite all my determination to hold myself together and be a pleasant person to be around, I fear that having the disease will make me lose that control and I'll turn into a shrew like my mother. And that will be what will alienate me from people. And that will be against my control, even though I won't want to do it. Like I see she does things that she doesn't want to do, but she can't help herself [woman, age 22].

The most reasonable resolution of the dilemma, and probably the most accurate, was for the individual at risk to perceive the ill parent as the victim of circumstances and background. At least the at-risk child did not share the parent's early environment. The real culprits thus become the grandparents and anger can be safely deflected from the parent. The grandparent is also to blame for passing on the gene, and both parent and child become the hapless victims. Yet, while the parent has succumbed to his or her upbringing, the child fervently hopes to transcend environment and become quite different from the parent.

When I first heard about it, I thought, "My God! She's gave me something else!!" I was bitter with my mother. Then, all of a sudden I started getting into her background and stuff [woman, age 23].

Remarkably few interviewees expressed conscious anger toward the parent who had given them this legacy. Compassion and grief were by far the most common feelings. It was considered in particularly bad taste to harbor hostility toward a parent who was already broken and ill. However, the child covertly disavowed the parent by trying to become as different as possible. Frequently, anger was turned against the self and expressed in depression. The following reaction of a young man, aged 20 when he learned of the disease, is quite typical.

I was angry and enraged [when I first found out about HD], you know -- gee, what a terrible thing! Just then, my mother died from it. I grew up with it in my family. To think, I might have the same thing someday, I don't even want to be sick a day with the flu or anything. I haven't missed a day of work in three or four years. I just try to stay as strong and healthy as I can. I think about it all the time. Not all the time but like when I go to church on Sundays. I pray, you know, give me strength and health. That's all I ask. I can take everything else from financial ruin to losing all my friends. As long as I've got my health, I can come through. I don't like to be sick and a burden on anybody. I just want to take care of my own life. I don't want to be on welfare. That's a terrible thought. It could happen. It'd be terrible if it does.

Q: When you think of HD, what do you think of?

A: I just kind of think your body'd waste away. You're just probably tired all the time from shaking. That's a lot of physical exertion, I'd imagine, moving all the time. It'd really take it out of you. In a couple of years, you'd be exhausted. I try to keep my weight up. I'm kind of over-weight for my size. I like that. I want to be heavy and real strong. The charts recommend 140 lbs. for my size. I weigh 175 lbs. That's fine -- I weighed 200 lbs. a couple of years ago when I first got married.

Q: Was your mother thin from HD?

A: Yeah. Your throat or anything doesn't function right. She'd try to drink a Pepsi and choke on it.

Q: Did that scare you?

A: Oh gee!! People probably feel like committing suicide! I mean, I wouldn't. I want to live as long as I

can. I imagine if it comes to that, if somebody's that sick, they'd want to get a whole bottle of sleeping pills.

Q: Do you think you would?

A: I don't know. I'd hate to just be a vegetable. That's all you are. You don't really function any more. Maybe you should be dead. You'd probably be so far gone that it would be better to be dead. It's hard to say. For myself, when I'm 35 years old it could happen then. Now I'm 24 and maybe I've only got ten more Christmases to have good times in. Maybe I shouldn't feel that way. I might be 60 like my other aunt. I sometimes wonder what families are like that never have it.

Mr. M's initial reaction was one of anger and outrage at his mother. He had had to put up with her severe emotional and physical problems and now she had passed the threat on to him. However, resolution of these feelings was aborted by the death of his mother and the feelings were internalized. While it is true that most HD patients become emaciated and cachexic, Mr. M seems to fear the total annihilation of his body. It is as if the internalized feelings of rage and disappointment will dissolve his very physical being. He tries to counteract this anxiety with exaggerated weight and solidity. If this defense should fail, he contemplates suicide, particularly by "eating" an overdose of pills. Mr. M acts as if he might wake up one morning when he is 35 and find that his life is over. He hopes desperately to be one who escapes and then wishes, rather wistfully, to be quit of the whole concern. Whether or not a person feels destined either to develop or to escape the disease has repercussions in attitudes towards siblings. A surprisingly large number of those at risks felt emotionally convinced, regardless of what they intellectually knew, that at least one of their sibship must develop the disease. They considered that feeling free of the disease was tantamount to inflicting it on a relative. One woman thought herself a "marked target" because she was the youngest of three and her sibs appeared to be healthy. A very thoughtful young man felt he should get the disease because his sibs all had children and he did not.

Respondents frequently expressed the feeling that they could be free of HD only by paying some price. One woman volunteered: "Spare me and I'll be a good Samaritan and care for the rest." Others make more modest requests; they wish only to be left for last. The notion of hoping that the entire family escape altogether is considered almost too greedy to be expressed aloud: God punishes hubris. One 25-year-old woman's dream graphically illustrates this point.

I have always had nightmares. Now, the fear in my dreams is that I'm being followed by someone. The last one I had was where my friends and relatives were in a big room, a big house. There was this one man. I could see him but I couldn't warn anyone. He would go up to them and stick pins in them. When he stuck enough pins in you, you died. He was eventually going to come back to me but he was going to leave me until the end and get all my friends and relatives first. I couldn't stop him from doing it.

"Waiting for Godot" One of the most psychologically unacceptable notions which confronts the individual at risk is to be the passive victim of a totally random genetic accident. One 28-year-old woman described being at risk as "playing Russian roulette with a two-barreled gun and somebody else's hand on the trigger." Subjects perceived their lives and their universes as conforming to the laws of cause and effect; true randomness was either unacceptable or unassimilatable on more than an intellectual level.

Surely they are not unique in this way of thinking. In general parlance we speak of "chance" in the context of being "lucky" or "unlucky" -- a personal attribute which, as it were, controls chance and mitigates against randomness. When an individual has been the victim of a violent crime, others often respond with accusations instead of sympathy. The victim is considered to have covertly incited the crime through some careless or inappropriate behavior. Even the victim often feels ashamed and self-

recriminatory. The advantage to this way of thinking is that the crime can thus be attributed to a specific action that then can be avoided by others in the future. If disaster, either natural or man-made, is truly random, then we are all and at all times vulnerable. Some, like Mr. M, manage their fears by turning to a higher order of control and explanation in the medium of religion. God can be influenced through prayer and good deeds. If He should choose to inflict the disease, then it is not randomly assigned but made meaningful through God's will.

On an unconscious level, many of the at-risk respondents view the transmission of HD in the context of crime and punishment. This feeling is especially fostered in families where the illness is regarded as a family curse and not discussed. To them, it is truly that "the sins of the fathers are visited upon the sons." Sometimes the "crime" stems from anger toward the ill parent or from the forbidden wish that another relative inherit the disease instead. A few subjects communicated a Kafkaesque sense of bewilderment at their feelings of guilt. A 20-year-old girl stated this explicitly;

It seems that so many things have gone bad in my life that with this HD thing, well, it almost seems like I must have done something wrong somewhere to deserve all this.

If an at-risk person is violent, moody, suspicious, jealous, or disagreeable, it is considered as an ominous sign that the disease is developing. The disease is sometimes considered as just punishment for these unpleasant traits. At-risk individuals often develop complicated and constant systems of monitoring themselves in mood and movement. They continually check their hands, gait, memories, and emotions, not really to identify the disorder if it occurs, but in order to exert control over what may be happening to them. Sometimes I hold my hands out to see if my fingers wiggle . . . I just hope they aren't shaking and I can control them. Usually I'm really cool about it. It just seems like maybe tense people are nervous people and it looks like a mild case of it. They get scared that way . . . I guess, maybe, I always try to walk straight with a good posture. I try to see if I shake. I don't know what it'd feel like. Sometimes I can feel my heart beating in my body. I imagine everybody gets that. Sometimes my heart will beat so strong I can feel my fingers move with each beat. I guess maybe that I watch my face . . . Sometimes when people are waiting they'll cross and uncross their legs and shake them. I try not to do that and hold perfectly still [male, age 24].

Shortly after learning of his at risk status, one artist in his mid-20s wrote a play in which the hero was also at risk. This fictional character would stand in front of the mirror for hours practicing facial grimaces, rehearsing, and thus accustoming himself through practice with the person he might become. Through this double distancing of hero- within-a-play, the author was trying to gain active mastery over the disease. He would go out and claim it, as it were, rather than passively wait for the illness to overtake him. This same man said rather ruefully that he envied his wife "her right to be clumsy." His use of the word "right" implicitly suggests that he considers himself to be deprived of his "rights," that is, to be punished.

Considering that most at-risk individuals were most frightened by the psychological aspects of HD, it is not surprising that many tried to influence the course and even the onset of the disease through the use of their minds. Almost every subject spoke of staving off the effects of the disease through "strength of will."

My mother would say, "With your heritage, I wouldn't get mad like that." In other words, don't get mad 'cause you'll go crazy. She would say things like: "Watch out . . . don't do this or don't do that, don't feel, don't trust people, they'll leave you." The people that got left -- they all got the disease. I got messages like: you have no right to your feelings. You can't feel angry. If you feel scared you'd better hide it. Don't ever cry. All of them had to do with the disease. Those were all heavy, strict injunctions on feelings. My mother's injunctions were that if you control your feelings well enough, be in charge of yourself at all times, then you can control your mind and you won't get this [woman, age 23].

The "power of positive thought" is a relief from the terrifying helplessness of passivity for many at risk. Its liability, however, is that the individual becomes responsible for the presence or absence of the disease. To develop the illness means failure of control or failure of faith.

I've made my prayers and asked to not get it. I have strong faith but it's not that strong in that area. I have really strong faith. I can ask a prayer and sit there and wait. But this thing is so powerful that I need a stronger faith. Maybe, eventually, I will get that. Faith might save me. You can ask a prayer but if you haven't got faith, then it won't get answered [woman, age 36].

Subjective Prediction: Second Guessing the Unknown through Magic

Although the laws of probability predict that approximately half the individuals will eventually develop this disease (disregarding sampling bias possibly introduced through the use of volunteers), three-quarters of the respondents felt certain that they would become afflicted. Nearly all those who felt that they would not develop HD as well as those who thought they would, expressed magical and highly unrealistic reasons to support their beliefs. Only a very few of those who were convinced that they would remain healthy gave as evidence that they were in their mid-30s and had been symptom-free on repeated neurological check-ups. One woman in this latter group was afraid to put her optimism into words, however, for fear it would be "bad luck." Others in the group who felt they had "escaped" gave far less reasonable explanations.

Typical of their responses was that of one man who was sure he would escape the disease because he had "always been lucky." He also considered himself to be somewhat psychic. (This man had been told by his older sister that every third child manifested HD; naturally, he was the third child.)

Those who felt certain they did harbor the defective gene had equally magical and unrealistic explanations: they had always been unlucky at lotteries, or everything else bad had happened in their lives. One woman felt that "the genes would have been stronger in the first conception" and that as the eldest she would certainly get it. A rather more powerful and subtle argument was that they looked or acted like their affected parents, that is, were nervous, moody, irritable, etc. Probably the most common dynamic operating in those who felt convinced they would develop HD was the attempt to combat the passivity of waiting through active control. As part of that activity there was also a frequently implied magical belief that if they "sacrificed" themselves to the disease, they would be rewarded by being spared. Like Abraham with Isaac, their devotion to their siblings, their humility, their unselfishness, and their willingness to suffer was being tested and at the last moment they would be reprieved. On an unconscious level, genetic randomness is seen as mediated by a moral universe. Consciously, many subjects expressed the feeling that if they expected and prepared for the worst they could only be surprised by something positive.

Predictive Tests

All of the subjects firmly believed that a predictive test should be developed, regardless of their own hesitations to utilize it. Approximately two-thirds of the subjects said, with varying degrees of conviction, that they would take a predictive test. Reactions ranged from thoughtful realism to bravado; all acknowledged that they would be terrified to avail themselves of the test. Some subjects responded as if they felt they ought to want to know, while others were adamant about the importance of being able to plan realistically for their future. For the latter, the ambiguities of limbo were psychologically more difficult to bear than the certain knowledge that they were carrying the HD gene. All suggested that counseling be made available with the predictive test. In this way subjects and their families could be aided in coping with emotional reactions and other repercussions both before and after taking the test.

Those who would not take a predictive test were vociferous in their wish not to know their genetic inheritance. Some said they would take a test only if there were successful ways to treating the disease. They clung to their 50-50 chances and would not want to risk losing them. One 23-year-old woman best expressed the feelings of many at risk, as follows:

Q: If there was an accurate predictive test available, would you take it?

A: Really, no, but knowing myself I probably would.

Q: Why?

A: For the fact that it's a step forward. For the fact that if it's a crisis, God, get it over with. I'm so tired of wondering. If they would tell me that I wasn't going to get it, they could take my arm off! What if they do tell you you've got HD, how do you live with that? Like, if they were to say to me today, "You're going to get HD when you are 30," do you know what every day would be like? Every day would not be a real life . . . I just couldn't live with that. Now, at least I have a 50-50 chance; knowing and not knowing. I can live with that. Now, I have optimism. Then it would be real.

Q: Do you think scientists ought to develop such a predictive test?

A: It would be good for science to have a predictive test but it wouldn't be good for the HD victim. Can you imagine knowing that there's some place you can just walk into? Every day, that would prod your mind, "I'm going. I'm going. Just for the sake of science, I'm going. For the sake of knowing, I'm going. I can't stand it anymore." Then all of a sudden, having somebody tell you you're going to have HD. I think this would prey on their minds and everybody would probably go. But they really wouldn't want to.

Family Planning and Prediction

One-fourth of the at-risk individuals interviewed had children after learning of the hereditary nature of the disease. Most subjects had already established families before learning of the genetic nature of their parent's illness. Of this latter group, the majority claimed that had they known, they would have chosen to adopt children. Many of those married without children, however, wanted their own. There were indications that several in the group with children had known at some level that the disease was hereditary, but did not acknowledge this awareness until after their children were born.

When we were kids, we didn't know what was wrong with them but we knew all three of them were alike. They walked alike. They smoked alike. They moved alike. We promised each other that if we ever got like that we were going to come and kill each other. We knew it was something bad [woman, age 28].

Only two childless subjects, a 24-year-old male and a 25-year-old female, had been sterilized; others were sterilized after creating a family. Over half of the single people at risk had decided not to have their own biological children, unless this decision prevented them from marrying the person of their choice. Many in this group were deeply concerned about the impact their decision not to bear children would have on their marriageability. They felt that being at risk in itself made them defective and denying their future spouse natural children made them even less desirable as marriage partners. Yet to be single and forced to cope alone with HD was a prospect filled with horror. An exceptionally attractive 22-year-old woman articulated one of the primary concerns of persons at risk.

Q: Do you think that maybe you turn away some of the men who are interested in you?

A: Well, I have had this feeling. When I meet somebody who's super intelligent, super good-looking -- this guy should have kids; this guy should propagate his kind. This guy wouldn't want to adopt. I do think about that. And having the guy smarter than me is an absolute requirement and that's something that I'm not willing to compromise. But having a guy not too good looking is one thing I

even search for. Number one, because then he won't feel like, oh, I'm so good looking I've got to have a whole bunch of me's running all over the world. And also, I feel like I'd be able to hold him easier, be able to trap him easier. Which is kind of a whole bad syndrome that I'm in . . . I think, wow, since I'm at risk, I'm less attractive as a possible mate, so I've got to compromise somewhere, give up something of my desires for a mate. So, I'm not willing to give up personality or strength or the intellectual capacity, so the one thing that's left is looks. So I'm sort of hunting for some ugly guy (laugh). I think the worst part is that no one will marry me and I'll have to be alone. I guess it's the fear of having HD and being alone at that point, and being abandoned by all my so-called friends. But that could happen with a spouse, too. I guess that's why having a sister is a real comfort, because I don't think she'd abandon me.

To marry and have children means to these individuals to lead a normal life. It strengthens the normal denial that anything may be wrong. If people at risk choose to forego having children or choose to adopt, they are acting as if the illness were a certainty. The woman who had a tubal ligation rather than risk passing on the gene felt she was "damned if she did and damned if she didn't." If she tried to accept the fact that she had been sterilized, it meant to her that she had HD; if she thought that she might remain healthy, she could not bear the thought that she had had herself sterilized for no reason. It was extremely hard for her to act in one circumstance as if she would have HD and take appropriate precautions and still maintain the belief that she could as likely be well. Childless subjects feel that they are faced with the choice of guilt or self-deprivation. To alter one's life to the extent of foregoing natural children means to acknowledge genuinely the reality of being at risk. Many genetic counselors report being surprised and disappointed by how many at-risk individuals have children. These counselors fail to realize the symbolic and magical significance of the child as an insurance of the parent's continuing health. It is also true that many at-risk persons are being asked to give up one "route to immortality" through their children at the same time that they are coping with their own potential death. If genetic counseling involved more in-depth counseling over a greater length of time, perhaps more at-risk couples would choose to adopt. The fact that many adoption agencies consider a person at risk not a suitable parent creates additional complications and emotional stresses.

Learning to Live in Limbo

There is an existentialist maxim that one cannot be really free until one has come to terms with one's own death. In their own words and through their actions, many of these at-risk individuals expressed this feeling: "If I can't live quantity, then I'll have lived quality." For some, of course, the potentiality of a reduced life span became translated into a constriction of their current lives and an unwillingness to take risks.

We're always talking new house. I don't know if it's good or bad but I always turn him off. In my opinion, we're living comfortably now and I can't see moving into a new house. I'm afraid to take a step into the future. I just can't talk about the future because I don't really believe in the future. I'm just living now. When somebody talks of the future, I just turn myself off. I don't believe I'm going to be part of it. If it's going to happen, I don't know how because I'm afraid to make a move [woman, age 28].

Most of the subjects were not as reluctant as this woman to plan for the dream of the future. All, however, shared her feelings of urgency, her emphasis on living for the here and now.

I'd say I started feeling an urgency to live; do everything right now and not wait for everything. I think I lived in the future a lot. It made me feel more pushed to finish school. That was always a dream of mine. None of my relatives had ever been to college [woman, age 33].

I mean, it's always in the back of your mind. The least little back-slide you have, you think about it. That's why I get depressed sometimes. If something doesn't turn out as I expect, I think maybe there won't be another chance. I don't think that very often. Sometimes I do [woman, age 36]. I feel there is no way I can escape it. I have to make my mark by a certain period of time. I have to do something very important with my life and not waste it. There has always been a sense of urgency in absolutely everything I've done [man, age 30]. There are times when I think about it but not when I'm dancing. I think that I love to dance so much. I dance every day -- I just dance. I think about this -- what am I going to do if I do have it? Because I really love to dance. I love to just be moving around. I've just kind of felt that I've got to get it all out of my system. If I can just dance now while I can and try everything I want to try [woman, age 25].

Many of the respondents felt that they had gained an enriched perspective on life in living more for the here and now. They questioned their previous values and felt more able to concentrate their energies on activities and relationships that were meaningful to them. An intensified wish to "make a mark early in life" often led to creative and productive work and, in fact, an increased willingness to take risks. Five of the women interviewed had gone back to school or had taken jobs which brought them a great deal of pleasure and pride. All of them claimed that the courage to make these changes in their lives stemmed directly from the knowledge that they were at risk. None of those interviewed wished to squander their lives but rather, in their own fashion, each voiced the desire to "see life whole and see it clear."

Psychotherapeutic Suggestions

Almost every respondent in this study could benefit from short- or long- term counseling focused on coping with HD in the family. When at-risk persons first learn of their own risk or when they come for genetic counseling, there is often so much substantive information to be imparted that there is not enough time for discussing emotional reactions. There is also an initial shock that shields against problems that arise later. None of the individuals interviewed had had any counseling other than that provided sporadically and on a volunteer basis by the Hereditary Disease Foundation, the Committee to Combat Huntington's Disease, or the National Huntington's Disease Association. For most, their only contact with a knowledgeable professional was their parents' physician. Genetic counselors, they felt, focus only on issues of procreation and are not available over a long period to discuss problems as they change over time. Given the mobility of our society, perhaps families should be encouraged to form an institutional alliance, a transference to an informed and responsive genetic or psychological clinic rather than only to an individual.

Based on my experience working with persons at risk for HD the following counseling suggestions are offered. Although they were developed with a specific population in mind, it is hoped that they will be relevant to many counseling situations in which a genetic illness is involved.

1. Listen. Many at-risk individuals find that their spouses, their immediate family, or their relatives are too involved, too frightened or too guilty to really listen. Most persons at risk do not want to frighten their families with their concerns. They also especially do not want other family members to watch them for symptoms.
2. Do not minimize the gravity of their concern but offer realistic hope. Because of their own difficulties in coping with the risk situation, family members often brush aside the at-risk person's concerns, scoff at them, or offer magical-omnipotent solutions. Spouses are notorious for such statements of denial as: "Don't worry, honey, I won't let you get it," or "It can't happen to us." Although optimism is a must, it can also be frightening to the at-risk person to feel that the spouse cannot afford to think it could happen to them. It means that the disease is truly too terrible to think about. Frequently the spouse has a realistic appraisal of the situation but a conspiracy of silence regarding

the disease grows between the couple because each does not want to frighten the other. A counselor can be extremely helpful in guiding the individual or the couple toward a realistic appraisal of the disorder, acknowledging the reality of their concerns, giving hope, and thereby demonstrating that the illness can be reasonably discussed without anybody coming to grief.

3. The fact that anyone might die suddenly in an accident is not effective consolation. Many people will try cheering persons at risk with statements like, "Don't worry about it, you could step off the curb and get hit by a truck." It is true, but not truly helpful. Most people at risk are concerned about the process of dying, not with death itself. It is more valuable to stress the quality of life, both in health and sickness. In this case getting HD is only a 50% risk, but many at risk make themselves 100% miserable worrying while they are healthy. Much of their apprehension concerns the treatment they will receive should they become ill. Many have retained childhood visions of a "lunatic" parent, strapped in bed, with no medication. The counselor should emphasize the new drugs which are now available, new health care insurance which is pending, better nursing facilities, increased awareness of HD in the medical community, and the efficacy of physical and psychological therapy in staving off some of the most frightening symptomatology. If the state of being ill is seen as less frightening, anxiety will decrease. Remind individuals that a 50/50 probability means as great a chance that they will not get HD as that they will. The likelihood that the disease will appear also begins to decline after the 40s.

4. The counselor should remember that the state of being at risk is qualitatively different from the state of knowing definitively either that one will be sick or healthy. The ambiguous condition of 50% risk is extremely difficult to maintain in one's mind, if not impossible. In practice, a 50-50 risk translates to a 100% certainty that one will or will not develop the disease, but the certainty changes from one to the other from moment to moment, day to day, month to month. It can be helpful to discuss this phenomenon with counselees so that they know that fluctuations in their convictions are a normal part of the coping process.

5. "Symptom Searching." Every at-risk individual is continually on the alert for any suspicious signs of the disorder. Even if they deny that they check themselves if asked directly, many will give examples in the course of general conversation of such self-diagnoses. Every time an at-risk person trips, stumbles, mumbles, falls, forgets, has a car accident, gets enraged, or gets divorced, the specter of HD is aroused for themselves and for others. Many are so hyperalert that they make themselves uncoordinated, frightening themselves even more. There are at-risk individuals who practice walking on lines, walking on curbs, controlling their handwriting, controlling their speech, touching their fingers to their noses, and even rehearsing Serial Sevens! Others practice how it would be to have HD. Occasionally they frighten themselves by not knowing when the practice stops and the real thing begins. They try to master the disease through activity in the same way that people who very much fear "going crazy" play "being crazy." Some at risk will imitate mannerisms of the affected parent as an identification with that parent. It can be extremely reassuring to explain that all who are at risk "symptom-seek" and that most feel convinced that they will develop the disease. The counselor should teach the individual about psychological defenses and how they may be operating, as well as about normal muscular tics and twitches such as myoclonic bursts and normal psychological lapses, including especially normal forgetfulness.

6. Differentiate HD from the rest of the environment. Freud once said that if you cordon off one portion of a city and tell the police they cannot enter, you can be sure where to find all the criminals. The prospect of having HD can feed into every conflict; and each problem can be interpreted in terms of HD, rendering it relatively hopeless in the eyes of the individual. For some it may be easier to lay the blame with HD rather than face vulnerabilities, failures, or weaknesses that have nothing to do with the disease. Work with the individual to differentiate realistic concerns regarding the illness from

fantasied concerns and from conflicts which are unrelated. Most problems stem from the usually disrupted environments in which these people have been raised.

7. Relieve guilt. The counselor should be attuned to any expressions of conscious or unconscious guilt on the part of the client. Often the guilt is pervasive and extends both toward the parents and toward the children. Guilt may be over anger toward and neglect of the parent, over envy of an obviously well sibling, over the desire to bear one's own children, and so forth. In particular, the counselor should try to make explicit the common belief that good or bad behavior will have an influencing effect on whether or not the individual develops the disease. Often the environment conspires with this belief: A 12-year-old girl at risk was told by a police matron that she had "better behave or she would get what her mother had."

8. Just as prediction is usually foremost in the minds of the at-risk individual, it is often foremost in the mind of the counselor: Will this person develop HD? In my opinion, most HD patients do not get diagnosed until approximately 3 to 10 years or more after the initial manifestations of the disorder. This does not mean that they are unaware of the disease prior to the diagnosis. A well-trained observer may be able to detect subtle neurological, cognitive, and psychological cues long before the person with HD feels it necessary (or is pushed) to be diagnosed. What have been thought in the past to be socio-psychological indicators of neurological pathology may be, in fact, indicative of a psychological reaction to a perceived change in performance, but should never be taken as sufficient indications of the disease in and of themselves. Extreme pain, anger, and/or an ill-advised decision to have children can result from an inaccurate prediction of future events and counselors should avoid speculation, even if they are optimistic. If, on the other hand, the counselor feels convinced that the individual is not manifesting any signs of the illness at that moment in time, it can be very encouraging for the client to hear this opinion. Above all, the counselor should be empathic and respond to the concerns of the client as they are expressed