

Huntington's Disease

Who should have a genetic test for Huntington's Disease?

The genetic variation (mutation) which causes Huntington's disease (HD) is located on chromosome 4. The HD gene contains a section of DNA which varies in size from one person to another, and is longer in people who have HD than in people who do not.

The laboratory can measure the size of this DNA segment by analysing DNA prepared from a blood sample from the patient. The genetic test is usually very reliable - most people get a result which is either clearly normal or clearly abnormal. However, a small number of people fall into a 'grey area' between the two. In this intermediate range, the size of the DNA fragment is not the only factor which decides whether the disease appears, since some people develop symptoms, while others with an identical fragment size do not. This 'other' influence is probably genetic rather than environmental as affected identical twins have very similar ages-of-onset and clinical symptoms. Additionally, in future generations, DNA fragments in the intermediate range may increase in size. Generally, the larger it is, the more likely it is to expand. With time, this may lead to the age-of-onset of the disease becoming earlier and some (but not all) symptoms more severe.



Jean lives in North London. Her husband Peter died two years ago, aged 54, from Huntington's disease (HD), a hereditary disorder that affects the brain and nervous system.

Peter first developed symptoms of HD when he was in his late twenties. His employers advised him to visit the doctor, and he was diagnosed with HD six months later:

'They said he was acting strangely - they sent him to his GP, and he thought it was some sort of a mental illness, so they sent him to a psychiatrist...but the psychiatrist eventually requested he went to the neurology hospital...he went there, and that's where he was diagnosed...We didn't really understand what it was about, never knowing anyone in the family with it... His dad died when my husband was 11, so he may have had it, we don't know.'

After Peter was diagnosed with HD, Jean contacted the Huntington's Disease Association, a support group for families affected by the illness. Although there is no effective cure for HD, Jean says that finding out about her husband's condition helped her to understand and deal with it:

'It makes you find out about what's going on. They can't exactly say how long the lifespan will be because it's a very progressive illness, but you get all the leaflets and they explain everything that's going to happen, so you're expecting that. You know what's going to

happen so you try and prepare yourself for the worst...A lot of people thought he was drunk, because of his speech, and they can't control their movements, and when they walk they stagger, so obviously people think they're drunk but they're not'.

Once Peter was diagnosed as having HD, he had to leave his job, because it involved working with machinery. The family's GP put her in touch with social services, who Jean says were 'brilliant' at sorting out benefits and help. As the disease progressed, it changed their lives completely:

'We used to have a caravan, and we had to give it up, because eventually he stopped driving...he was told he had to stop driving, so that was gone. He couldn't work, obviously, so I had to be the breadwinner. Caring for Peter was down to me. I could have had a carer come in, but my husband was a bit funny, he didn't want anyone else to look after him. The only thing that the social worker got him into was respite care for two weeks, more or less every three months, so I had a break from it then'.

The couple have two children, who were offered a genetic test to find out if they had inherited HD from their father:

'There was a counsellor in the hospital where my husband was diagnosed and he said that if they wanted to, they could have it done when they were older.'

After discussing it with Jean, her daughter decided not to have a test, but her son went ahead with genetic testing, and found out that he has not inherited HD.