

Diagnosis of Huntington's Disease

How is a diagnosis of Huntington's disease made?

HD is diagnosed when:

- The person has a positive gene test for HD. With a few exceptions, you must be eighteen or over to have the test.
- The person has motor symptoms of HD (involuntary movements, problems with balance & coordination, changes in speech)

Huntington's disease affects each person differently and some people with the condition may not have motor symptoms first, but instead have cognitive (thinking processes) changes or experience mental ill health. This can make diagnosis more difficult and it's important to see a doctor who is experienced in the condition, in order to get an accurate diagnosis. You can find out where to get specialist advice about diagnosis from your local Huntington's Disease Specialist (staff employed by the SHA to give specialist support to people living with the condition), which can be found on the SHA website.

How is a diagnosis of Juvenile Huntington's disease made?

Juvenile Huntington's disease (JHD) is a rare form of HD that occurs in about 4% of people who have the gene that causes the condition. A young person is said to have HD if they have onset of symptoms before their 21st birthday.

The initial symptoms of JHD may be different from the adult form of the condition and include problems with attention and concentration (accompanying difficulties in school), changes in behaviour (e.g. problems with temper control) and slowed movements. Involuntary movements are far less common in young people.

Many of the symptoms described above, especially if they are mild, can be caused by other things. A child who is experiencing stress in school might have problems with concentration or attention or may be moodier than usual. For those reasons it is not uncommon for parents to worry that their child is showing symptoms of JHD.

If you are worried about a young person, you can contact one of our HD Specialists for advice or you can speak with a member of our Youth Team.

If there is concern about a young person having JHD the following advice may be useful.

- In the first instance talk through your concerns with a specialist.
- If there is sufficient concern the young person's GP can refer to a Paediatrician for a neurological assessment.
- If neurological changes are present, the neurological assessment should be repeated six months later. If neurological symptoms have changed, referral for the gene test for HD may be considered and this ideally should occur after a multi-disciplinary meeting with the family has taken place.

Useful links and information

More can be found out about HD on the SHA website.

hdscotland.org/what-is-hd/

Publications that may help

Huntington's Disease, The Facts, by Oliver Quarrel

The Juvenile Huntington's disease handbook