The Basics of JHD

HDYO has more information about HD available for young people, parents and professionals on our site: www.hdyo.org

This section is here to explain the basics of Juvenile Huntington’s disease (JHD). As well as provide some insight into the kind of support available for families impacted by JHD.

What is JHD?

When George Huntington first identified Huntington disease in 1872, he suggested that the disease affects adults only. It is now known that Huntington disease can occur in individuals at any age in their life - young or old. When someone who is 20 or younger develops symptoms of Huntington’s disease, they are classed as having Juvenile Huntington’s disease (JHD). So effectively ‘JHD’ means a young person with Huntington’s disease.

Why do people get JHD rather than Huntington’s disease?

Huntington’s disease is hereditary - that means that it can be passed down, from parent to child, through genes in our DNA. Genes are passed to you from your parents - that’s why you might have hair like your dad, or brown eyes like your mum. We have thousands of genes, and they are all sectioned into different segments in our DNA. These segments are known as chromosomes and in total we have 23 pairs of chromosomes. The reason we have ‘pairs’ of chromosomes is because we inherit one from each parent.

In 1993, a team of researchers discovered the gene responsible for Huntington’s disease on chromosome 4. They found that one part of the gene repeated itself over and over again, like a stutter or when you hold a key down too long on a computer keyboard. This repeat is what causes the development of Huntington’s disease and is known as a ‘CAG repeat’.

Higher CAG repeats

Everybody has two copies of the Huntington’s disease gene, whether they’re at risk for Huntington’s disease or not. One copy was inherited from each parent. And every Huntington’s disease gene contains a CAG repeat in that gene.

What causes the development of the disease is the number of times the CAG is repeated. Put simply, people who develop Huntington’s disease have longer CAG repeats than those that don’t. To make things easier to follow, we will split the CAG repeats into different ranges.

26 And lower (Unaffected range) - As we mentioned, everybody has a CAG repeat in their Huntington’s disease genes (even those not in a family affected by Huntington’s disease). Most people who don’t have Huntington’s disease have around 10-20 CAG repeats. In fact, anything up to 26 CAG repeats is fine and will not cause any problems. But, once you go above that, there may be complications.

40 Repeats and higher (disease range) - Anyone with 40 CAG repeats or above is, unfortunately, certain to develop Huntington’s disease in their lifetime, with a 50% risk of passing the gene on to each of their children as well.

Repeats in the middle range between 27 and 39 are quite rare. They’re a bit less straightforward, and in fact scientists don’t understand them fully, so they’re sometimes referred to as the ‘grey area’.

Most people that have Huntington’s disease have a CAG repeat between 40-50. For these people, it is hard to tell when their symptoms will begin. For example, two people with the same CAG repeat of 45, could get symptoms of Huntington’s disease at completely different ages, decades apart. But JHD tends to be different, people with JHD usually have higher CAG repeats - around 50+ repeats. This difference in CAG repeats results in people getting symptoms at a younger age, earlier in life.

Is JHD common?

Less than 10% of cases of Huntington’s disease come in the Juvenile form. When you consider that
Huntington’s disease itself is rare, this highlights just how rare JHD actually is.

For reasons which have only recently become clearer, most people (around 90%) with JHD inherit the gene from their father. The reason for this is that the CAG repeats tend to be less stable when passed from the male. In general, CAG repeats usually increase a little when passed from generation to generation. So after a few generations of the gene being passed on, the likelihood of getting symptoms earlier in life also increases. However, this ‘increase’ seems to be larger when the gene is passed from the male (the father), as opposed the female - where the CAG repeat usually stays around the same. It’s important to state that this is not always the case. As with anything regarding Huntington’s disease, it is very unpredictable.

Symptoms of JHD

Support for young people with JHD is vital, and their needs differ from typical adult-onset Huntington’s disease, in both the symptoms and the ways that it changes the lives of the individuals and families that it affects. JHD presents unique challenges to affected individuals, their caregivers, and the various professionals who are called upon to assist them.

Typical early symptoms of juvenile HD

- Stiffness of the legs
- Clumsiness of arms and legs
- Decline in cognitive function
- Changes in behaviour
- Seizures (In 25-30% of cases, there is a tendency to epileptic seizures - something almost never seen in adults with HD.)
- Speech problems
- Behavioural disturbances

It is thought that the progression of JHD may be quicker than that of adult-onset Huntington’s disease in some younger people with JHD, although as with adults there is always a huge variation in how someone’s condition progresses so it is difficult to know for certain how fast each person’s condition will progress.

Diagnosing JHD

Diagnosing JHD can be very difficult. As mentioned previously, JHD is extremely rare, and few physicians will have encountered the disease before. This can lead to a great deal of time being spent eliminating other possibilities. Even the best physician may have to see the child several times before being confident that the symptoms are clear. A neurologist can usually determine that the child has a disorder affecting a portion of the brain called the basal ganglia (which is affected by Huntington’s disease), but it can be difficult to distinguish Huntington’s disease from other similar diseases. It can also be difficult to be sure that early symptoms of JHD are caused by JHD, as the symptoms of JHD, (such as changes in behaviour, difficulty at school, being a bit clumsy) can have so many possible causes besides Huntington’s disease, particularly in children that are changing. So, even if someone is worried about changes in a child, it’s still important to check other things so you don’t miss something else by just assuming it has to be Huntington’s disease because the condition is in the family.

What makes things even harder for a professional to diagnose JHD is the fact that testing the child, to see if their CAG repeat was suspiciously high, would not be enough on its own for a diagnosis either. This is
because although some young people with JHD do have very high repeat counts which might be more suggestive of JHD, some people don't. The risk then is that you test for the gene and get a result that could be juvenile- or adult-onset HD. In other words, you know from the result that the young person is going to develop Huntington’s disease, but it’s still not clear whether the young person has symptoms of Huntington’s disease now, or whether they are going to develop symptoms in the future. This would then mean that a person has essentially had a predictive test without having made the decision to know their result - which goes against the rights of that person. It is not advised to perform a genetic test on someone under 18 unless absolutely sure there are symptoms of JHD. All the genetic test can really tell us is that this person will develop symptoms at some point in their life, so in diagnosing JHD it can only be part of the picture.

As a result of all these difficulties it tends to take a while for a young person to be diagnosed with JHD. This can be quite frustrating for all involved and although support can be a bit harder to find during this time, people should be able to get help and support with whatever difficulty they’re facing while they’re waiting for a diagnosis.

Support for JHD

Young people with JHD and their families need plenty of support to help them at different stages of the progression. Below you will find those HDYO considers key.

Neurologist: Having a neurologist who specialises in Huntington’s disease can be a huge help. Many doctors won’t have much experience with Huntington’s disease, let alone JHD. Having a neurologist who specialises in Huntington’s disease can bring with them a wealth of knowledge and understanding that may be vital in dealing with this disease.

Physiotherapist: Most physical therapists have never seen a case of JHD and they may assume that the affected child will relentlessly decline and that, therefore, therapy is a waste of time. But the opposite is the case.

A physiotherapy programme for children with JHD should focus on maintaining the range of motion in the joints and independent mobility. Although most children with JHD have stiffness, physical therapy and activity can help to prevent muscle contractures. So there’s real benefit to physiotherapy for those with JHD.

Occupational Therapist: As the disease progresses, more equipment and mobility support may be required to help the person with JHD. This is where an occupational therapist can help you.

Speech and Language Therapist: A speech/language therapist can help to create strategies which will help maintain speech ability for as long as possible. At the appropriate point, they may also be able to introduce new tools to facilitate on-going communication abilities.

For example, there are devices which allow the user to “speak” a word or phrase by pressing squares on the surface. Specific words and phrases can be programmed into the computer according to the child’s needs - from “I need a drink” to “I need a hug.”

A speech/language therapist can also help with swallowing difficulties when eating becomes an issue.

Dietician: And when eating does become an issue a dietician can be a real help in setting up a diet that helps maintain the child’s weight. A classic issue as a result of Huntington’s disease is the weight loss people suffer. But this can be controlled with a high-calorie diet.

HDAs: The Huntington’s Disease Associations are there to provide support to families impacted by Huntington’s disease in their region. Most nations have a HDA, and yours may be able to offer some support with JHD. Unfortunately the support available in different regions varies hugely. But there is a lot of good information on JHD available from the HDAs. Along with the neurologist, the HDA should be your first port of call. Find the contact details of your local HDA here.

JHD is a difficult issue to learn about. If anything has upset you in this section you can speak with HDYO about it. We are here to help.

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