

Huntington's disease out of the closet?



In her Art of Medicine essay in *The Lancet* today, Alice Wexler¹ recounts the stigma faced by those with Huntington's disease, a cruelly progressive and incurable neurodegenerative condition that usually (not invariably) presents in mid-life. The disease is inherited as an autosomal dominant trait so that children of a parent with Huntington's disease are all at risk, with each having a 50% chance of developing the disease if they live a normal lifespan. The stigma of Huntington's disease, therefore, attaches to those who are at risk as well as to those with symptoms.

As well as having profound implications for the families with a member who has Huntington's disease, the stigma has negatively affected research, particularly studies that have sought to investigate epidemiology and, most especially, prevalence. Various attempts have been made to estimate the prevalence of Huntington's disease in the USA and Europe.² The most intensive efforts, however, have been made in the UK, and we now have prevalence estimates in 15 localities.² The prevalence observed in these UK studies (expressed per 100 000 of the population) ranges from 2.5 (Essex) to 9.95 (Grampian), with an overall average of 6–7. These estimates presumably formed the basis for the assertion, in an answer to a parliamentary question, that there were 4800 cases of Huntington's disease in the UK in 2003.³

The true prevalence of Huntington's disease is unquestionably greater than that rate. The Huntington's Disease Association—a not-for-profit organisation that provides services for patients with the disease and their families in England and Wales—currently cares for 6702 people with symptoms. Specialist neurologists have referred all of them, so their diagnoses can hardly be in doubt. From these numbers alone, the minimum prevalence in England and Wales must therefore be at least 12.4 per 100 000 of the population. The Huntington's Disease Association, however, does not provide services in all areas of England and Wales; and there are an unknown number of patients with the disease who have never been referred to the Association. This prevalence estimate of 12.4 per 100 000 of the population is, therefore, unquestionably an underestimate but by how much is uncertain.

Why is the prevalence of Huntington's disease so hard to estimate? And why does it matter? There are

two reasons why it has been so difficult to estimate the prevalence. First, until fairly recently, the diagnosis was entirely based on clinical features. As a result of the discovery of the underlying genetic abnormality⁴ by a team led by Nancy Wexler—Alice Wexler's sister—other patients, with atypical neurological and psychiatric features, have now been found to have the disease. Second, and probably most importantly, the families with a member who has Huntington's disease often try, because of the stigma associated with the condition, to conceal the true nature of their familial trait even from their own general practitioners. Huntington's disease is the only genetic condition for which the insurance industry loads those at risk. The stigma—to the insurance industry's eternal shame—is not only societal but also actuarial.

Reliable estimates of the prevalence of both symptomatic Huntington's disease and of the individuals who are at risk are important for two reasons. First, the UK National Health Service (NHS) needs to ensure that there are appropriate services available now—and in the future—to care for those living with the disease and for their families. Second, combining individuals with symptomatic Huntington's disease with those at risk emphasises the importance of encouraging research that will ultimately lead to treatments that will arrest progression. Such treatments, even if only partly effective, would need to be started long before

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Members of the Sussex Branch of the UK's Huntington's Disease Association

the appearance of symptomatic disease. By the time symptoms appear, there are already substantial neuropsychiatric changes,⁵ and prophylactic therapy in those at risk, and who carry the genetic abnormality, would be essential even though at present we do not know when treatment might best be started.

To promote greater understanding and awareness of Huntington's disease, an All Party Parliamentary Group is launched on June 30, under the chairmanship of Lord Walton of Detchant. With the organisational skills of the former NBC news and war reporter Charles Sabine—who is not only himself at risk but who also carries the abnormal gene—this Group aims to eradicate stigma, promote research, and chivvy the NHS to provide the best possible care.

Michael Rawlins

London School of Hygiene and Tropical Medicine,
London WC1E 7HT, UK
michael.rawlins@nice.org.uk

I have been involved in establishing the All Party Parliamentary Group on Huntington's disease.

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