POLICY CHOICES AVAILABLE IN GENETIC COUNSELING FOR PEOPLE AT-RISK FOR HUNTINGTON’S DISEASE

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Staff on 94 wards in 78 Veterans Administration Medical Centers caring for Huntington’s disease patients were interviewed by telephone to determine their policy, practice, and attitude regarding counseling HD families. Considerable variability among wards within a given hospital and among different hospitals was reported. Presentation of genetic information was found to be significantly associated by statistical tests with existence of some kind of ward counseling policy, treatment on a neurology ward, active seeking-out of relatives, construction of pedigree charts, and specific staff designated to provide such counseling. Genetic discussions with families of HD patients and at-risk kindreds raise some sensitive ethical questions, notably the vigorous denial of disease in these families, the high suicide rate of those at-risk, and consideration of patients’ rights regarding privacy of medical data. A consistent genetic information-giving policy, at least among wards of each hospital and possibly among all hospitals within a large hospital system, is recommended as a logical and desirable goal.

Considerable publicity has been given to the complicated medical/ethical problems1,2 which confront hospital staff in the formation of policies concerning cessation of treatment of the terminally ill, dealing with the markedly impaired newborn (pediatric euthanasia), abortion, and specifying priorities for organ transplants. These are prominent examples of issues which foster ambivalence in treatment-team members and present difficult decisions for professional and administrative staff as they attempt to maintain a logical, ethical, and consistent stance.

Less conspicuous, but deserving attention, are the difficult choices required in counseling kindreds and patients with a lethal inherited disease. Huntington’s disease (HD) is a notable example, and one which demands special consideration because of the characteristics peculiar to this illness. Clinical onset in the 30s and 40s is beyond typical marriage and reproductive age and the 50:50 chance of occurrence reflecting autosomal dominant transmission allows for a genetic “Russian roulette”3 attitude from the asymptomatic but at-risk relatives.

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Pronounced mental and physical deterioration incapacitate the HD patient with loss of muscle control, eventually resulting in constant writhing and jerky movements; they cannot feed, dress, or bathe themselves or speak understandably. Mental changes proceed steadily with loss of attention and concentration, depression, irritability, and emotional withdrawal, occasionally accompanied by psychotic delusions and hallucinations. Misdiagnosis as schizophrenia is frequent in the early phase. Suicide is common (seven times that of the general population), and death usually results from aspiration, falls, and decreased resistance to infection about 12-15 years after onset of symptoms. There is no cure and no reliable presymptomatic test to identify potential victims.

Huntington's disease is emotionally, physically, and financially devastating to both parents and relatives that must observe that this is one of the least attractive ways to spend the last few years of your life.

In haste, we might agree that all at-risk kindreds receive full genetic counseling; they have a “right” to know. However, a minority prefer not to know their risk status but to live instead in ordinary circumstances, untroubled and content in ignorance until symptomatic onset. Here then is the dilemma. Initial signs of HD usually begin after marriage and after the disease has had an opportunity to affect the next generation. Yet, one half of the at-risk children fail to carry the lethal gene and so are spared this malady to live ordinary lives. Providing genetic information to all at-risk for HD presents difficult choices to each individual: whether to marry or not and whether to have children or not. Bad news of at-risk status may be terrifying to some who become fearful, depressed, and may attempt suicide although for half of the group, their worry is unnecessary. Mere knowledge of the possibility of being at-risk will likewise provoke anxiety, guilt, and other emotional reactions which the genetic counselor must be prepared to handle. It approaches a no-win paradigm: failure to inform at-risk teenagers allows them an ordinary life style for a few years until they marry and have children, with major problems striking half of the group in the third and fourth decades. Informing this same at-risk group, however, may markedly alter their marital and reproductive plans, and generate nagging worry until they clear the period of onset of symptoms (age 55-60 in 90% of the cases), still not too late for marriage, but rather late for reproduction.

Truly successful genetic counseling not only halts the disease but also eliminates the biological family. Despite these drawbacks, the preponderant medical opinion and practice favor giving information as a reasonable and theoretically defensible position both from genetic and patients' rights considerations. It is less clearly defined who shall provide this genetic information, when, and to whom, and if there shall be uniformity in genetic counseling policy within a particular hospital or a particular hospital system. This study is a survey of the practice and policies of providing genetic information to relatives of HD patients within the Veterans Administration System.

METHODS AND RESULTS

A survey by telephone was conducted by one of the authors (K.S.) on 78 VA Medical Centers reported by Sugg to be caring for HD patients who were