INTERDISCIPLINARY COLLABORATION

Multidisciplinary Management at Key Stages in the Huntington’s Disease Neurodegenerative Process

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Introduction

Huntington’s Disease (HD) is an adult-onset neurodegenerative disorder with significant motor, cognitive, and psychiatric manifestations. Symptoms are progressive, with no effective treatments currently available and death occurring 15-20 years after onset. HD is inherited in an autosomal dominant manner. Affected individuals display an expanded CAG trinucleotide repeat in the HD gene on chromosome 4. Given the devastating natural history of the disease and its hereditary basis, HD can present many challenges for patients, their families, and healthcare providers. In this article, we describe some of the complex issues that arise during key stages of the HD neurodegenerative process and emphasize the importance of comprehensive, interdisciplinary team management at each stage.

Predictive Testing for HD

Predictive testing for HD based on trinucleotide repeats has been available since 1993, with a less definitive linkage analysis test available in the mid-1980s. While some argue that the test can relieve uncertainty and enable planning for the future, only 4-24% of those at risk for HD go through with testing. Decisions regarding testing have many ethical, legal, and psychosocial implications and at-risk patients must find a way to navigate through these concerns. Patients must consider the psychosocial effects of declining or postponing testing, as well as the effects of undergoing testing and receiving either a positive or negative result. Moreover, the results of testing can have a significant effect on the patient’s family, who may become future caregivers or future patients due to their at-risk status. Patients also face the possibility of discrimination based on genetic information, particularly regarding eligibility for disability or life insurance and obtaining employment. While there are only anecdotal reports of genetic discrimination in Canada, the fear of discrimination remains a very powerful influence on patients and physicians.

The healthcare team at this stage takes a primarily counselling and supportive role. Genetic counselling, often done by genetic counsellors or medical geneticists, is particularly important during this time. Counselling provides patients with disease information, takes them through the implications of testing, and offers psychosocial support. Counselling must provide the patient with all necessary and relevant information, because the far-reaching consequences of testing necessitate truly informed consent for the procedure. Genetic counsellors must present information regarding HD and predictive testing in a balanced and non-directive manner, ultimately respecting the patient’s autonomy. Patients must be assured of privacy and confidentiality. In order to support the patient through their decision of whether or not to undergo testing, other services that may become involved at this stage include social work, family support and counselling, psychology, psychiatry, neurology, and family medicine. Management should be specific to the individual patient, while taking the family and cultural contexts into account.
After Undergoing Testing for HD – Living with the Results

The results of predictive testing for HD can have a profound impact on patients and their families. A Canadian study examining the psychological consequences of predictive testing found that although psychological well-being was significantly improved for the majority of participants, 6.9% experienced clinically significant adverse psychological events, including diagnosed clinical depression and suicide attempts. These adverse events occurred in individuals given positive and negative test results. The paradoxical reaction of some patients with a negative test result (noncarriers of the HD gene) has been called “survivor’s guilt” and involves psychological distress from being spared from HD. Patients receiving a positive test result (carriers of the HD gene) must come to terms with the meaning for their own health, as well as implications for their family, career, and life plans. Although fear, anxiety, and depression may be understandable responses to a positive test result, these are also possible psychiatric manifestations of HD and patients must be followed for early onset of symptoms. A positive result can also have a tremendous impact on family dynamics, with studies showing higher divorce rates among carriers than noncarriers in the 6 months following test results, and parents possibly experiencing guilt for having transferred risk status to their offspring. Thus for HD and other genetic diseases, the unit of care is often the family rather than the patient.

Following predictive testing for HD, patients are faced with new challenges that may require multidisciplinary involvement. It is clear that psychological counselling and follow-up must be available to all patients undergoing predictive testing regardless of test result, and psychiatric care may also be required at this stage. Support and education should be offered to the patient’s family and may require the help of social workers, nurses, psychologists, and therapists. Early collaboration between the family doctor and a specialist in HD can monitor for onset of disease symptoms. Physicians must also formulate a plan with the patient for future management. In addition to the formal healthcare team, patients and their families may benefit from joining a support network like the Huntington Society of Canada.

Reproductive Decisions

HD presents complex dilemmas for patients and their families with respect to reproductive decision-making. Although HD follows autosomal dominant inheritance, the penetrance of the disease varies with the extent of the CAG triplet expansion. Triplet repeats of 40 or more are fully penetrant, those 36 – 39 are thought to be variably penetrant and those less than 35 repeats do not typically manifest as disease. The expansion (and, in some cases, contraction) of the repeats during gametogenesis further complicates reproductive choices and can present a challenge for health care professionals when communicating risks.

Qualitative research has identified several difficult decisions facing patients and their healthcare teams: whether to have children at all, whether to undergo prenatal testing or pre-implantation genetic diagnosis, and whether to abort gene-positive fetuses. Many patients who wish to have children express opposition to giving birth without attempting to prevent passing on the disease; however, this concern is often superseded by patients’ fears of their illness preventing them from providing for and raising children. For this reason, patients must be counselled with consideration and potential involvement of their partners, families, and wider social support networks. This in turn can be a concern for patients, as many report the influence of others as a major factor in decision-making. This includes the influence (whether or not intentional) of the healthcare team, which is an important consideration in light of a recent study which indicated that 38% of Mexican neurologists, psychologists and psychiatrists felt that those with the HD mutation should not have offspring.

Prenatal testing for HD currently exists via amniocentesis or chorionic villus sampling. However, testing rates for those at risk for HD remain low; within the United Kingdom and
Australia only 5 – 25% of at risk populations undergo prenatal testing.\(^8\) Within Canada, a survey examining 15 of the 22 centres offering testing found that only 15 prenatal tests were completed between 1987 and 2000. The 12 families who underwent these tests were estimated to represent approximately 0.1% of the at-risk population.\(^11\) Women were shown to be more likely to request prenatal testing as well as predictive testing for themselves.\(^11\) More recently, pre-implantation genetic diagnosis has become available to ensure only mutation-negative embryos are implanted. This process, however, can be difficult for many couples and may not be an option for many due to personal beliefs and preferences.\(^12\) Pre-implantation genetic diagnosis is now also offered without informing the parents of their own HD gene status, which is preferable to some patients who desire certainty that they are not passing on the disease to their children, but do not want to determine their own gene status.

Despite these difficulties, many HD patients and carriers do opt to have children. A European study demonstrated that 14% of HD carriers had subsequent pregnancies following predictive testing, compared to 28% of non-carriers.\(^12\) Furthermore, many patients consider adoption as a potential alternative depending on their expectations of disease onset.\(^8\)

**Interdisciplinary Care of the Symptomatic HD Patient**

The management of symptomatic HD patients is a broad topic, and in its entirety is beyond the scope of this overview; instead, a brief outline of the interdisciplinary nature of HD care is presented here.

The nature of HD requires a team-based approach to symptom management. An optimal healthcare team includes leadership not only from a neurologist or psychiatrist specializing in HD, but also a family physician to monitor complications in the late stages of the disease.\(^3\) Allied health care professionals, including nurses, dentists, dieticians, physical, occupational and speech therapists, psychologists, and social workers also play major roles. Advance care planning is also extremely important in establishing quality of life goals and end-of-life care directives.

The stages of symptomatic HD dictate the need for each of these team-members. The Shoulson-Fahn scale, originally designed for research, has become useful in evaluating the severity of HD. The five stage scale is based on a Total Functional Capacity (TFC) score from 0 to 13 that assesses the patient’s functional skills and ability to carry out activities of daily living. In stages 1 through 3, the focus of care includes treatment of chorea, sleep disturbance, depression, anxiety, impulsivity, irritability and other psychiatric symptoms.\(^3,13\) In these stages the physician members of the team are foremost. However, the role of psychologists and social workers may be crucial in the transition from working life to disability status for some patients and their families. It has been suggested that one of the most prevalent deficiencies in HD treatment is disproportionate focus on minor findings of chorea, without proper management of depression and deterioration of family relationships.\(^13\) Medical therapy including anti-depressants and mood stabilizers may be of value and neuroleptics may be warranted in cases of more aggressive behaviour.\(^13\)

Mid- to late-phase HD requires follow-up visits to physicians to assess for complications, but also increased involvement of other healthcare professionals. Occupational therapists may be involved in assessing driving ability and determining necessary restrictions. Patients often have large appetites, and may have difficulty meeting nutritional requirements given increasing dysphagia and lack of coordination (aspiration and subsequent pneumonia are often the terminal events for HD patients). For this reason, dieticians' expertise and nursing or personal support worker assistance with feeding can be of great benefit. Physician follow-up may involve the reduction of medication for chorea, which paradoxically decreases in the late stages and is supplanted by rigidity and dystonia. These symptoms are often worsened by excess pharmacotherapy for chorea, which must become more judicious.\(^13\) Physicians must also help
patients and families deal with increasing cognitive impairment and mood alterations, and as such, behavioural counselling is often required. In the end stage of the disease, hospice nursing and palliative care specialists ensure patients are as comfortable as possible and assist families with the patients’ death.

Conclusion

Huntington’s disease presents significant challenges for the healthcare team. Despite the paucity of treatments available to delay disease progression, a well-coordinated, multidisciplinary team can assist patients and their families with managing the evolving nature of their illness and help them make informed decisions. Through appropriate collaboration, care can be optimized and the burden of this multi-faceted disease can be reduced.

References