class 9

Fighting Dragons: The Construction of Explanatory Systems in Genetic Disease

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This article reports the authors' observations on coping strategies used by families who are at risk for or who have a family member showing early signs of Huntington's disease, a neurodegenerative genetic disease. The nature of the disease in its biopsychosocial context and how its diagnosis is approached by the medical community is discussed in relation to a family's construction of reality. By attributing HD-related signs to other causes, families are able to maintain their present orientation and delay having to deal with the difficult psychological, financial, and social problems associated with caring for the affected family member. The need to construct an explanation of why this disease attacks is examined in terms of its protective functions for the individual and the family. In light of the recent identification of a genetic marker that can identify an HD gene carrier, the role of genetics has particular significance for families at risk for Huntington's disease. The article discusses the implications of a family's explanatory system for genetic screening and counseling.

INTRODUCTION

Throughout history people have explained the unexplainable by using what they believe to be true, what they would like to be true, and what, true or not, brings them some inner peace. These explanations, shaped and transmitted down through the generations, develop particular meanings for the people who use them, and they form part of an individual or a group's explanatory system (13). The passing on of this knowledge is an important

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process, as it is often isomorphic with interactional patterns utilized under stress, which says something about how people cope with the unknown.

Becoming ill is one experience that often demands explanation. It is not surprising that scientific developments to the contrary, we continue to utilize multiple sources of beliefs in an effort to understand why a particular disease strikes.

The construction of an explanation to achieve psychic relief becomes subject to clinical judgment and defined as a problem when it gets in the way of treatment. Often labeled as "denial," it is possible that a range of unorthodox, unusual, or scientifically unfounded explanations are helpful in increasing life satisfaction, level of functioning, and longevity (7, 8, 9, 11, 14, 19, 22, 23, 24), even when they may inhibit or prevent medical treatment. Norman Cousins, in a thoughtful essay on denial, questions the applicability of the term in certain cases where what appeared to be denial of a serious medical problem or even noncompliance with treatment, was closely connected to the patient's effort to explore other options. He comments: "To the extent that a positive or hopeful attitude was behind the efforts, treatment may actually be enhanced" (7, p. 210). In fact, the medical literature is replete with studies beginning to consider the beneficial aspects of denial. It is not, however, our intent to review them here.

Rather, it seems clinically important to be able to assess the nature and function of a patient's explanatory system, as it may inform us about conflict-resolution patterns and interpersonal processes currently operating in the patient's life. We begin by exploring the construction process as it relates to coping with genetic disease.

GENETICS AND THE NEW UNCERTAINTY

For those involved in clinical research and disease prevention, the increasing role of genetics is among the more exciting technological advances. Molecular breakthroughs have located genes responsible for alterations that result in a disease process. At the same time, the ability to identify a carrier of a defective gene creates a whole new set of problems because of implications that we will be able to identify either a person's predisposition to a disease, or the certainty of future affliction (2). Of particular concern are the breakthroughs as they relate to diseases with an onset that is not immediate, whose course is degenerative, and for which we currently have no cure.

It is through these diseases, whose beginnings and endings are so frightening and powerful, that we can begin to unravel how explanatory systems work, their transmission over time, and their impact on patient functioning. Let us discuss how one disease, Huntington's disease (HD), lends itself to the construction of alternative explanations, with the realization that in the near future, we are also likely to be faced with the capacity to predict who will develop other similarly debilitating illnesses, such as Alzheimer's disease, affective disorders, and cancer, all of which have genetic implications.

HUNTINGTON'S DISEASE AS A MODEL

Huntington's disease is a frightening and devastating illness. One needs only to see a single patient with HD to be sobered by the thought of what it would be like to live with the prospect or the reality of the disease.

Huntington's disease is a neurodegenerative disorder that is characterized by progressive dementia, psychiatric disturbance, and choreiform movements (4). The reported mean age at onset of symptoms ranges from 33.8 years (3) to 51.59 years (15), although cases have been reported with onsets as early as four years and as late as 65 years of age (16). Reported estimates of life expectancy after onset range from 10.6 years to 15.8 years (1). Huntington's disease is inherited in an autosomal-dominant mode, so each child of a parent who has HD has a 50 percent chance of becoming affected himself, regardless of sex.

As a disease complex, HD involves the intertwining of biological, psychological, and social systems. At the biological level, HD is a disorder that includes alterations in many biochemical parameters (12). At the psychological level, it involves the individual patient's adaptation to degenerative disease and his or her perception of self. At the social level, it involves the patient's relationships with his family and the complementary social systems (family, social networks, and institutions), all of which must deal with the disruptive effects of the illness as well as the care of the patient (17).

The deteriorating course of HD is fairly predictable. Shoulson divides the course into five stages, each with increasing functional incapacitation. Each stage brings its own set of psychosocial crises for the patient and family, and include when the diagnosis is rendered, when driving must be eliminated, when the patient can no longer walk, and when the family can no longer manage with home care and must face institutionalization (20).

Treatment of HD is palliative at best. There are medications that can reduce the involuntary movements and treat psychiatric symptoms with some effectiveness (6). Until such time as the gene is actually isolated and sequenced, however, we are not likely to understand the basic defect well enough to completely prevent the disease or to eliminate the symptoms once they have begun.

Recently, scientists identified a DNA marker that is genetically linked to the HD gene (10). This discovery has led to a predictive test and serious consideration of the implications of screening of HD family members who want to know if they carry the gene (25).

THE COMPONENT OF DIAGNOSTIC AMBIGUITY

The diagnosis of Huntington's disease is difficult. Autopsy-confirmed pathology in a family member is helpful, but in the absence of a positive family history, the diagnosis becomes one of ruling out other clinical causes of

chorea, dementia, and/or psychosis until such time as a CT scan can show the more specific caudate nucleus degeneration (18). Huntington's disease is so rare that a general physician may practice for a lifetime and never see a single case.

Historically, the medical diagnosis of HD reflected a larger societal confusion about the disease and misinterpretation of symptoms. Huntington's disease is still confused with Saint Vitus' dance, a reference to the characteristic choreiform movements of the afflicted. In our experience with HD families, we frequently find a recurrent uncertainty as to what deceased relatives who probably had HD are reported by family members to have had. This uncertainty has been complicated, not only by the lack of a definitive test, but also by the overlay of symptoms or illnesses associated with, or secondary to HD, such as psychiatric disturbances and alcoholism (5). In taking family histories of individuals at risk for HD, we often find a parent, grandparent, or relative who was considered "strange," who "fell a lot," who had "bad nerves" or a "nervous breakdown," or who was an alcoholic, but who, despite showing signs and symptoms of HD, was never diagnosed.

The absence of a medical diagnosis allows one to construct and to perpetuate what Helm Stierlin refers to as a "soft" relational reality:

> The soft relational reality depends more on subjective perceptions, interpretations, emotions, and fantasies than the "hard" reality. Thus, it encompasses the opinions, expectations which individuals, family, friends, and colleagues openly or covertly hold with regard to the individual, or the loyalty which ties the individual to certain people or groups or the desires, fantasies, and prohibitions which determine the individual's conduct (21, p. 45).

Stierlin points to the diagnosis as the critical point of differentiating the "soft" reality from "hard" reality. When the family settles on an explanation that is sanctioned by an authority, Stierlin maintains that the "soft" relational reality is subsumed by a "hard" one (21).

Because of the difficulty of diagnosing HD with 100 percent certainty, there is a tendency to prolong a "softer," more subjective reality for as long as possible so the closure, the "hard" reality, which usually accompanies a diagnosis, is delayed or never occurs. In addition, the emotional investment surrounding the explanations of Huntington's disease is often profound and pervasive. In the following section we will look at the explanations of four families, each with a family member showing signs of the disease.

THE FAMILY HEIRLOOM: FOUR CASE EXAMPLES

Mrs. M

Mrs. M is a 62-year-old woman from a family with a well-documented late-onset HD. She is one of 10 siblings, five of whom seem to be affected.

Mrs. M and her husband of 25 years have three adult children, none of whom has been told about the family history of HD.

About three years ago, Mrs. M's husband requested an evaluation for his wife because her employer was asking for an explanation of her deteriorating job performance. At the time of the evaluation, Mrs. M was showing mild choreiform movements and decreasing cognitive abilities. When the subject of genetic counseling for the couple's children was brought up, Mr. M was insistent that this information be kept from them.

Most recently, Mrs. M's CT scan revealed specific caudate atrophy, and she currently presents with adventitious involuntary movements (AIMS) and memory difficulty. A specific diagnosis was given, but even as her functioning deteriorates, Mrs. M opposes discussing HD with her adult children and voices hope that her symptoms are psychological.

Mr. B

Mr. B is a 37-year-old male who has a positive family history of HD. His mother, uncle, and maternal grandmother all suffered from the disease. He has four siblings, the eldest of whom has been diagnosed as having the disorder.

Mr. B is married and has two children. About five years ago he began to complain of headaches, intermittent diplopia, upper-extremity weakness and numbness, and decreased memory. He was subsequently diagnosed as most likely having both CNS Lupus and Huntington's disease. Additionally, Mr. B, who served in Vietnam, received a workup at the Veteran's Administration Hospital for exposure to Agent Orange.

Currently, the Bs' maintain that Mr. B's exposure to Agent Orange is responsible for his symptoms.

Mr. T

Mr. T, a 43-year-old man, is symptomatic for HD and has a positive family history of the disorder. He is the youngest of three siblings and his brother and sister have been diagnosed with the disease. His mother, who was also anemic, was diagnosed with HD at the age of 50. Mr. T has four teenage children by a first marriage and has been remarried for five years.

Three years ago, Mr. T became aware of experiencing fatigue, an inability to concentrate, shaking, feelings of weakness, and mood swings. At that time he was seen by several physicians and found to have anemia. He was also evaluated by a neurologist who told him that he probably had HD.

Currently, Mr. T watches his food intake and takes naps when he gets tired. The couple rarely mentions his past medical evaluations, his family history of HD, or any connection between his symptoms and his siblings' symptoms. They do not discuss HD with their children. Both he and his wife believe that his problems are attributable to anemia.

Mrs. C

Mrs. C, a 51-year-old woman, has a well-documented family history and is symptomatic for HD. She has two older living sisters, one of whom has HD. Her grandfather and father were also affected with the disease. The Cs have two teenage children. The Cs have a history of marital and family problems, including emotional and behavioral disturbances among all family members.

Around eight years ago, the family sought a clinical evaluation for Mrs. C. Mr. C noted signs of depression, subtle loss of cognition, unsteady gait, and slight movements of the hands and fingers. Also, the family noticed that Mrs. C had clear aggravation of her symptomatology when she was in the company of her affected sister. The possibility of a conversion reaction was entertained at that time by one of the clinicians who evaluated her.

Currently, Mrs. C presents a neurological and neuropsychological picture of HD. The CT scan, however, remains inconclusive. Despite being told by the neurologist that a diagnosis of HD is "virtually" certain, the family holds to the earlier explanation of a conversion reaction, saying that as long as the CT scane is inconclusive, they remain unconvinced.

DISCUSSION

Families develop explanations of illness to bring some measure of certainty to the unknown, the unexplainable, and the unjust. Constructing the explanation itself includes interpreting and organizing past medical, individual, familial, and social information to explain the past and present and to predict the future (17).

In our four case examples, each family has chosen another label to explain symptoms commonly associated with HD. The patients had all shown various HD-related signs and symptoms, such as AIMS, memory loss, and psychiatric disturbance, yet all symptoms were considered by one or more family members to be of uncertain origin. Alternative explanations were based either on the interpretation of similar behavior in the patient's family of origin, or on competing or alternative diagnoses. We saw these explanations holding firm in spite of strong positive family histories and mounting clinical evidence consistent with the course of HD. The fact that the unmitigated certainty of the diagnosis had not been communicated to the families by a physician appears to contribute to the maintenance of the family's reality.

One could look at the function of the family's explanatory system by beginning with the disease itself. By labeling the symptoms as psychological, biochemical, or occupational, the family attempts to protect the individual and themselves from the horror associated with the disease and the possible guilt surrounding its transmission. The explanations may also help the family maintain its present orientation and delay having to envision the family

member in an unattractive, even grotesque state. Explanations also postpone having to deal with the financial and logistical problems associated with caring for a person with HD. Also, it is psychologically easier to blame an external event or agent for causing this misfortune than to accept the bewildering genetic transmission process that has brought on this "injustice." Thus, a temporary escape from the realities of HD is provided through an alternative focus or attribution of blame.

By uniting around an explanation, the family also derives support from each other, even though it may intensify an "us against them" identity. The family's adherence to its explanation promotes this group identity, and at the same time conveys to unaffected family members some values associated with why disease attacks. This may be, for example, because the person is morally weak, or a victim of the political system, or due to physical or psychological vulnerability. An unspoken fantasy may exist in which family members avoid certain behaviors or habits of the affected member, such as the expression of anger or drinking, as part of an effort to avoid developing these symptoms.

The family's interpretation of the symptoms also supports patterns of interaction and of conflict resolution or avoidance. The maintenance of alternative explanations in the T and M families, for example, functions to support the couples' reluctance to discuss HD between themselves or with their children, respectively. Outreach by our staff to the adult children in these families has been opposed and blocked by the caretaker and/or the couple.

If explanations in HD families are such powerful coping tools, what effects on family coping should we be expecting in light of the presymptomatic testing? We continually observe the tenacity with which some families with a history of HD hold to their beliefs and explanations about HD symptomology. One might predict that there will be many families who will present for testing and who, like our four families, have shaped and developed explanatory systems that have been useful to them in coping with the illness. It is naive to assume that the identification of the HD-gene carrier would have equal efficacy in altering a patient's and family's explanatory system regarding HD.

The clinician would be well advised to spend time unraveling the family's history of coping with HD. This would include obtaining information about the beliefs and attitudes surrounding HD in the patient's family of origin, and about how the patient's family of origin organized to cope with affected members. In addition to screening for the presence of significant individual or family psychopathology, it would be useful to identify families in which

1) alternative explanations have been maintained, despite multiple consistent evaluations of HD in one family member;
2) there is present crossgenerational (mis)explanations of HD; and

3) more than one family member holds strongly to an alternative explanation.

Eliciting beliefs and explanations about HD can be valuable for understanding how the family unit has adapted to and copes over time with the threat of HD. It can also help identify which families may be the most vulnerable and have more to lose if their reality is directly challenged. Careful consideration should be given to assessing the impact on the family of relinquishing an explanation and the effect this would have on the family's functioning.

The moral and ethical dilemmas for individuals at risk for HD unfold and intensify with the availability of a presymptomatic test. When viewed from the "outside," the advantages, particularly for preventing the transmission of the gene to unborn generations, may seem initially straightforward. Seen from the perspective of the at-risk individual, however, the complexity involved in whether or not to present for presymptomatic testing is quite profound. A certain amount of optimism helps those at risk to maintain hope for themselves or for their adult offspring in making choices about life plans and goals, intimate relationships, and childbearing. If the identity of being at risk is altered to one of being a carrier of the HD gene, the issues of individual responsibility create a new burden. As medical advances afford new knowledge about disease processes, clinicians need to continue to broaden their understanding of the implications of scientific progress for individual and family health.

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