Huntington Disease

General Information

Neurogenetic Clinic,
Groote Schuur Hospital

Division of Human Genetics,
University of Cape Town

The disease that you or your relative has is called **Huntington disease.**

PURPOSE OF THIS HANDBOOK

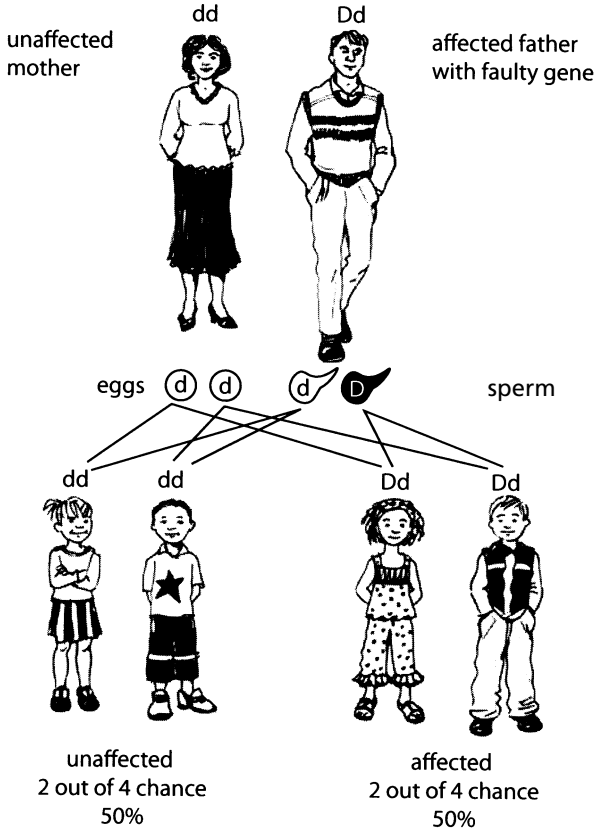
The aim of this handbook is to familiarise you with Huntington disease, so that you can recognise its symptoms and know what to expect as it progresses. By reading the book from time to time you will be able to see that many of the difficulties you are facing are part of the symptoms of the disease. Although you may find it depressing to read about the course of the disease when you only have mild symptoms, remember that by knowing all about the disease it will prepare you in recognising the symptoms that are part of the disease process. By having this information you will be able to anticipate and plan now for future changes that you will need to make in your life when the time comes.

WHAT IS HUNTINGTON DISEASE?

Huntington disease is an inherited disorder caused by a faulty gene which affects the brain. In other words the disease is passed down from generation to generation through family members who have the disease or who have the faulty gene. You need to understand the fact that it is a family disease and that nobody is to blame for the disease.

The disease starts gradually, usually between the ages of 30 and 50. The symptoms affect both the body and the mind and last for 15 to 20 years or longer. There is no cure and there are no easy answers to manage the symptoms, but knowing all about the disease will help you to face the challenges you meet from time to time.

HOW DO YOU GET HUNTINGTON DISEASE?



As you can see in the drawing above, people who have Huntington disease have one 'good' copy of the gene and one faulty copy of the gene causing Huntington disease. When they have a child they may pass on the 'good' copy or they may pass on the faulty copy. There is a 50:50 chance each time, so each daughter and son has a 50% risk or a '1 in 2 chance' of developing the disease. If you have inherited the faulty gene you will develop the disease some time during your life and your children and grandchildren will also be at-risk of developing the disease. If you do not inherit the faulty gene you will not develop the disease and will not pass it onto your children or grandchildren.

SYMPTOMS AND PROGRESS OF THE DISEASE

There are 3 major groups of symptoms and each person is affected differently. The symptoms start slowly and mildly and you may not notice them for a few years after they have developed. Not all difficulties mentioned below affect everybody with Huntington disease. Nor do they affect everybody in the same way or in the same order. One person may start with clumsiness or uncontrolled movements; another with depression or irritability; and a third with changes in thinking processes which make her or him forgetful or not able to do her or his job as well as in the past.

Movement difficulties

Individuals may find that they are clumsy; dropping objects, knocking things over or bumping into objects when walking. They may develop slight twitching of the face, particularly around the mouth, and fidgeting movements of their fingers and toes.

Gradually the arms and legs and body may develop jerky movements which are worse when the individual walks or becomes anxious or distressed. The movements are present all the time the person is awake, but generally stop or become much quieter at night during sleep. The movements cannot be controlled and are often not disturbing to the person, but are irritating and tiresome to those watching or hearing the movements.

As the disease progresses the person starts to stumble and lurch when walking. Speech becomes slow and slurred and difficult to understand which the individual finds frustrating and often leads to irritability and temper outbursts. However, even though the speech might be difficult to understand the person can still understand conversation. These speech and walking difficulties are often mistaken for drunkenness by members of the community.

Chewing food and swallowing liquids and food is difficult and often leads to bouts of choking.

Thinking difficulties

The person may start to be forgetful, have difficulty with concentrating or remembering recent information and may not be able to perform duties at work correctly or as quickly as before. There may be difficulties with routine activities such as washing, dressing, eating or setting the table, that previously were performed easily.

Even though memory is affected the person recognises people, knows where they are and the day and time. They have the insight to know that their thinking has been affected and that they are no longer able to think as fast as they used to and are unable to perform certain thinking tasks that they used to do.

Although they understand what is being discussed they may have difficulty in starting a conversation or staying on the topic of conversation and may switch from one topic to another. They may also repeat themselves over and over on one subject after the conversation has moved on and have difficulty getting off it and moving onto the next conversation.

Personality or emotional symptoms

Individuals may show irritability, agitation, moodiness or outbursts of anger which are out of character for the person or much worse than previously if that was already present in their personality. These aggressive outbursts may take the form of swearing or physical lashing out at others in the vicinity. This may be with their arms or legs or any objects that are close to them.

The individual often suffers from periods of depression.

He may also appear to lack motivation and appear lazy. He may show no interest in the people around or in the environment or in previous hobbies or leisure activities. He may be difficult to get along with and suspect family members or others around of planning to deceive them.

The family may find the individual's behaviour embarrassing in public. He may use bad language or become impatient with others very quickly if he doesn't get what he wants immediately. Because the individual has difficulty in controlling his behaviour he may make derogatory personal comments about people who are able to hear them, make sexual advances to his partner in front of the children, take other people's cigarettes right out of their mouths or scratch himself inappropriately in the company of others.

These emotional and personality symptoms are the most difficult for the family to manage although not everyone faces every difficulty and not every difficulty is present at once or in the same order as for somebody else. The person is often unable to see changes in himself and vigorously denies their existence.

Some individuals deny that they are depressed because they lack insight. However, the caregiver often can notice it and should report this to the doctor at the clinic visit as it is possible that medication can relieve it and make the person feel better which will help him to be less irritable and angry.

Other disturbances

Weight loss: occurs in spite of the person having a good appetite and eating large amounts of nutritious food.

Sleep: some individuals sleep a lot during the day and very little at night.

Functional independence: gradually the person loses the ability to dress, wash and care for himself becoming more and more dependent on the caregiver for assistance and finally for doing everything for him.

Incontinence: very late in the disease individuals may lose the ability to control their bladder or bowels and wet or soil their clothes or bed linen.

PREDICTIVE TESTING

Some individuals find it very stressful to live with the knowledge that they may have inherited the faulty gene and feel that they want to know for certain whether this is so or not. There is a blood test that can be done which will be able to give you this information. Only very occasionally is the test not able to give you a definite result.

Although this test is available, it does not mean that you have to have it. You need to think about it very carefully to see whether this is the right thing for you to do or whether you would prefer to live without knowing for certain whether you will develop the disease or not. If you do decide to have the test, you should first clarify any life insurance implications that this may have. You need to be over eighteen years of age to be able to have the test done.

If you have only recently found out that you are at risk of developing the disease, then be careful not to rush into making a decision about having the test. Once you have been given your test result this information will be with you for the rest of your life.

You are the only person who can decide about whether you want to have the test or not. Parents, partners, other family members, employers or doctors may try and persuade you, but it is entirely your decision.

If you decide that you would like to have the test done, it can be arranged through the clinic sisters whose contact details are provided at the end of the booklet. The predictive testing programme provides you with all the information about the way Huntington disease is passed down in families. It also gives you the opportunity of thinking about and discussing all the consequences that the test might have and for you to ask any questions you may want clarified. The testing programme is usually spread over six sessions, so that you have a good understanding of all the information to help you to make the decision. You may decide at any stage that you do not want to continue with the programme, even just before you are due to be given the result. It is entirely up to you and will not affect your or your family members' future treatment at the clinic.

After you have been given the results you will have follow-up visits which will give you the opportunity of discussing any concerns about your result or any aspects of your life which are worrying you.

DECIDING ON WHETHER TO HAVE CHILDREN

If you are at-risk for developing Huntington disease it may affect your decision about having your own children or not. Some people decide to never have children at all so that they cannot pass on the risk, or because they are concerned about being able to care for their children once they develop symptoms of the disease. Others decide to go ahead and to have children, based on the fact that they and their children (even if they do inherit the faulty gene) will most likely have many years of normal life before developing the disease.

Your decision to have children may depend on the result of your predictive test. If your test is negative, showing that you ***do not*** carry the faulty gene, then you will not pass it on to your children. If your test shows that you ***do*** have the faulty gene and you still want to have children you can discuss the different

options during your follow-up sessions with the genetic counselor before becoming pregnant if you are concerned about passing the faulty gene on to your children.

WHAT TREATMENT IS THERE FOR HUNTINGTON DISEASE?

At present there is no cure for Huntington disease, but there are medications that help to control symptoms such as the movements, sleep disturbances, mood swings or depression. It is important that you report any problems you are experiencing to the doctors at the clinic so that he or she can prescribe medication that might be able to help these difficult symptoms.

WHAT CAN BE DONE TO KEEP HEALTHY AND ACTIVE?

You should remain physically, mentally and socially active for as long as possible. This means that you should continue with your normal activities and hobbies such as walking, reading, listening to the news, visiting friends, going to church, attending a gym or continuing with anything that you have enjoyed in the past. Because people with Huntington disease often have difficulty in organising themselves and getting started with an activity your caregiver may need to help you to get them started and support you in engaging in any activity.

There are no specific exercises that will delay the progress of the disease, but you should do exercises to maintain your strength, balance and posture. It is also important to keep your lungs and heart healthy. The best form of exercise is to take a walk every day for at least 20 minutes.

One aspect that will help you to remain independent for a longer time is for you to develop a set routine with all activities such as dressing, washing, setting the table and washing the dishes. You

should follow these routines as soon as you have been diagnosed so that the routine of each activity becomes absolutely automatic and you do not have to think about the way in which you are doing it. For example, each time you get dressed, lay your clothes out on the bed in a specific way. Check that they are the right side out and that the back of the garment is facing upwards. Lay them down in the order in which you are going to put them on: first the shirt, then the trousers, then the socks and finally the shoes. Keep to this exact routine every time you dress yourself.

'SYMPTOM WATCHING'

Sometimes people who are at risk of developing Huntington disease become anxious and start noticing actions or behaviours in themselves such as dropping objects or moodiness or forgetfulness, similar to what their affected relatives had, and think that they have started with the symptoms. This increases their anxiety which in turn makes them more forgetful or irritable or clumsy. Many of the symptoms of Huntington disease are also experienced by the general population who do not have Huntington disease and so they are not necessarily an indication that the disease has started. Anyone who is anxious should attend the Neurogenetic Clinic for examination by a neurologist.

TALKING TO YOUR CHILDREN ABOUT HUNTINGTON DISEASE

It is important to talk to your children about your disease to explain the symptoms of the illness. This can be done when answering their questions about your movements or changes in behaviour. When they are told that it is a family disease they may want to know whether they will also develop it one day. This is a good time to discuss their risks and the possibilities there are for predictive testing once they are over eighteen years of age should they want to know whether they have the faulty gene or not.

DIFFICULTIES SOME PEOPLE WITH HUNTINGTON DISEASE MAY EXPERIENCE

At the time the doctor tells you that you have Huntington disease.

- Accepting that you have the disease. This may be difficult if, by nature, you have always been an independent person and now have to come to terms with losing your independence; or having to rely on others to prompt your memory for events that have occurred or activities that have taken place; or to remind you to do your daily routine tasks;
- Deciding who and when and what to tell other people about Huntington disease;
- Coping with the different ways in which your spouse, family, friends and working colleagues relate to you since you were told you had the disease;
- Telling your children about the seriousness of your illness;
- Telling your children about Huntington disease;
- Dealing with difficulties at work – deciding when and how you should stop working;
- Planning and managing your future financial and legal matters;
- Deciding when to stop driving and planning how to manage transport without being able to drive yourself;
- Feeling unmotivated and not being interested in anybody or anything;
- Coping with emotional changes like anger, sadness and anxiety;
- Coping with increased clumsiness – dropping and breaking things, bumping into objects;
- Unable to keep up at work and having difficulty with going from one task to the next; and
- Thinking about and planning for the future in general.

Later on you might have difficulty with:

- Having difficulty in doing two things at once. For example, you cannot continue with a conversation while you are drying the dishes or dressing yourself. You need to concentrate on one task at a time without any interruption or distraction;
- Getting organised and having a sense of time. You may have difficulty in planning and completing tasks within a certain time or even forgetting about appointments;
- Caring for your children;
- Coping with lots of noise and lots of things happening at once;
- Being embarrassed by clumsiness, stumbling when walking outside and being mistaken as being drunk;
- Communication. Although you understand everything, you may slur or trip over your words or have difficulty in saying some words. Sometimes you can start a sentence, but you cannot finish it even though you know what you want to say. In conversations with friends and family you may prefer just to sit and listen and only respond to their questions as you have difficulty initiating conversation;
- Your writing may be difficulty to read. People notice that you fidget and are restless;
- Confusion. You may have difficulty in making wise and rational decisions in situations that you previously used to do this comfortably on your own. Even making the choice between two options may take you a long time and then you still may not be sure about your final choice. You sometimes get confused about finding your way to places that are very familiar to you;
- Not being able to control your emotions and lashing out at people you love;
- Shopping and paying bills or managing money. You have difficulty in adding the costs of items at a shop and cannot work out the change. You sometimes forget to pay bills and find it difficult to work out your monthly budget;
- General routine chores around the home;

- On social occasions you might not react appropriately by offering help where it is needed or showing concern for others who have problems. You do not make contact with friends or family yourself.
- Safety around your home. You may forget to switch off electrical appliances such as the stove or kettle or to lock the doors when you leave the house or go to bed. If you smoke you forget to put the cigarette out before leaving the room or drop live ashes on your clothes or the furniture without noticing it.

Even later on you may have difficulty with:

- Monitoring yourself and the things you need help with;
- Showering, dressing and feeding yourself;
- Feeling lonely and isolated;
- Feeling tired all the time and having no energy to do things;
- Feeling hungry and losing weight;
- Eating and swallowing difficulties;
- Having extreme feelings that are hard to control or understand;
- Your partner or caregiver getting tired while caring for you and needing a respite break.

Remember that the above lists of difficulties do not mean that you will develop all of them or when you will start with the difficulties you do develop. It is just a guide to provide you with information on the disease so that you will be able to recognise that the difficulties you develop are part of the disease.

RECOMMENDATIONS

Driving

Once you have been diagnosed with Huntington disease you should consult the doctor at each clinic visit and ask her or him whether it is safe for you to continue driving, especially if family members or work colleagues have commented on your driving. Remember it is your responsibility to ensure that it is safe for you to drive as you might not be needing to see the doctor regularly and your driving skills might change between clinic visits.

Smoking

You should start developing a habit of smoking outside or in a room with cement or tiled floors.

Medication

If the doctor prescribes medication for you it is important that you take it regularly even if you experience side effects such as a dry mouth as some medicines take a few weeks before they start to benefit you.

Routine appointments should be kept so that the doctor can monitor the action of the medication and regulate it accordingly. This might mean prescribing a new medicine, increasing or decreasing the dose of the current medicine, or taking you off the medication.

INTERNET SITES FOR MORE INFORMATION ON HUNTINGTON DISEASE

International Research Consortium:
www.huntington-study-group.org

International Huntington's Disease Association:
www.huntington-assoc.com

Hereditary Disease Foundation: www.hdfoundation.org
Huntington's Disease Society of America: www.hdsa.org

Huntington's Society of Canada: www.hd-ca.org

The centre for genetics education:
www.genetics.com.au/factsheet/07.htm

CONTACT DETAILS

For information on disability grants, grants-in-aid, school fees rebates and Medic Alert bracelets contact the social worker, Belinda Bailey, at 021-404-5446.

For assistance for more information regarding medication, predictive testing or arranging clinic appointments contact Sr Sklar or Sr Legg at 021-404-6235 or 021-406-6304.

BIBLIOGRAPHY

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Smith Steve (2005) Huntington's disease: A nursing guide. Fern House: Cambridgeshire