Huntington's Disease Association

Huntington's disease:

A professional guide to Juvenile Huntington's disease

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What is Juvenile Huntington's disease?

Huntington's disease is a rare disease which is caused when somebody has a faulty gene in their DNA. If someone develops symptoms of Huntington's before the age of 20, this is known as Juvenile Huntington's disease - sometimes called JHD or Juvenile-onset Huntington's.

Juvenile Huntington's is quite rare; about 5% of people with Huntington's will have Juvenile-onset. This can make dealing with it quite an isolating experience for everyone involved, including professionals involved in that person's care.

People with Juvenile Huntington's can sometimes experience symptoms that don't feature in adult Huntington's. This is particularly the case for those who are young children when they begin to develop symptoms. They are less likely to have the involuntary movements, known as chorea. They're more likely to experience muscle contractions and stiffness, making their movements slower and more difficult. They may also develop epilepsy.

Mood changes, such as feeling frustrated or angry and finding it hard to manage behaviour are a common symptom in people who develop Huntington's in their teens. This can often be the first sign of the illness. However, many children and young people with Juvenile Huntington's do not have these problems, and it's important to remember that these symptoms may relate to other causes and not be due to Huntington's.

These are just a few of the possible symptoms, someone with Juvenile Huntington's may not experience them all, or may experience different ones. There are many different kinds of help and support available to reduce symptoms and their effect on someone's life.

The genetics of Juvenile Huntington's disease

A gene is a piece of biological information that a person inherits from their parents. They are present in every cell of the body and they tell the cells what to do. Genes control cells by producing proteins and different genes make different proteins. Chromosomes are threadlike structures which store the genes. Each human cell contains around 25,000 pairs of genes, stored on 23 pairs of chromosomes.

The gene that determines whether a person could develop Huntington's disease is stored on chromosome pair 4 and is known as the 'Huntingtin' gene. The gene provides the code for the Huntington's protein. Everyone has two copies of the gene – one inherited from each parent.

The code for the Huntington's gene contains a particular sequence known as a CAG (cytosine-adenine-guanine) repeat. Everyone has two CAG repeats – one for each copy of their Huntington's gene. If a person has Huntington's disease, this means they have inherited an expanded copy of the Huntington's gene and the recipe for the protein it produces is incorrect, or 'faulty'. If there are too many CAG repeats on one copy of the gene the protein that is produced can damage nerve cells in the brain. Every child conceived naturally to a parent who has the faulty gene has a 50% chance of inheriting the gene, and the disease.

If someone has 40 or more CAG repeats, they will definitely develop Huntington's at some point in their life. If they have more than 60, it is highly likely that they will get Juvenile Huntington's, although it is not definite. If they do develop Juvenile Huntington's, it is more likely

that the faulty gene came from their father, as CAG repeats tend to be more unstable when passed on from a man. It is thought that this is because the gene becomes more unstable in sperm.

Juvenile Huntington's is what's known as an "autosomal dominant disorder". In plain English, autosomal refers to a gene that is not on the X or Y chromosome and dominant means that the mistake in one copy of the gene dominates over the normal copy.

Diagnosis

If the child of someone with the Huntington's gene starts to show symptoms, a diagnostic blood test may take place to see if they have Juvenile Huntington's.

Because Juvenile Huntington's is so rare and the symptoms hard to recognise, they can be mistaken for something else, and this can make the actual diagnosis more difficult. Not knowing can be stressful, and it is important for the family to have support at this time.

The effects of Juvenile Huntington's disease

If someone has Huntington's disease or Juvenile Huntington's disease, it means they have a faulty version of the gene responsible for producing a protein that helps nerve cells (neurons) in certain parts of the brain to develop before birth.

The faulty version of the gene produces too many repeats of a particular piece of genetic code. This means that the protein it produces damages neurons instead of helping them to develop, causing them to function poorly and reduce in number over time. As this happens, changes occur in how the neurons function, resulting in the various symptoms of Juvenile Huntington's.

In the case of Juvenile Huntington's, the gene produces an even larger number of repeats than with adult Huntington's, resulting in symptoms appearing earlier (before the age of 20). There are also other factors which influence the age of onset, but these are not yet known.

The parts of the brain affected include the basal ganglia and cerebral cortex. These inter-connected areas are associated with different types of activity, including movement, learning, thinking, planning, motivation and emotion.

The ways that Juvenile Huntington's affects the body, and the speed at which these changes happen, are different for different people. Not everyone experiences the same symptoms, and they change over time as the disease progresses. However, changes to the body may occur faster in some young people.

Access to care that fits the person's needs and adapts to changes that they experience will make a huge difference.

Symptoms of Juvenile Huntington's disease

In some ways, symptoms of Juvenile Huntington's are similar to those of the adult disease, but there are some key differences. As with adult-onset Huntington's, Juvenile Huntington's symptoms can vary from one person to another. However mostly they affect three main areas:

- Movement
- Thinking
- Behaviour

However, children and young people affected by Huntington's are less likely to experience the involuntary movements, known as chorea, that often characterise the adult illness, and more likely to be affected by muscle contractions and stiffness. Epilepsy is also more common in Juvenile Huntington's, particularly in younger people and children.

Different types of symptoms generally occur at different stages of the illness. Often the first indication that someone has Juvenile Huntington's is a change in their thinking or behaviour. For example, they may experience difficulty concentrating and following instructions, and there may be a noticeable drop in their performance at school, college, or work. Family members and teachers may not initially interpret these changes as being linked to an illness.

Not all behavioural changes will necessarily be caused by the disease. Children of families affected by Huntington's may also be

experiencing disruption and difficulties in their home life, which could also impact their behaviour, or they could be facing other challenges in their lives.

As a person must be over 18 years old to have the genetic test for Huntington's, and because Juvenile Huntington's symptoms can resemble those of other diseases, such as autism, depression, or attention deficit disorder (ADD), the illness can be misdiagnosed or remain undiagnosed for some time. This is particularly true in cases where family history of the disease is not known.

There are many ways that children and young people living with Juvenile Huntington's can get help and support with their symptoms, in order to help them cope in school and greatly improve their quality of life.

Early stages of Juvenile Huntington's disease

Changes in behaviour or a drop in school performance are often the first noticeable symptoms of Juvenile Huntington's disease.

- It may become harder to concentrate, learn new things, follow instructions and remember things.
- It may take longer to respond to questions and perform tasks.
- It might be harder to start tasks. Sometimes this can be misinterpreted as lethargy or laziness.
- The person may start to feel more frustrated, impatient, irritable or angry than normal.
- The person may start to experience stiffness in their limbs. Their movements may slow down and they might find that they start to stumble or walk unevenly.

These are just a few of the possible symptoms – someone with Juvenile Huntington's may not experience them all or they may experience different ones. Some young people with Juvenile
Huntington's may find that their symptoms progress more rapidly than adults with Huntington's.

Many symptoms can be greatly helped with extra support at home and in school. As Juvenile Huntington's is so rare, it's important for schools to have information to help them understand the disease and how best to support the young person. For example, it may be that they need longer to eat or to have frequent snacks throughout the day to keep their weight up.

Middle stages of Juvenile Huntington's disease

Symptoms of the middle stage of Huntington's often revolve around changes in muscles and movement, although changes in behaviour may also become more significant and challenging.

- Muscles may start making involuntary contractions and may become stiff and rigid.
- A person's movements may slow down and their arms and legs may become clumsy.
- There may be changes in how the person speaks, as it may become more difficult to form words. Speech therapy and specialist equipment can help with this.
- It may become difficult to swallow, making eating difficult, and the person may also find that they lose weight. Support from a speech and language therapist and dietitian can help to make sure they keep their weight up.
- Behaviour may change as feelings of anger, frustration, or depression become more intense. This can be incredibly challenging for the young person and those around them. It doesn't happen to everyone but it's more likely to happen if symptoms started developing in the teens.

Everyone's journey through Juvenile Huntington's is unique. Not everyone will experience all these symptoms, and some people may experience them at different stages. This means that care will need to be tailored to the individual's specific needs and will need to change and evolve as they do.

Later stages of Juvenile Huntington's disease

The nature of Huntington's is such that gradually, often over a period of many years, the disease progresses until the end of life.
Later on, a person may experience difficulties with

- Weight loss and nutrition
- Speech and swallowing
- Movement and stiffness
- Communication

It is important to pay attention to the symptoms that are causing the greatest difficulty, emotionally or practically, at the time.

Sometimes the psychological and emotional sides of living with the disease are more of a problem than the physical side. It can be mentally difficult to cope with having a serious illness, and extremely frustrating to not be able to do things that could be done easily before.

There are many different kinds of support and help that someone with Juvenile Huntington's can access to manage their symptoms and live as well as possible. The person's carer and family may also need additional support, practically and emotionally. In the later stages of the disease more care and support will be needed.

Support from the Huntington's Disease Association

Working with someone with Huntington’s disease can seem quite daunting at first, there is so much to take in and understand. It will take a lot of kindness and patience to fully assess this person and you may have to rethink the ways you do things. The most important thing to remember is that they cannot adapt their behaviour to you, **you** must be the person who adapts to them. The person's family members can be an invaluable resource in the assessment process but may feel they cannot openly discuss the issues in front of the person with Huntington's for fear of upsetting them. Be tactful and flexible in your approach.

Specialist Huntington's Disease Advisers (SHDAs)

At the Huntington's Disease Association, we have a specialist advisory service operated by Specialist Huntington's Disease Advisers who have a background in health or social care and are knowledgeable about Huntington’s disease. They operate throughout England and Wales and are able to provide bespoke training to staff working with clients with Huntington's disease. They are also able to discuss individual problems, provide tailored advice and suggest ways of managing certain behaviours. To get in touch with your local Huntington's Disease Adviser, contact us at:

- 0151 331 5444
- info@hda.org.uk

Specialist Huntington's disease Youth Engagement Service (HDYES)

At the Huntington's Disease Association, we have a Youth Engagement Service (HDYES) who work with young people whose families are affected by Huntington's. This confidential service is for anyone aged 8-25 living in a family affected by the disease (including extended family such as a cousin, grandparent, etc.). If you are supporting a young person living in a family affected by Huntington's and you wish to refer them to HDYES, get in touch with your local Specialist Youth Worker at:

- 0151 331 5444
- info@hda.org.uk

Website

Our website offers practical advice and sources of help and support, including downloadable information guides. We have guides with more in depth information about particular areas of Huntington's disease such as mental illness and mental capacity and care in advanced Huntington's. It also holds information about events, webinars and national training events that you can attend:

- www.hda.org.uk

Membership

There is a £30 lifetime membership fee to become a professional member of the Huntington's Disease Association. Members receive regular eNewsletters and other communications from the charity, meaning they are among the first to hear about our work, news, events and opportunities to get involved. Members are eligible to vote at our Annual General Meeting and receive a copy of our Annual Report and Summary of Accounts. Further information about professional membership can be found on our website.

- www.hda.org.uk/get-involved/membership

Juvenile Huntington's Disease Weekend

The Huntington's Disease Association's Juvenile Huntington's Disease Family Weekend gives young people with Juvenile Huntington's and their families the unique opportunity to meet others in a similar situation, while also having a great weekend full of exciting activities. It's also a chance for families to find out more about Juvenile Huntington's, both from other parents, and healthcare professionals with expertise in Huntington's disease.

The whole family is welcome on the weekend, including the young person with Juvenile Huntington's, parents and carers, as well as brothers and sisters. Our website has a video which shows more about the weekend.

Social media and mailing list

We have a number of social media channels that we share stories, resources, events, webinars, and announcements on frequently. They offer a way for the Huntington's community to interact and connect with each other. We also have a YouTube channel with lots of videos and webinar recordings for people to catch up on anything they've missed.

- @hda_tweeting
- @hdauk
- @hdauk
- @hda_uk
- Huntington's Disease Association
- Huntington's Disease Association

We also regularly send out information about the charity, events and updates by email to those signed up to our mailing list. To join, visit our website:

- www.hda.org.uk/get-involved/join-our-mailing-list

Support from other organisations

Getting the right information and support is vital, and the Huntington's Disease Association is here to help.

Supporting a young person with Juvenile Huntington's and their family can feel daunting. Because the disease is very rare, it is likely that most health professionals or schools won't be aware of it or knowledgeable about it. Families often say they would prefer professionals to be honest about how much (or how little) they know, but to listen to them and to try and help them get the support they need. Families often build up a lot of expertise and knowledge, but the responsibility of this can be a burden, and overwhelming for them at times.

Key professionals

Types of specialists that may be involved:

- **Neurologist** - A neurologist is a doctor who specialises in conditions that affect the brain, spinal cord and nerves. Having a neurologist, paediatric neurologist, or another doctor who specialises in Huntington's disease can be a huge help. Depending on their age, children and young people with Juvenile Huntington's are usually referred by their GP to a paediatrician, and then to a paediatric neurologist, or a neurologist.

- **Occupational therapist (OT)** - An OT will help the young person to move around, carry out normal everyday activities and do the things that matter to them. They will identify any difficulties that the child is experiencing and will provide practical solutions. As the disease progresses, equipment and mobility support will become necessary.

- **Physiotherapist (or physio)** - Physios help people to maintain and restore movement and function in their muscles through techniques like massage, exercises, and advice. Most physios won't have met someone with Juvenile Huntington's before, but they will look at the child's symptoms and decide how they can be managed. A physiotherapy programme for children with Juvenile Huntington's should focus on keeping the range of motion in the joints and on supporting independent mobility. It will also help to prevent the muscle contractions that cause the stiffness many young people with Juvenile Huntington's experience.

- **Speech and language therapist (SALT)** - Speech and language therapists help people who have difficulties with communicating, eating, drinking or swallowing. A SALT can help to keep up speech and swallowing ability for as long as possible, and introduce tools to improve on-going communication abilities. Young people with Juvenile Huntington's tend to perform much better with speech when they have an early referral to a speech therapist.

- **Dietitian** - Dietitians provide advice about diet and nutrition. Many people with Juvenile Huntington's start to lose weight. If eating becomes difficult or they are losing weight, a dietician will help by setting up a high-calorie diet to keep weight up. They can also help with advice around foods that are easy to eat.

- **Palliative care team** - Palliative care teams provide holistic care and support for people who have conditions that can't be cured, as well as that person's family or carers. They will often be able to offer support at any point in the condition. Different healthcare professionals provide palliative care support, but there are also some healthcare professionals and teams who specialise in palliative care.

Education and Juvenile Huntington's

Given their age, many young people with Juvenile Huntington's are likely to have regular contact with teachers at school.

Difficulties with movement may put the child at a higher risk from falls, and they will probably find it difficult to get to parts of the school without easy access. They may also find it difficult to carry their own books and equipment, and may require assistance. As the condition progresses, it may be necessary for them to be accompanied between classes, especially at times when the corridors are very busy and crowded. It may be useful to think ahead about what specific parts of the school may cause problems, so that there is time to find funding and establish any changes that need to be made. Often, however, all that is needed to overcome these difficulties is to make a few simple changes (e.g., switching the child's classes to ones in classrooms with easy access).

Young people with Juvenile Huntington's may also have cognitive (or 'thinking') changes that cause them difficulties within the school environment, and sometimes these changes have been happening for some time before they are diagnosed. Young people with Juvenile Huntington's may have a slowed response time. They may also find it harder to concentrate and they may have memory problems. It can be harder for them to initiate actions that they are perfectly capable of completing, which may make them appear to be lethargic and lazy. However, all of this is part of the condition, and is easily helped if they are given assistance when starting these activities. They also may be more impulsive and may have difficulty waiting for things, which is again a part of the condition. Finally, these difficulties with thinking tend to cause them to have problems with organisational skills (e.g., having equipment and books for

homework). This may also mean that they need extra help in classes. Sometimes, this can be given by a friend or the class teacher, although at other times they may require a learning support assistant.

Challenging behaviours are fairly common, in particular where symptoms begin in the teenage years. In some cases, this can be the most obvious symptom in the early stages of the disease, before changes to movement have begun. For some years, it may be uncertain whether their behaviour is due to Juvenile Huntington's. Some young people with Juvenile Huntington's may become very reluctant to go to school, particularly if they are beginning to struggle.

Epilepsy can become present in young people with Juvenile Huntington's, although less so when symptoms begin in the teens. Many teachers may be worried about how to deal with somebody who is having a seizure. However, there are many sources of support that can be used to get information and advice on epilepsy.

Young people with Juvenile Huntington's may also have problems with speech and with swallowing. They also often need a high calorie intake. To be able to get enough calories, they may need to eat small, frequent snacks in-between meals. They may also need more time to eat, because they will have to eat slowly, and will need a calm, quiet environment as they need to concentrate on eating. If drinking is difficult because of swallowing problems, care must be taken to ensure that they drink enough to avoid getting dehydrated, especially in the summer months.

Speech problems usually start off with mild slurring, but this will continue to become increasingly more affected as the disease progresses. Speech and communication problems are one of the aspects of the condition that can make it most difficult for the child with Juvenile Huntington's to maintain good relationships with peers. In class, the young person may need more time to speak,

understand a response and form their own response. Speech aids can be of help, but it is important that an assessment is made early on by a speech and language therapist and that any suggested solutions (e.g., flash cards, computer aids) are put in place as early as possible to allow the young person to get used to using them. It is also important that the school is aware of what aids are being used at home, so that a consistent method is being used.

It is also important for a school to consider the wider family. Any siblings of the person with Juvenile Huntington's may also be at risk. The school should be aware of how the situation may be impacting the siblings, especially their education, and their development. It can be very useful for staff and fellow students to gain a basic knowledge of Huntington's, as misunderstandings and misinformation can lead to problems.

Transition

When a person moves from child to adult services this is known as ‘transition’. This term covers many different areas of a young person’s life, including healthcare, education, employment, relationships, finances, and housing.

Planning for these transitions is an important role for those involved in the care of someone with Juvenile Huntington’s. It is important to consider that during this time someone’s condition or circumstances may also be changing, and they may require a lot of support. A key time for many young people will be when they finish school/college, and this should be carefully planned.

Palliative Care

Palliative care services can play an important role in the care of someone with Juvenile Huntington's.

Considering the wellbeing of family members is particularly important because of the inherited nature of the condition. This means that there are often other family members who have the condition. A child with Juvenile Huntington's may have a parent with Huntington's who is still alive, and they may be being cared for by the same carer. It also means that other members of the family may be at risk of developing Huntington's, including siblings of the child with Juvenile Huntington's. In some cases, there may be more than one sibling with Juvenile Huntington's at the same time, so there is also a possibility that a carer could be looking after multiple children in one home.

Parents may ask for advice or support in talking to their children, either about Juvenile Huntington's or about being at risk of developing Huntington's. Palliative care services can have an important role in supporting families who need help with this.

There is currently little guidance about symptom control in Juvenile Huntington's, with some clinicians using their own knowledge of treatments for similar conditions to treat the Huntington's symptoms. This is one of the reasons why palliative care services can have such an important role in the care of a child with Juvenile Huntington's. Given that children with Juvenile Huntington's can present very differently to adults with Huntington's, different treatments are often used.

It is important to use common-sense when prescribing medications. Anything prescribed should be directed at symptoms, but should consider potential sensitivities to certain medications and drug interactions. Medication should ideally be kept to a minimum and polypharmacy (the constant use of multiple medications) should be avoided where possible. Working closely with Huntington's specialists and palliative care teams can enhance symptom control.

Pain is a common symptom which is often under-treated due to fear of strong medication, as well as difficulty in assessing pain due to communication difficulties. Morphine appears to be well-tolerated and effective for the pain experienced by people with Juvenile Huntington's. Alternative pain relief may be required for muscle spasm related pain.

A high calorie intake is important for people with Juvenile Huntington's and this can help reduce symptoms. However, swallowing often becomes a problem and tube feeding may be offered as an option. This can help to ensure a good calorie intake. Decisions should be taken early as inserting a feeding tube later in the disease can be risky. It is important that young people with Juvenile Huntington's and their families are fully informed of the risks and benefits and have the opportunity to consider end of life decisions, such as withdrawing food.

Adult Huntington's disease usually progresses over 15-20 years, with death often being caused by Huntington's-related complications (e.g. pneumonia). There is some suggestion that the progression of Juvenile Huntington's might be quicker, but there is little evidence for this, and it is likely limited to a smaller group of those with Juvenile Huntington's. The reality is that, with both adult Huntington's and Juvenile Huntington's, it is very difficult to predict exactly how each individual person's Huntington's will progress.

In view of the natural progression of Huntington's disease, it is sensible to make advance care plans which outline preferences about end of life issues, such as place of care, resuscitation and feeding.

Resource library

The following charities, organisations, publications and websites may be able to provide further information and guidance on the topics covered in this guide:

Organisation	Contact details	Support
Huntington's Disease Association	0151 331 5444 info@hda.org.uk www.hda.org.uk	Advice Support resources Online forum
HD Buzz	hdbuzz.net	Research news
Huntington's Disease Youth Organization (HDYO)	info@hdyo.org en.hdyo.org	Advice Support resources Online forum

Notes

Get in touch

For advice and support
or to speak to a
Specialist Huntington's
Disease Adviser

email **info@hda.org.uk**

phone **0151 331 5444**

www.hda.org.uk

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