Huntington's Disease Association

Huntington's disease: A guide for GPs and primary care teams

We'll be there

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Presentation

What is Huntington's?

Huntington's disease is an illness caused by a faulty gene on chromosome 4. It affects the body's nervous system. Huntington's causes changes with movement, cognition, mental health and emotions. Once symptoms begin, the disease gradually progresses, with prognosis usually around 15 – 20 years. Living with Huntington's means having to adapt to change, constantly, for both the person affected and their family.

Most people develop Huntington's between the ages of 30 to 50 but there is a juvenile form of the illness which affects children and young adults under 21 at the point of diagnosis. As people live longer we are seeing onset in older people.

Living with Huntington's disease can be very challenging. Getting the right information and support is vital. The GP and primary care roles are essential in supporting families through their journey.

The triad of Huntington's symptoms

The symptoms of Huntington's vary widely between people. Even people in the same family may be affected differently. However, changes usually affect three main areas:

Movement

This is usually characterised by the presentation of involuntary movements (chorea) and impairment of voluntary movements. Although chorea is the most obvious symptom, it is usually the disturbance of voluntary movements that is more problematic to the individual as it results in reduced manual dexterity, slurred speech, swallowing difficulties, problems with balance, and falls. Both chorea and impairment of voluntary movements progress in the middle stages of Huntington's disease, but later, chorea often declines as the person becomes rigid and unable to initiate voluntary movements.

Rigidity, spasticity and dystonia tend to emerge later in the course of Huntington's, except in cases of childhood onset, in which they are often present from the beginning. Myoclonus, sudden brief jerks involving groups of muscles, is more common in juvenile-onset Huntington's disease, where it may be mistaken for a seizure. Epilepsy is uncommon, though not unheard of, in adults with Huntington's, but is said to be present in 30% of individuals with juvenile-onset Huntington's.

Cognitive

This symptom may not be initially obvious but causes the person with Huntington's a great deal of frustration. The cognitive disorder is characterised initially by a loss of speed and flexibility in thinking and processing information. This may be seen first in complex tasks, when the person is unable to keep up with the pace and lacks the flexibility required to alternate between tasks. The most prominent cognitive difficulties in Huntington's involve the so-called 'executive functions', abilities such as organisation, regulation and perception.

These fundamental abilities can affect performance in many cognitive areas, including speed, reasoning, planning, judgment, decision making, emotional engagement, perseveration, impulse control, temper control, perception, awareness, attention, language, learning, memory and timing. Cognitive losses accumulate and people develop more global impairments in the later stages of the disease.

Several studies have suggested that cognitive and behavioural impairments have a greater impact upon quality of life and a person's ability to function than the movement disorder in persons both in the work place and at home.

Mental health and changes in behaviour and personality

The most common specific psychiatric disorder in Huntington's is depression. Some people suffer from conditions such as major depression, bipolar disorder, or obsessive-compulsive disorder. Many, if not most, people with Huntington's also experience less well defined, non-specific changes in personality and mood, such as irritability, apathy, or disinhibition. Most of these psychiatric problems are believed to be related directly to the central nervous system injury caused by Huntington's.

People with Huntington's who have psychiatric disorders are often under diagnosed and are not treated appropriately. It is important to remember that psychiatric problems, particularly depression, are very common and very devastating in Huntington's, but they are also very treatable. Relieving depression and irritability in someone with Huntington's may be the single most effective help a doctor can offer.

Sometimes, symptoms are present for a long time before a diagnosis of Huntington's disease is made. Professionals and families can mistake Huntington's for a different illness such as Parkinson's disease or Alzheimer's disease. This is especially true when people are not aware that the faulty gene is in their family and that they are at risk.

If a person develops symptoms before the age of 21, this is known as Juvenile Huntington's disease. The symptoms may at first appear as stiffness and clumsiness in the arms and legs. Parents may notice a change of performance at school, behavioural changes and disturbances in speech.

The disease is progressive, meaning that symptoms increase over time and greater care and support is needed the longer someone has it.

- **The movement disorder is usually the most obvious first symptom.**
- **The cognitive disorder is usually the symptom that affects people most in daily life.**
- **The behavioural disorder usually gives the person with Huntington's and their carers the most concern.**

There are many ways that people living with Huntington's can get help and support with their symptoms to greatly improve their quality of life. The GP and primary care team can play a vital role in this support.

Aetiology and physiology

Huntington's is caused by a faulty version of the gene responsible for producing a protein called Huntingtin, which helps neurons in certain parts of the brain to develop before birth. If you have a faulty version of the gene, the protein it produces damages these neurons instead of developing them. This causes them to function poorly and reduce in number over time. It is autosomal dominant so each child of a parent carrying the Huntington's gene has a 50% chance of inheriting it, and therefore developing Huntington's.

The parts of the brain affected include the basal ganglia and cerebral cortex. These interconnected areas are associated with different types of activity including movement, learning, thinking, planning, motivation and emotion. As the cells in these parts of the brain reduce in number, changes occur in how they function, resulting in the various symptoms of Huntington's. Some of the effects of this may be:

- **loss of weight, despite a good appetite. This may be due to a combination of factors, including difficulties with chewing and swallowing, and the increased energy used by the body due to involuntary movements as well as metabolic changes. A referral to a dietician or a speech therapist can help with issues related to eating and swallowing.**
- **a person may eventually experience changes in their muscles. They may become more rigid over time, resulting in a slowing down of movements. Physiotherapy can be very helpful to ease stiffness and maintain movement for as long as possible.**

The ways Huntington's affects the person is unique which means the symptoms experienced will differ and will change over time. Having care that fits the person's needs and adapts to changes will make a huge difference. The GP and primary care can play a pivotal part in this.

DID YOU KNOW?

Each person with Huntington's is unique and will display different symptoms.

Early referral to a dietitian and speech and language therapist can help maintain weight for longer.

Physiotherapy can help reduce pain from stiffness.

Genetics and genetic testing

Some facts about genetics and Huntington's

- If a person has Huntington's it means they have inherited a faulty version of the Huntington's gene and the recipe for the protein it produces is incorrect.
- The faulty gene that causes Huntington's repeats a particular coding sequence known as CAG (cytosine-adenine-guanine) too many times. This means the protein it makes damages nerve cells in the brain.
- If a person has 40 or more CAG repeats, it is certain that they will develop Huntington's at some point.
- Every child conceived naturally to a parent who has the faulty gene has a 50% chance of inheriting it and the disease. If both parents have one faulty gene the child has a 75% chance of inheriting it.

There is a genetic test that can show whether the person is carrying the faulty gene by taking a blood test known as a predictive test. You normally need to be 18 years old to take the test.

Although the test is available many people choose not to take it. There are many reasons for this. Some of the things the person needs to consider are:

- The person at risk of Huntington's should be the person who makes the decision about testing and they usually have to be over 18. Parents, partners and other family members may try to coerce or dissuade one way or the other, but it remains their decision.
- If the person at risk decides to get tested, in addition to any emotional impact on them and their family, they also need to think about things like the effect of a positive result on other aspects of life such as life insurance and job opportunities. In certain occupations, such as the armed forces, it may be an obstacle to recruitment.

- If they have only recently discovered that they are at risk, it is important not to rush into making a decision about testing. Once a person has their test results, they can't change your mind about whether or not they wanted to know.

About the test

- Testing is only available at regional genetics clinics, which are located throughout England and Wales. GPs can arrange a referral. For more information go to www.hda.org.uk
- Going to a genetics clinic does not mean that a person is obliged to take the test but it does give them the opportunity to talk over all the implications and any other concerns they may have. Each clinic follows an agreed counselling 'protocol'; usually at least three sessions where you can talk over the implications of testing and any questions you may have, before you decide.
- A person can withdraw from the testing process at any time.
- If they decide to have the test carried out they may have two separate blood samples taken (to double check the results). The DNA which is extracted from the blood is then analysed in a specialised laboratory.
- According to national and international guidelines, follow-up counselling – after a test result – should be available.

Reproductive options

There are a number of different options that a person at risk of Huntington's can consider, such as:

- Conceive naturally, and accept the risk of a child inheriting the genetic condition.
- Try to conceive naturally and, if successful, undergo a test called prenatal diagnosis (PND), to find out if the developing embryo is affected. In most cases, this test is only carried out if you intend to terminate the pregnancy if the embryo is found to have the faulty Huntington's gene. (Although if you change your mind, this cannot be enforced.)
- Seek pre-implantation genetic diagnosis (PGD).

Pre-implantation genetic diagnosis (PGD)

In the case of PGD, after the fertilised embryos have developed for a few days, one or two cells are removed from each one. The genetic material (DNA and chromosomes) from the cells are then tested for Huntington's. Up to two unaffected embryos are then transferred into the woman's uterus. If the pregnancy is successful, the baby should not be affected by the disorder it was tested for.

- PGD is the only way for parents to have an unaffected child to whom they are both biological parents - but it is not an easy option.
- Like all IVF, it is a physically and emotionally demanding process, especially for the woman, and it may not result in a pregnancy. It requires a highly skilled technical team and laboratory set up which means only a few places deliver this service.
- The NHS Commissioning Board will pay for three cycles of preimplantation genetic diagnosis (PGD) for couples who have a proven genetic disorder and who wish to avoid the birth of an affected child, in accordance with the criteria outlined in Clinical Commissioning Policy: Pre-implantation Genetic Diagnosis, April 2013.

Use of donated eggs, sperm or embryos

Another way a person could conceive a child is through donated eggs, sperm or embryo. Depending on the treatment they need, they may well be looking at assisted conception via IVF, so again, this is not an easy option. But it is a way of avoiding the risk of Huntington's disease.

As with other kinds of assisted conception, it can be demanding, and funding for cycles to attempt it might be an issue.

For more information about clinics, PGD and other assisted conception techniques, visit the Human Fertilisation and Embryology Authority website **www.hfea.gov.uk**
You can also find information from the Genetic Alliance UK **www.geneticalliance.org.uk**

Adoption

This may be an option but the person's health and ability to look after a child will, however, be a factor under consideration. More information about adoption is available from organisations including Adoption UK and your local authority, among others.

DID YOU KNOW?

There is a test that shows whether someone carries the Huntington's gene, but many people at risk choose not to take it.

An embryo can be tested to see if it carries the gene.

Couples who are at risk can have PGD (pre-implantation genetic diagnosis).

Treatment

General principles of treatment

Due to the progressive nature of Huntington's, symptoms will change constantly over time. It is important to constantly re-evaluate treatments. Sometimes the most helpful thing a doctor can do is discontinue an unnecessary drug. People affected by Huntington's are highly vulnerable to side effects, therefore starting on low doses and increasing slowly is recommended. Polypharmacy should be avoided where possible and non pharmalogical interventions can also be helpful.

Treating the movement disorder

Discuss with the person with Huntington's if their movements are causing them problems or distress, as often they are not. Consider non drug intervention such as massage. Treating chorea can very often affect other aspects of the movement disorder, and will often have a detrimental effect on cognition and mood. Chorea generally lessens over time so reassessment of medication and reducing or withdrawing it may be necessary.

Medications used to suppress chorea

Class	Medication	Starting Dose	Max Dose	Adverse Effects
Neuroleptics	Olanzapine	2.5mgs at night	15-20mgs per day	Sedation, parkinsonism, akathisia, raised triglycerides, weight gain from increased appetite. Caution should be exercised in patients with diabetes and blood glucose.
	Risperidone	0.5-1mg/day	6mg/day	As above but less effect on increasing appetite.
	Quetiapine	25mg od	750mg/day	As above, less effects on lipids and glucose.
	Sulpiride	50-100 mgs/day	2400mg/day	Agitation, dystonia, akathisia, sedation, hypotension, dry mouth, constipation.
	Haloperidol	0.5-1mg/day	6-8mgs/day	Sedation, more parkinsonism, dystonia, akathisia, hypotension, constipation, dry mouth, weight gain.
Benzodiazepines	Clonazepam	0.5mgs/day	4mgs/day	Sedation, ataxia, apathy, cognitive impairment may be exacerbated, withdrawal seizures.
	Diazepam	1.25mgs/day	20mgs/day	Same as above.
Dopamine Depleting Agents	Tetrabenazine	12.5mgs once a day and titrating slowly up.	200mgs/day	Depression and sedation.

Specialist Huntington's services

Families often report that they have a more holistic service from a specialist Huntington's disease service. There are a number of these services around the country and will offer access to a team experienced in treating Huntington's.

Further information about services in the local area can be found at the local information section of this guide or by speaking to your local HDA specialist adviser. Contact details can be found at **www.hda.org.uk/supportnearyou**

Depression

It is easy to assume that developing depression is a natural reaction to a diagnosis of Huntington's. Research and clinical experience shows that many people with Huntington's are not depressed, and are able to adapt gradually to having the disease. Nonetheless, even severe depression in someone with Huntington's is often explained away as an 'understandable' reaction, therefore not requiring additional treatment. In fact, people who have a depressive syndrome, even when the depression is 'understandable', and even when there are clear triggers, usually respond to standard treatments, including medications and psychotherapy.

Because depression in Huntington's appears to be directly related to the brain disease, pharmacotherapy is usually indicated. Depressed patients should always be asked about suicide, and this should be regularly reassessed. The management and treatment of depression in people with Huntington's will improve their quality of life significantly.

Key points in the treatment of depression

- Avoid over interpretation of symptoms.
- Depression is very common in Huntington's. If in doubt start treatment and review.
- People with Huntington's are sensitive to side effects. Start medications at a low dose and increase gradually.
- Ask about substance abuse.
- Ask about suicide.

Signs and symptoms of depression

The signs and symptoms of depression may include:

- low or irritable mood
- loss of interest or pleasure in activities
- change in appetite, or weight loss
- insomnia or hypersomnia
- loss of energy
- feelings of worthlessness or guilt
- impaired concentration
- thoughts of death or suicide
- loss of libido
- feelings of hopelessness
- social withdrawal

Medications used to treat depression

Class	Medication	Starting Dose	Max Dose	Adverse Effects
SSRIs	Fluoxetine	10-20mg	60-80mg	Insomnia, diarrhoea, GI, initial agitation, upset, restlessness, weight loss, impaired erection or orgasm. Withdrawal syndrome if stopped suddenly.
	Sertraline	25-50mg	200mg	Similar.
	Citalopram	10-20mg	40mg	Similar. Doses above 40mg should be used with caution and ECG monitoring.
	Paroxetine	10-20mg	40-60mg	Similar, more sedation.
Tricyclics	Nortriptyline	10-25mg	150-200mg	Dry mouth, blurry vision, constipation, hypotension, tachycardia, sedation. Cardiotoxic in overdose.
Other	Mirtazepine	15mg	45mg	Sedation.
	Venlafaxine	25-37.5mg	225mg	Hypertension, nausea, headache, constipation.

Communication

Dysarthria

Dysarthria, a difficulty with the physical production of speech, results largely from impairment of voluntary movement. Speech becomes slurred, dysrhythmic, variable in volume due to inconsistent breath support, and increasingly difficult to understand. The listener must do everything possible to promote successful communication, beginning with allowing enough time. Many people with Huntington's thought to be incapable of communication can be understood if the listener is patient enough. The person may need to be moved to a quieter, calmer environment, and urged to speak slowly.

Useful strategies for communication

- Allow the person enough time to answer questions.
- Offer cues and prompts to get the person started.
- Give choices. For example, rather than asking "what do you want for dinner?" Ask "do you want beef burgers or corned beef hash?"
- Break the task or instructions down into small steps.
- If the person is confused, speak more simply and use visual cues to demonstrate what you are saying.
- Ask the person to repeat phrases you did not understand, or spell the words.
- Yes-no cards, or other communication devices may be helpful.
- Use of technology such as mobile phone or tablet.

Language

Communication, or the transfer of information from one person to another, requires a complex integration of thought, muscle control and breathing. Huntington's disease can impair all three of these functions.

There are two main aspects to communication: getting the information IN (understanding) and getting the information OUT (talking). Both of these aspects can be impaired by Huntington's, making communication a difficult task. The most prominent language difficulties in people with Huntington's are speaking clearly (articulation), starting conversation (initiation), and processing what has been said and responding.

In contrast to the basic impairments in language output, the basic capacity to understand language remains relatively intact in Huntington's. Even in later stages of the disease, language comprehension may remain when the ability to speak is significantly diminished. This fact is important to communicate to family members, staff at care facilities and other professionals involved. Even if a person cannot express themself, it is likely that they can understand what is being said.

Early referral to a speech and language therapist is recommended who will:

- Advise on strategies for communication.
- Arrange for assessment and provision of communication aids.

Swallow and nutrition

Difficulty in swallowing can indirectly be the most common cause of death in people with late stage Huntington's disease, whether through choking, aspiration or malnutrition.

Dysphagia results from impaired voluntary control of the mouth and tongue, impaired respiratory control due to chorea, and impaired judgment, resulting in eating too rapidly, or taking overly large bites of food and gulps of liquid.

Swallowing tips

- Eat slowly and without distractions.
- Prepare foods with appropriate size and texture.
- Eating may need to be supervised.
- Caregivers should know the Heimlich manoeuvre.

In late Huntington's, when even liquids may be difficult to swallow, the texture of food should be soft and smooth, and liquids may be thickened with an additive. The issue of feeding tubes should be discussed with the person and family before dysphagia becomes severe, to ensure that appropriate nutrition can be maintained throughout the illness. A gastrostomy tube can clearly improve nutritional status of someone with swallowing difficulties, however the advantages and disadvantages should be discussed with the person and their family before any decisions are made. A person may also have completed an advance decision to refuse treatment which should be adhered to.

Early referral to a speech and language therapist and dietitian is recommended, who will help identify swallowing difficulties. Periodic reassessment can identify changes in swallowing ability and suggest appropriate non-pharmacological interventions such as a change in food consistency.

Nutrition

The main aims of dietary treatment in Huntington's are to encourage people to achieve a well-balanced diet, which includes all the essential nutrients, to prevent or minimise the weight loss frequently seen in the mid to later stages of the disease, and to help people regain lost weight where possible.

It has been well documented that many people with Huntington's find it difficult to maintain their body weight and therefore need a higher than normal calorie intake. While the exact reasons for this are unknown, several studies have shown some people have a higher metabolic rate. This is frequently, but not always, associated with increased chorea movements. In the later stages of the disease increasing swallowing problems contribute to weight loss as mealtimes take longer and it can be difficult to ensure a puréed diet provides adequate nutrition.

Maintaining a healthy body weight is essential because people who are underweight (i.e. BMI of less than 18.5) lose muscle mass and therefore feel weaker, become apathetic and depressed, are more prone to catch infections, develop pressure ulcers if their mobility is compromised and take longer to recover from illness, operations or wounds. There is some anecdotal evidence that shows that providing a high calorie intake can help to reduce chorea movements, improve cognition and improve speech and swallowing.

It is important to monitor weight on a regular basis.

Juvenile Huntington's disease

Juvenile Huntington's disease is when the person is diagnosed with the disease before the age of 21. Juvenile Huntington's is quite rare. Less than 10% of people with the disease will have Juvenile-onset. This can make dealing with it quite an isolating experience for families, and meeting others in the same situation can help.

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

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**

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 an illness at all.

Not all behavioural changes, however, may be directly caused by the disease. Children of families affected by Huntington's may also be experiencing disruption and difficulties in their home life, which might impact their behaviour, or they may be facing other challenges in their lives.

For these reasons, because children and young people usually need to be over 18 before they can have the genetic test for Huntington's, and because Juvenile Huntington's symptoms can resemble those of other diseases (such as Parkinson's disease, depression or attention deficit disorder), sometimes the illness can be misdiagnosed or remain undiagnosed for some time. This is particularly true where a family history of the disease is not known.

Key facts:

- **Changes in behaviour or a drop in school performance are often the first noticeable symptoms of Juvenile Huntington's disease.**
- **Children and young people with Huntington's can remain undiagnosed for some time, as doctors and specialists may be reluctant to give a diagnosis or refer them for genetic testing before physical symptoms appear.**
- **Challenging behaviour tends to be more common in people who develop the disease in their teens, but many young people and children are not affected in this way.**

There are many ways that children and young people living with Juvenile Huntington's can get help and support with their symptoms to help them cope in school and greatly improve their quality of life. The HDA has a specialist Juvenile Huntington's adviser. Details can be found at **www.hda.org.uk**

Genetics

It is the same gene that determines whether a person will develop Huntington's disease or Juvenile Huntington's disease.
If they have 40 or more CAG repeats, it is certain that they will develop Huntington's at some point. If they have more than 50, it is highly likely that they will get Juvenile Huntington's (in other words, you will start to experience symptoms before the age of 21).
Less than 10% of people with the faulty Huntington's gene have more than 50 CAG repeats, making Juvenile Huntington's a very rare disease.

If a child or young person has more than 50 repeats, there is a 90% chance they got the faulty gene from their father, as CAG repeats tend to be more unstable when passed on from the man. It is not completely understood why this is, but it is thought to be because the gene becomes more unstable in sperm.

Diagnosis

If a child who has a parent with the Huntington's gene starts to show symptoms of Juvenile Huntington's, and if there is a strong suspicion that they may have it, a diagnostic blood test may be done to see if they do have it.

However, because Juvenile Huntington's is so rare and because its symptoms can be hard to recognise, it can often take a long time before a definite diagnosis is established. Not knowing can be stressful, but support is available to help them through this situation. The Huntington's Disease Association has an experienced adviser who specialises in Juvenile Huntington's.

Please visit **www.hda.org.uk** for more information and contact details.

DID YOU KNOW?

Juvenile Huntington's disease is very rare.

Juvenile Huntington's disease can be complex to diagnose.

The Huntington's Disease Association has a specialist adviser for Juvenile Huntington's.

Social issues

Equipment and adaptations

There is a variety of equipment and adaptations to the home which can often be helpful for people with Huntington's. For example, specialist seating, specialist beds and adaptations to the bathroom. Referrals to an occupational therapist and social worker can be very useful in exploring these options. Regular re-assessment is also very important due to the progressive nature of the illness.

Falls are common in people with Huntington's disease, and can be a source of significant morbidity. They are usually seen more in the moderate to advanced stages, and often result from the combination of spasticity, rigidity, chorea, and loss of balance. Most efforts at prevention do not involve drugs, but modification of the environment and behaviour of the person. Occupational and physiotherapists can advise people on how to sit, stand, transfer, and walk more safely. Installing handrails in key locations, and minimising the use of stairs can help to reduce falls. Some families convert a ground floor room into a bedroom. Furniture such as tables and desks, particularly items with sharp corners, should be arranged along the periphery of the room, where they will present less of an obstacle. People who fall out of bed may have a mattress placed beside the bed at night. The person with Huntington's will eventually become unable to walk and will need to be transported in a wheelchair. A weighted and padded chair, perhaps with a wedge to keep the hips tilted, or a pommel between the legs, may minimise the chance of someone with dystonia or severe choreic movements falling or sliding out, or knocking over the chair.

Accepting using a wheelchair can be difficult, but the person does not need to use it all the time. Mobility may be extended by using the wheelchair for longer excursions and using other assistive devices such as a walker for shorter distances, or in the home. Walkers with wheels may be particularly useful when rigidity or loss of balance is a problem. People who are particularly prone to falls sometimes wear helmets, or elbow and knee pads to minimize injury. Physiotherapy may also help by teaching patients how to minimise injury in a fall and how to get up again after a fall, and also teaching exercises to improve core strength.

Disability and benefits

The progressive nature of Huntington's will eventually force people to retire from employment. Unfortunately, the performance of people with Huntington's at their job may already have begun to deteriorate before they have received a diagnosis, or before they have made the connection between Huntington's and the problems they are having at work. Therefore, early identification of problems relating to Huntington's at work is very important, to help the person to stay in employment as long as possible. The employer must make reasonable adjustments to allow them to continue working.

There may also be issues of work safety. A doctor or specialist Huntington's disease adviser may be able to help the individual inform those at work of the nature of the problem, decide when to take retirement, and navigate the application process for benefits relating to their disability. Huntington's is a complex condition and the person may be unable to work, but may not have a single sign or symptom which, by itself, would qualify them for benefits. Therefore, disability benefits letters must be comprehensive, must stress functionality, and should include specific examples of dysfunction at work. Because of the particular nature of the dementia found in Huntington's, routine IQ test scores may not be relevant to the level of impairment because they do not reflect the organisational and task-switching problems found in Huntington's. Tests specifically directed toward executive function will better identify Huntington's-related cognitive deficits.

Insurance

In 2001 a moratorium on the use of genetic information by British insurance companies was agreed. Huntington's disease is, however, the only condition to be exempt from this, and insurers may ask for the results of predictive testing for life insurance valued over £500,000. Insurance eligibility also impacts on the success of mortgage applications. These issues are included in pre-test genetic counselling.

Driving

All people with Huntington's eventually lose the ability to drive. This can be a severe blow for people, who see driving as a sign of competence and a way of maintaining independence. There is a legal duty imposed on the holders of a driving licence to inform the Driver and Vehicle Licensing Agency (DVLA) as soon as they are aware of any disability which could affect their ability to drive now, or in the future, and which is expected to last more than three months. The obligation is printed on every driving license and Huntington's comes into the relevant category of disability, but only applies when signs and symptoms become apparent and not to asymptomatic people who have had a positive test.

Guidelines concerning a doctor's duty to inform the licensing authority without the patient's consent have been issued by the GMC – "Professional Conduct and Discipline: Fitness to Practise"; it is permissible but only in exceptional circumstances. The medical branch of the DVLA is willing to give advice to doctors and other health professionals.

Respite and care

People with Huntington's and their carers will often need support at some point. Exactly what form this takes will depend on individual circumstances. Referral to a social worker can be helpful in exploring these options.

DID YOU KNOW?

Referral to an occupational therapist can help maintain independence for longer.

Once a person has symptoms they must inform the DVLA.

Insurers can ask about the outcome of a genetic test for an application for life insurance over £500,000.

Psychological support

People with Huntington's, their families and carers often face considerable psychological and emotional distress. Much can be done to alleviate this, and to help people adjust and make the most of their coping skills. Research shows that individuals with Huntington's manage better with their illness if they have adequate support.

Impact of diagnosis

The diagnosis will have direct and profound implications for that person's children, siblings, and perhaps their parents and other relatives. A diagnosis may raise difficult questions for people as to when and how to discuss this with other family members. Any doctor who diagnoses Huntington's in a patient must be prepared to answer questions from and about these additional family members. It is also particularly important to consider the needs of other family members when thinking about the care of someone with Huntington's.

As the disease progresses

The major challenges experienced as the disease progresses are coping with repetitive loss and living with change, and this is true for all those whose lives are affected by Huntington's. Adjustment or coming to terms with the disease is made more difficult by the progression of the condition and the accompanying changes and losses. Early referral to a palliative care team is recommended.

Person symptomatic with Huntington's

The person with Huntington's will have to face many changes and losses as their condition progresses. They may not have insight into their symptoms or be fully aware of how they affect them. It is important to remember that they are a person with previous life and experiences.

Person at risk

Being at risk of Huntington's can create a huge variety of emotions. In thinking about the genetic test, people at risk may have mixed feelings.

The advantages of testing are that the uncertainty about gene status is removed. If there is no mutation, there is no more concern for themselves or their children. If the mutation is present they can make plans for the future, inform children, or decide whether to have children.

The disadvantages of testing is that if the mutation is present it may reduce the feeling of hope. There is uncertainty of when and how symptoms will start. There may be children or potential children at risk. There may be an impact on the individual, their partner, family and friends. The person may experience difficulties with getting insurance or a mortgage. If the mutation not present the person may experience 'survivor' guilt.

Although those at risk may have many thoughts and feelings relating to being at risk, it is important to remember that they may be caring for a parent or other family member and all the emotions that this brings.

Carer

Here we are mainly thinking of spousal carers, but this may also include other carers of people with Huntington's who are not at risk themselves (e.g., a family member who has had a negative genetic test). Although the person may not have the gene for Huntington's, it will affect their lives in many ways. Caring for someone with Huntington's can be incredibly difficult, tiring and stressful. The carer may have also cared for other family members in the past, and may face caring for other family members in the future. They may also worry about other family members, such as their children if a spouse has Huntington's.

Children

It is important to think about any children in a family affected by Huntington's. They will have to face many changes in their parent and often in their home situation, not all of which they will understand.

They may also need to take on additional caring roles. It can be difficult for parents to know how and when to discuss Huntington's with their children. The Huntington's Disease Association employs specialist youth workers who can support children. Details can be found at **www.hda.org.uk**

Impact on professionals

Huntington's disease is a complex illness and can often create feelings of inadequacy and emotions for professionals involved. Good multidisciplinary teamwork is necessary to provide support and opportunities to discuss concerns and responses to difficult situations. It is important to include the palliative care team to offer support and advice to other health professionals.

DID YOU KNOW?

The impact of a diagnosis has a ripple effect on a family.

People from families affected by Huntington's suffer repetitive loss.

Someone living at risk of Huntington's maybe seeing a mirror image of what may happen to them.

Palliative care

Huntington's is a progressive illness and due to the cognitive symptoms it may mean that someone is unable to express their wishes later in the disease.

It's important that people are given the opportunity to think about and discuss their wishes and preferences for their treatment and care. It may also be helpful to help sort out various practical, financial and emotional matters. Although it may be difficult for a patient and their family to think and talk about the end of life, some of the things you may like to encourage families to think or talk about include:

- **An advance care plan for future treatment and care.**
- **Where they would like to be cared for in the final stages of their illness.**
- **Setting up a lasting power of attorney.**
- **Writing a will.**
- **Creating an advance decision to refuse treatment.**
- **Organ and brain donation - perhaps supporting research into Huntington's if they wish to.**

When looking at the palliative care needs of someone with Huntington's and their family, it is important to remember that the hereditary nature of the illness offers some unique challenges. The person with Huntington's may have seen several family members die from the illness, which may impact upon their perception as to what will happen to them. Equally they may have some very firm views about end of life choices and these should be established as soon as possible. Equally, it is important to remember that siblings and children of the person with Huntington's are themselves at risk of developing the illness and so sensitivity is essential.

Pain

People affected by Huntington's have a varied response to pain. Sometimes pain is masked, and so they display no symptoms and may go on to develop severe difficulties such as pressure sores, burns etc. They may experience altered sensation. At the opposite end of the scale the person with Huntington's may suffer from severe nerve pain, or muscle pain.

Often simple analgesia is effective. It should be noted that the person with Huntington's may not be able to articulate that they are in pain, so may demonstrate symptoms such as agitation or challenging behaviour. If a response to simple analgesia is not evident then progression through the analgesia ladder is needed. Referral to the specialist palliative care team can be helpful.

Research

It is an exciting time in the world of Huntington's research with different strands of research being undertaken including looking into stem cell research, gene silencing techniques, and biotechnology research all looking into ways to slow down or delay the onset of Huntington's.
You can read more about how people can get involved at

www.hda.org.uk/research

where you can also read information about Huntington's research.

Local information
Further information can be found at **www.hda.org.uk**

Get in touch
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