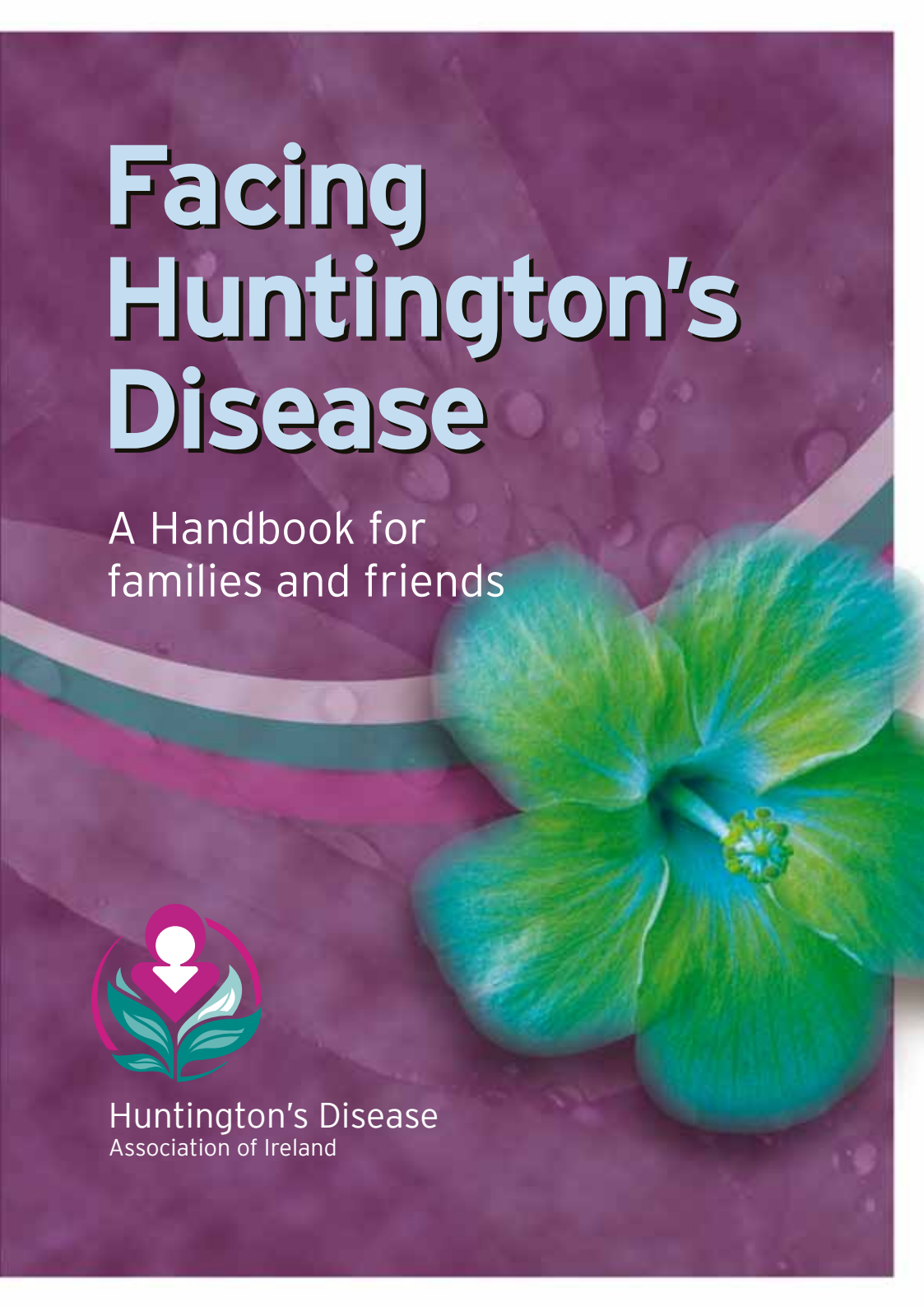
Facing Huntington's Disease

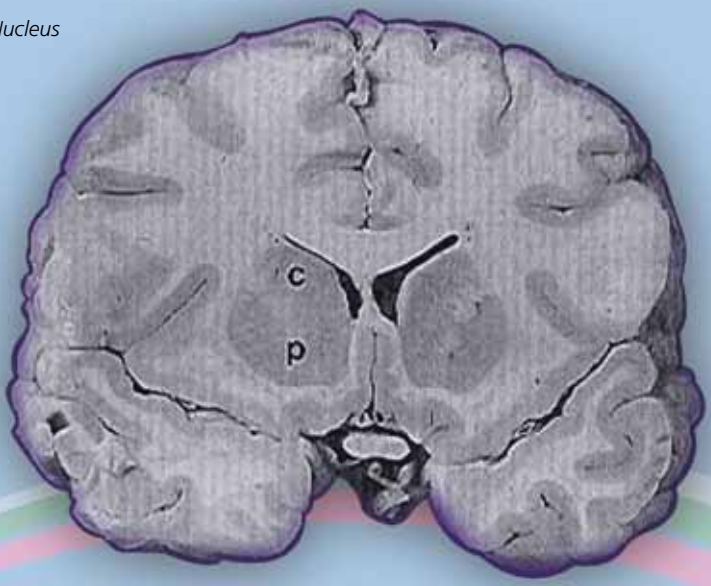
A Handbook for families and friends



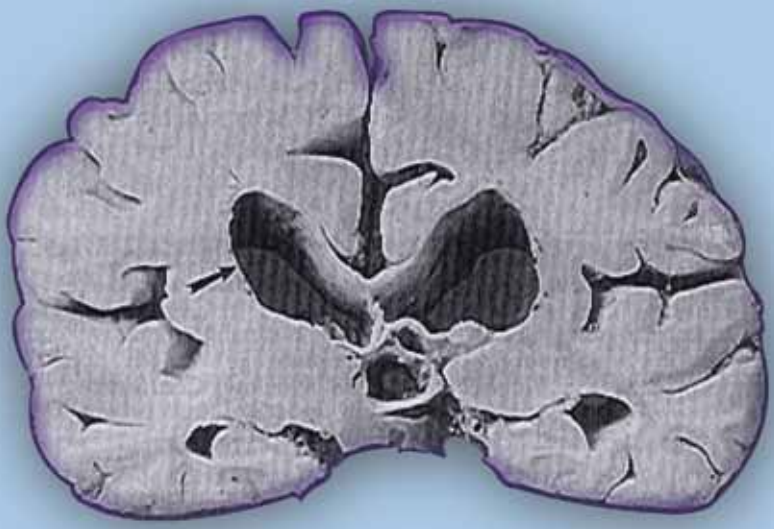
Huntington's Disease
Association of Ireland

Coronal section of a normal brain

c = Caudate Nucleus
p = Putamen



Coronal section of a HD brain



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1 INTRODUCTION

THE PURPOSE OF THIS BOOK

This booklet is intended to give an introduction to Huntington's Disease (HD) and the issues that this condition raises for the symptomatic individual, those at risk, family members, family carers and professionals working with people affected by HD.

It is not a replacement for more detailed texts or professional information but presents the facts in what is hoped is a straightforward and helpful way. Further information is available in the HD Handbooks:

Caregivers Handbook, Physicians Guide and Understanding Behaviour.

Various Thank You's

The contents of this book owe much to other publications, especially material kindly made available by International Huntington's Disease Organisations. It has been edited by the Huntington's Disease Association of Ireland with contributions from Professor Andrew Green, National Centre for Medical Genetics, Dr Niall Pender, Neuropsychologist, Beaumont Hospital and HD family members. The 2012 edition is printed with financial assistance from the Hospital Saturday Fund.

Feedback Invited

Huntington's Disease is a condition with many facets and in attempting to describe these facets, the authors do not underestimate the range of problems that the condition brings; it is a feature of HD that there are many examples of differences from the norm, and thus readers may have direct experience which differs from that detailed in this publication. Huntington's Disease Association of Ireland (HDAI) welcomes your feedback.

HD OVERVIEW

Huntington's Disease is a hereditary neurodegenerative disorder that causes progressive deterioration of the physical, cognitive and emotional self. Most people with HD develop the symptoms between the ages of 30 and 50, although there may be subtle changes much earlier. About 5 -10% of people affected have onset of symptoms before age 20 (Juvenile HD) and 10% have onset after age 60. In this booklet a person with HD symptoms is referred to as a pwHD.

HD is a genetic condition with each child of an affected parent having a one in two chance or 50% likelihood of inheriting the gene. Both men and women have equal chances of being affected. People with the abnormal gene will almost always develop the disease, unless they die of other causes prior to developing symptoms.

People who do not inherit the HD gene will not develop the disease, neither will their children, or their children's children. The disease does not skip a generation.

The average survival time after diagnosis is about 15-20 years, but some people have lived 30 or 40 years with HD.

Maintaining general health and well-being, will bring about improvements in the pwHD's quality of life.

HD is a complex disease and requires a multidisciplinary approach, involving a range of services that are required at each, differing stage of a person's life with the disease.

Clinical assessment of motor, cognitive, emotional or mental health symptoms should determine services required. Rehabilitative therapy including: physio, occupational, speech & language and diet & nutrition expertise can help maintain physical function. Support and information, psychological, neuro psychological or psychiatric services can help with cognitive and psychiatric changes.

Maintaining a healthy body weight is essential because people who are underweight lose muscle mass and therefore feel weaker, become more apathetic and depressed are more prone to catch infections, develop pressure ulcers and take longer to recover from illness.

Anecdotal evidence suggests that providing a high calorie intake can help to reduce chorea movements, improve cognition and improve speech and swallowing.

Absent family history of HD

Some individuals develop HD without ever knowing they were at risk, because they have no known family members with the condition. This occurs in 2-5% of all cases. Sometimes this can be explained by early death of a parent who carried the gene, but did not live long enough to manifest the symptoms, by adoption, or by mistaken paternity. Others represent *"new mutations,"* caused by rare expansions of parental genes with a high-normal CAG repeat number (27-35 repeats) into the affected range in the child. A genetic counsellor can provide information on this.

Finding a Cure or Treatment

Huntington's Disease is a neurodegenerative disorder caused by an expansion in the IT-15, or huntingtin, gene on chromosome 4, which encodes the protein Huntington. Although there is currently no cure for HD, many of the symptoms can be treated. Research continues, bringing hope for the future in terms of finding better treatments or delaying/reversing the progression of symptoms.

Delaying onset

Recent research suggests that leading a more active physical and intellectual lifestyle can delay the onset of symptoms. Keeping physically and mentally active from an early age can have a positive impact for people at risk.

Prevalence of HD

Professor Patrick Morrison conducted a survey in Northern Ireland which estimates the prevalence for 2001 of 10·6 people per 100 000 (Lancet Neurology). Based on the Republic of Ireland population of 4,670,976 (July 2011) this would indicate approximately 500 with the condition with a further 2500 at risk. While HD is relatively rare, over 9000 family members in Ireland may require support and information.

ABOUT HDAI

HDAI is a national voluntary organisation established to provide consultation, information and individualised support to those diagnosed with Huntington's

Disease, their families and their health and social care teams. HDAI is a registered charity incorporated in 1998.

Information is regularly updated through links with International, European and other National Associations. Because HDAI has been dealing specifically with HD in Ireland for over 25 years, the Association has extensive expertise and knowledge in supporting individuals and families affected.

HDAI exists to provide a unique service offering understanding, information and support to all those affected by HD. Confidentiality is respected.

Services include:

- A national information, support and advocacy service based in Dublin which provides information and support to families; individuals at risk of HD; carers and health and social care professionals throughout the country
- A Family Support Officer available to meet family members in crisis
- Access to counselling for those in need
- Support group meetings /carers workshops regionally
- An annual information meeting and respite weekend available to people with HD and their families
- Publications including leaflets, booklets, and articles covering the many issues specific to HD available for families, social care and health professionals on request
- A quarterly newsletter and annual magazine
- Loan of specialised HD aids and equipment
- Talks and information seminars on request
- Therapeutic treatments for people in the mid stages of HD
- HD ID cards provided on request

Information is available by contacting the HDAI office or referring to the website **www.huntingtons.ie**

HISTORY

Huntington's Disease was first described in 1872 when an American doctor, George Huntington, had his paper published in the Medical and Surgical Reporter of Philadelphia and the hereditary disorder he described became known as Huntington's Chorea. The word *"chorea"* is derived from Latin and Greek words meaning chorus or group of dancers. Today the term Huntington's Disease rather than Huntington's Chorea is more common because we have learned that some patients display a more rigid form of the disease and some may show more intellectual and behavioural deterioration rather than physical symptoms especially in the earlier stages of the illness.

Today, earlier diagnosis and better management provides an improved quality of life for people with this condition. Ongoing research provides greater hope for the future.

2 THE FACTS

WHAT HAPPENS IN THE BRAIN

Huntington's Disease is caused by the destruction of brain cells, particularly in those parts of the brain known as the basal ganglia and the cerebral cortex.
(See diagram of HD brain on inside cover).

The disorder manifests as a triad of motor, cognitive, and psychiatric symptoms which begin insidiously and progress over many years. If you fear that HD may be present the best way to resolve worrying questions is to see a specialist who is experienced in the diagnosis and treatment of neurological disorders. Ask your GP to refer you to a Neurologist.

SYMPTOMS

The movement disorder is characterised by the emergence of involuntary movements, or chorea and by impairment of voluntary movements. Symptoms include 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 patients become rigid and unable to initiate voluntary movements.

A minority of people develop a form of muscular rigidity rather than chorea.

The cognitive disorder is characterised initially by a loss of speed and flexibility. 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.

Cognitive losses accumulate and individuals develop more global impairments in the later stages of the disease. The most common specific psychiatric disorder in Huntington's disease is depression. Individuals may also suffer from mania or obsessive preoccupations.

Other symptoms (which may not fit a specific psychiatric category) include irritability, anxiety, agitation, impulsivity, apathy, social withdrawal and obsessiveness. Symptoms vary over the course of the disorder. Symptoms also vary from individual to individual, even within a family. Some symptoms may not be HD related. Individuals with HD can develop other unrelated illnesses therefore if in doubt it is best to discuss with the person's GP.

JUVENILE HUNTINGTON'S DISEASE

In Juvenile Huntington's Disease (JHD) symptoms manifest in people before the age of twenty years. JHD occurs in about 5-10% of cases of HD. JHD can present with somewhat different features to adult onset. Chorea is a much less prominent feature.

DISEASE PROGRESSION

The disease can be roughly divided into three stages. Early in the disorder, individuals may continue to work, drive, and live independently.

Symptoms may include minor involuntary movements, subtle loss of coordination, difficulties in problem solving, and perhaps a depressed or irritable mood.

In the middle stage, individuals will probably not be able to work or drive, manage finances or perform household chores, but will be able to eat, dress, and attend to personal hygiene with assistance.

There may be problems with swallowing, balance, falls, and weight loss. Problem solving becomes more difficult because individuals cannot sequence, organise, or prioritise information as they did before. In the advanced stage of HD, patients will require assistance in all activities of daily living.

Regular respite breaks and community support can help keep the pwHD at home for longer. It is important to discuss issues relating to end of life care before the person loses their ability to communicate.

PSYCHOLOGICAL ASPECTS

By Dr Niall Pender, *Principal Clinical Neuropsychologist, Beaumont Hospital, Dublin*

A diagnosis of HD in a family is a devastating and confusing time for everyone, whether you are a parent, spouse, sibling or child, the diagnosis brings a great deal of emotions. These range from fear, anxiety, anger, sadness and guilt whether you are unaffected, positive with the gene or have not had the test. Getting help and information is the first stage in trying to manage the disease.

For the person with the gene it can be difficult to live a normal life as one waits for the initial signs of the disease. This is very normal but very often people misinterpret normal everyday lapses in thinking and behaviour or simple clumsiness as signs of the onset of the condition. Family members watch the person and it can be difficult to forget about the disease.

Many people feel that they become defined by the disease. Unfortunately, HD can slowly affect one's ability to manage relationships and control emotions. People can become more irritable and at times irrational. This is because HD affects pathways from the frontal lobes of the brain (which control and manage behaviour and emotion) to other parts of the brain.

The person themselves has no control over these changes and they are as confusing to them as everyone else. The brain changes can affect thinking skills such as memory recall, learning, understanding emotion, planning and mental flexibility. These are often very early changes for people. As HD progresses it can also be associated with high rates of depression and mood changes and these are terribly disabling for many people.

Sometimes these changes in emotion and thinking can result in people stopping work or education and falling out with their family. Due to the manner in which the brain changes occur, many people with the disease have little awareness of the severity of their symptoms.

This can, of course, be protective for the person themselves but can cause great heartache for family and friends.

MANAGEMENT OPTIONS

I have worked with people with HD for many years at various stages of the disease. One of the many questions asked is *"What can be done about these changes"?* Well the initial point is to begin to get help. The HDAI is a first port for many people and knowledge is vital to a clear understanding of the condition. For many people it is hard to talk to other's about the disease. I would encourage you to attend your GP and seek help and advice.

Often many difficulties can be managed with a clear multi-disciplinary treatment plan. While specialist services for people with HD and their families are sorely lacking in Ireland there are attempts to improve these. There is a need for symptomatic management of thinking changes, mood changes and behavioural treatment. These go together with the ongoing medical treatment. It is vital that the care of people with HD and their families is seen as a multi-disciplinary experience. Through this planned input, a good quality of life can be maintained for many years.

SELF MANAGEMENT

Self-management is the idea that each person plays a role in managing their own condition in so far as that is possible for them. There are many factors that can help a person mange their condition better such as getting better informed (from reliable sources) about the illness, getting assistance and support from family members, and regular support from specialists about medication, diet and fitness.

By putting plans into place to manage difficulties one can offset the development of the symptoms for a long-time. I would also urge carers and family members to seek help to address their concerns and worries. The extent of distress experienced by carers is significant and can also result in psychological difficulties. Many gene negative family members suffer from terrible emotional difficulties and can benefit from supportive psychological treatment.

COGNITIVE CHANGES

Cognitive changes are those that I see most often as a neuropsychologist and these include changes in memory, understanding emotions, poor concentration and poor planning, mental flexibility and multi-tasking. Here is some advice for managing these on a day to day basis.

1. Any changes to the brain can slow down brain function. Therefore, do not overload the person with lots of demands
2. Remember the *"little and often"* rule for memory. Small amounts of information delivered frequently are more likely to be remembered

3. Do not rush the person to provide answers or make decisions
4. Fatigue is very common in neurological conditions. Take regular rests
5. Try to reduce distraction when learning information or preforming tasks as this will help concentration
6. Remember to use diaries, memory aids, calendars, phones or computers to remember. Prompting helps. Recognition is easier than recall
7. Take time to plan your day and activities. Review the day at the end and make lists of activies and tasks to complete
8. If you have worries and concerns please ask your doctor or the HDAI for advice and referral to specialists. Sometimes the worry about symptoms can be eased with help and advice

MENTAL CAPACITY AND LEGAL ISSUES

Following from the fact that HD affects thinking and intellectual ability over time it is important to consider the legal issues involved in decision making. Importantly, as HD is associated with a known pattern of thinking difficulties which can affect insight, awareness, planning and memory it is always wise to consider talking to a solicitor about issues around decision making. This could be about financial planning, treatment decisions (which should also be discussed with your doctor) and issues about living arrangements and palliative care. It is best to begin to think of these (even though it can be upsetting) early in the disease course so that your own wishes and feelings can be considered. Your solicitor can help with these issues.

IN SUMMARY

HD is associated with a range of behavioural, thinking and emotional changes but due to the very nature of the disease, there is no uniform pattern of cognitive impairment. Variation within and between families is common and it is very hard to predict the exact changes that will happen to each person. I would urge careful planning, seeking treatment for both affected and unaffected family members and trying to avoid watching for symptoms too closely. Many everyday lapses in thinking and changes in mood are normal and not sinister signs of the disease.

1. Cognitive difficulties are common in HD but their pattern is variable within and between families
2. These difficulties come from inefficiencies in how the brain processes information
3. The most frequent changes are in attention/concentration, memory and planning
4. A neuropsychological assessment will identify which aspects are weak and which are well maintained for each individual
5. Depression, worry, and behavioural difficulties are all common symptoms and psychological help is effective in managing these difficulties

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GENETIC ASPECTS OF HUNTINGTON'S DISEASE

INHERITANCE OF HD

HD is passed from one generation to the next because of an alteration in one of the many genes each of us inherits from our parents. The gene that causes HD is called IT15. It is inherited in an autosomal dominant manner. This means that if either parent has the altered HD gene, each son and daughter has a 50% or 1 in 2 chance of inheriting or not inheriting Huntington's Disease.

A fifty per cent chance does not mean that exactly half the children will get the disease in a family where the altered HD gene is known to be present. Each individual child of a person with an altered HD gene stands a 50% or 1 in 2 chance at the moment of conception of inheriting the altered HD gene. This could mean, for example, that one child in a family of four children will develop HD, or two may get it, or three, or perhaps all four, or none. Each person faces his or her own fifty per cent chance irrespective of whether any of his or her brothers or sisters is affected or not. An altered HD gene never skips a generation. It does not appear in one generation, skip the next, then reappear in a third or subsequent generation. If a person does not have an altered HD gene, they cannot pass HD on to their children.

However, not everybody with HD will have a clearly affected parent. Sometimes a parent who has an altered HD gene can die from something unrelated to HD, before ever showing signs of HD. In that case, their son or daughter could be affected, without an apparently affected parent.

THE GENETIC TEST

Discovery of the HD gene

The gene which when altered causes HD was isolated in March 1993. The IT15 gene is located on the tip of the short arm of chromosome 4. The abnormality which causes HD is an expansion of the DNA sequence of an otherwise normal gene.

Normal huntingtin genes contain 10-35 *"CAG repeats"*. Repeat sizes of 27-35 are at the upper end of the normal range, and will not result in Huntington's disease, but sometimes increase into the abnormal range in future generations.

36-39 repeats are at the low end of the abnormal range; an individual with this result may develop HD or may live a normal lifespan without developing the condition. People with 40 or more repeats will develop Huntington's disease if they live a normal life span.

What is a diagnostic and a predictive test?

A diagnostic test is used to confirm the diagnosis of HD in a person showing symptoms and signs of the disease. The test is carried out to confirm a clinical likelihood that a person already has HD.

A predictive test for HD is carried out in a healthy person, who has no signs or symptoms of HD, but who has a family history of HD. The predictive test will determine whether that person has a HD gene alteration. If the person has a HD gene alteration, they will almost certainly develop the condition in their lifetime. Not everyone with a family history of HD wishes to pursue a predictive genetic HD test, and having the test remains an individual choice.

The results of the test would be one of the following three possibilities:

1. A person has inherited an altered HD gene
2. A person has inherited a normal HD gene
3. Rarely, the tests result is uninformative

Why have genetic counselling?

Genetic counselling gives people who wish to have a predictive HD gene test the opportunity to discuss the issues involved in testing, to have their questions answered, and to consider the possible consequences of a predictive HD gene test result.

Once a person starts on the predictive test programme, a partner, close friend or family member should come to the appointments. At least three appointments are usually necessary before testing. A referral to a psychiatrist is made, to make sure that a person is able to deal with an unfavourable result, and also for follow-up if necessary when a result is unfavourable.

A person receiving their test result, needs to be accompanied to that appointment.

Why are partners or close friends asked to attend?

It is extremely important that a partner close friend or relative accompanies the individual to their appointments.

The result may have implications for members of the family, therefore they should have the opportunity to consider the implications of the result for themselves. Individuals accompanying a person should understand the anxieties the person may be experiencing.

Can children have a predictive test for HD?

No. Usually only adults are offered predictive testing. This is because the test is voluntary, and children usually cannot understand the complex issues involved in the decision to take the predictive test. In addition, children cannot legally themselves give consent for the test.

GENETIC CLINICS

If any person at risk of HD is interested in finding out more about the genetic aspects of HD, or about the predictive genetic test, they can talk to a clinical geneticist or genetic counsellor. The National Centre for Medical Genetics holds genetic clinics in Dublin, Cork, Galway and Limerick. More details are available from the National Centre for Medical Genetics (01 409 6739) **www.genetics.ie** or the HD Association (01 872 1303) **www.huntingtons.ie**

GENETIC COUNSELLING

The definition of genetic counselling is *"the process by which patients or relatives at risk of a disorder that may be hereditary are advised of the consequences of the disorder, the probability of developing or transmitting it, and of the ways in which this may be prevented, avoided or ameliorated"*

(P. Harper - 1983).

Genetic counselling is mainly an educational process that seeks to help those affected by HD or at risk of HD to understand the genetics of the disorder, the way in which it may have been inherited, and the options that are available to them in management and further reoccurrence.

TALKING TO CHILDREN

There are two main issues to consider when talking to children about Huntington's Disease. The first relates to talking to them about an affected parent, relative or friend, in order to explain the effects of the illness. The second concerns the genetic and hereditary nature of the illness and the implications for the child.

If someone in the family has HD, it will be difficult to avoid giving a child some sort of explanation. Small children are more accepting than adults are, but become less so as they grow older and therefore need to know that someone is actually ill in order to understand and accept certain behaviour.

Children are also very sensitive to atmosphere, and if they realise that something is not being discussed, can imagine all kinds of things as the cause of the family *'secret'*. They may feel they are in some way to blame and this anxiety and guilt can be more harmful than knowing the facts.

Ideally the telling should be a gradual sharing so that children can grow into an understanding made possible by parents acting openly and answering questions as truthfully as possible appropriate to the child or young person's age. It may help to think about the type of questions that your child may ask and to have prepared some answers.

Parents should be as reassuring as possible without denying the risks. As the child gets older it is important that they can discuss their worries and feelings in an environment where their feelings are accepted and understood.

All families are different so how, and when, may be dependent on the family and the individual child e.g. is it a family who talks about things over the dinner table together? Or does one parent (or on occasion grandparent) usually take the lead in talking about things? Do some children need to be told more information than others (e.g. older siblings versus younger siblings)?

Some parents may be able to face this task on their own, but many will find it difficult and may need to find someone with whom to talk over their own feelings and the needs of the child.

This could be a relative or close friend, doctor, social worker, HDAI staff or other professional. The important thing is to find someone with an understanding both of HD and of children, who will assist you to work out the best approach.

Moodiness and irritability can make life very difficult for the carer and children. The first and probably most difficult step to face is the truth, that a family member is affected. The second step is to speak openly about the situation. This won't come easily but it is important to work toward it. The whole family must understand what is happening.

Truth and honesty within the family helps friends and relatives feel more comfortable in their relationships with the person with HD and makes it easier to lend much needed help and support.

COPING WITH BEING AT RISK

Studies of people at risk have shown that each individual reacts to this challenge in a variety of ways. Even though there is just as great a mathematical chance that the person will escape the disease as there is that they will get it, for some people being at risk means a constant struggle to master the odds and they may live lives of dread, denial, fear, emotional disarray and gloom.

Understandably at times this struggle makes their situation seem much worse but at other times they can live with it.

Some people respond to the risk situation by trying to ignore the disease. Pretending that it either does not exist or couldn't possibly touch them, they push the reality of the disease out of their minds. They do not talk about it nor do they seriously consider it when they contemplate marriage and a family. Unfortunately, the disease itself may not co-operate with this attitude of denial. It cannot be wished away.

Families may be helped by recognising a five step coping process we may all experience:

1. **Denial**
   The person at risk to HD may refuse to accept the information and says *"No, it couldn't happen to me."* Sometimes the person accepts the information well, but later cannot recall the details of what was said.
2. **Anxiety**
   The person can suffer headaches, fatigue, insomnia and irritability as a response to fear. They may benefit from emotional support at this time.
3. **Anger**
   The person sees his/her misfortune as *"the act of a cruel and uncaring world"* and may be openly hostile to friends, relatives and health professionals. There may be a great deal of angry behaviour. Resentment builds between family members. Counselling helps to channel the guilt and redirect the anger in constructive ways.

4. **Depression**
   This is a critical phase, necessary for eventual readjustment. The person will need support to accept changes, try new behaviours and plan for a new way of life.
5. **Stability**
   The person at risk *"cycles"* back and forth between these five phases but with family and professional support and reinforcement, can be helped to live with the threat of HD.

RELATIONSHIPS

If you have never seen or even heard of HD, you may try to ignore the risks and possible consequences. Some of you will feel so frightened that you break off the relationship almost immediately without going further into the subject. Learning the real facts and weighing up the risks and what they mean to you, against the quality of the relationship, is the only way a proper decision can be taken. If you understand the implications and can work out plans for the possibility that your partner either may or may not get HD, then your relationship can be happy and a success whatever happens.

This is an extremely complex area but the right of all individuals to make decisions, provided they are well informed, should be respected. If people have sufficient information it will help them to make an informed decision.

If you are in a relationship with someone at risk they may not have told you because they may never have *"told"* themselves.

WHETHER OR NOT TO HAVE CHILDREN

The hereditary nature of HD, makes the prospect of starting a family particularly difficult. However, many individuals at risk to HD have already established families before they learn about HD or fully understand the hereditary nature of it. Some who fully understand HD and its hereditary implications may choose to have their own children. Others at risk may decide not to have children of their own in order to avoid passing the disease on to another generation.

Through genetic counselling the full implications of the genetic characteristics of HD and reproductive options should be discussed and all the alternatives available should be considered. In other words people at risk to HD should know all the facts before making decisions appropriate to their individual situations.

4 PRACTICAL PROBLEMS

ROLE CHANGES

Roles within the family are likely to change as the person with HD is unable to complete all their previous tasks. Carers/ partners may require support to adapt to increased responsibilities.

They may also need financial assistance due to:

- Loss of income
- Cost of care
- Childcare
- Additional food and clothing

Over time the relationship will become altered, and the pwHD will be less of a friend, companion and lover. This adds personal grief to a complex situation and both individuals may need support to deal with these challenges.

Although working outside the home brings it's own problems and worries about what is happening in your absence, it can also be a relief from the demands of home and in fact can help you cope with the physical and emotional problems because you are not subjected to them all the time.

SEXUAL AND MARITAL ISSUES

Problems related to sexual adjustment for people with a lengthy illness are of great concern both to the patient and the spouse/partner.

These anxieties are often hidden. Even in the midst of the current abundance of information about sexuality, one can find little about sexual adjustment in conditions such as HD.

Hopefully each partner will feel free to discuss their needs with the other. In the words of someone in this situation- *"thoughtfulness and consideration can work wonders"*. Professional guidance may also be sought through the family doctor.

FAMILY TENSIONS

Common experiences of people with neurological conditions and their families include lack of appropriate information, social isolation, high levels of stress, financial strain and changes in family roles. The hereditary aspect of HD has implications for the extended family. This can cause further tension as individuals cope in different ways. Some use exercise as a means of reducing stress, others prefer to keep it private and think things through for themselves whereas others need to talk to family and friends.

Some may find it hurtful if they are not getting the support they expected from other family members. There is no right or wrong way to cope but it helps if you know what works best for you and for your family members. Good psychological supports can reduce stress and uncertainty. Get in touch with HDAI or talk to your GP if you need support.

STOPPING WORK

The person's ability to continue working is frequently a critical issue. The length of time somebody can work will depend on the progression of the disease and the kind of job. It may be difficult for the person concerned to admit that he or she can no longer do the job. They may need some help to accept that there is a problem.

Occupations which are potentially dangerous such as welding, or intellectually demanding such as accountancy, may have to be given up earlier than others. Some employers can be sympathetic and may be able to offer alternatives, though this may not be acceptable to the person in terms of income or status.

It may be beneficial to seek independent advice before making a decision.

Your doctor could intervene if, for instance, you were worried about the person driving or where their actions could put themselves or others in danger.

INCOME AND FINANCIAL MATTERS

The employment of the carer is also an important factor. You may already have been the main earner in which case you will have to decide whether to carry on as before while bearing the extra stresses of organising the home and being a carer.

If you were the secondary or part-time earner before, you will have to think about whether your income will be sufficient to maintain the family, or how you can supplement it. If you were not going out to work at all, you may consider whether a job is an option or whether family demands are too great. It is advisable to consult your solicitor at an early stage in the illness to discuss legal and related issues, e.g. the making of wills, palliative care options etc.

Normal outgoings can be set against expected income. New expenses may have to be taken into account, for example, extra food, extra heating if the house has not been used in the daytime before. Some people move to a house or ground floor flat which is physically more practical and cheaper to run, though this can be disruptive to neighbourhood social life. Probably some financial sacrifices will have to be made, though different people have different priorities as to which they should be.

OTHER WORRIES

Other important worries about HD exist in relation to insurance, mortgages and so on.

If you need information in relation to these or other issues, contact the HDAI office.

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CARE OF THE PERSON WITH HUNTINGTON'S DISEASE

DIET AND NUTRITION

People with HD need a well balanced diet, which includes all the essential nutrients, to prevent or minimise weight loss. A higher than normal calorie intake is often required to maintain normal body weight.

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, and are more prone to catch infections. Evidence suggests that providing a high calorie intake can help toreduce chorea movements, improve cognition and improve speech & swallowing.

- Good nutritional care is essential in the management of HD
- The individual's weight should be monitored regularly and calorie intake adjusted appropriately
- Early individual assessment and regular reviewing of nutritional care plans are vital
- Many individuals have very increased energy requirements and it is essential to provide adequate nutrients
- Nutritional assessment and care planning will vary with the stage of the disease and the individual's eating difficulties

As HD progresses, food will need to be cut in small pieces or liquidised to facilitate chewing and swallowing. The person should eat slowly and without distractions. Hunger and lack of hand control can lead to *"cramming"* at a time when the patient can least cope with such large amounts.

Meals should be smaller but more regular (5-6 per day) with nourishing drinks in between. Many food supplements, rich in protein and calories are available. They can be taken on their own or added to the person's favouite food.

EATING

Early referral to a speech and language therapist will help identify swallowing difficulties. Periodic reassessment can identify changes in swallowing ability and suggest interventions such as a change in food consistency, environment etc. Eating aids such as enlarged grips for silverware and nonslip plates with raised edges to prevent spilling may prolong independent eating. People with HD should be encouraged early in the disease, to eat slowly and deliberately, to sit in an upright position during and after meals, to take small bites, and to clear the mouth of food after each bite by taking sips of liquid.

Individuals should avoid doing other activities (such as talking or watching television) while eating, in order to concentrate on chewing and swallowing.

Speech and language therapy has an important role in the management of HD.

Swallowing difficulties affect most individuals and require management with timely and effective therapeutic intervention.

TIPS FOR GOOD COMMUNICATION

- Reduce distractions
- Offer prompts
- Don't change topics too quickly - ask one question at a time
- Allow time for an answer, Give your full attention
- Keep questions simple - Offer choices (Do you want milk or juice?)
- Don't pretend to understand - Ask if you are unsure

TIPS TOWARDS AVOIDING PROBLEMS

Dental care is important as the patient will have difficulty using dentures.

Incontinence occurs in the later stages of HD because control is lost over the muscles of the bladder and bowels. A regular toilet routine (a two hourly pattern) should be observed. Pads, protective mattresses and absorbent bed sheets provide comfort for the patient and eases the workload of the carer.

Clothes should be comfortable and easy to wash and dry. Items of clothing which are easily worn (e.g. few buttons or hooks) allow the patient greater independence.

Any prospective hazards in the house etc. should be removed, guards placed in front of fires etc. Ensure all electric appliances are safe.

There is a variety of equipment and adaptations to the home which can often be helpful to people with HD such as specialist seating, beds and adaptations to the bathroom. Falls are common and can be a source of severe injury. The Occupational therapist (OT) can give advice on seating and walking aids and can assess the person at home and arrange for rails and banisters to be put in. OTs and Physiotherapists can instruct people in how to sit, stand and walk more safely.

The Primary Care Team can help in the management of choking, infections, chiropody and stress. They can provide advice on communication, recreation, respite care and day centres.

If the person is unwell do not always assume it is HD. It may be another problem, e.g. digestive or chest infection. It is best to contact the GP.

CARER SUPPORT

People with HD and their carers will often need support. Care support choices are dependent on individual circumstances but can include home care, day centre services, respite care and residential care. Referrals to a social worker can help in exploring options. Talk to your G.P. regarding referrals to the appropriate health or social care professional.

Caring for a person with HD can be a huge challenge. The progression of the condition combined with the changing physical, cognitive and emotional symptoms require huge resilience from carers. It is hurtful when your loved one directs their anger at you and hard not to take it personally. Figuring out how to avoid or redirect a person's anger, discovering a new way to approach an old problem or getting support from friends and family or from others in a similar situation can ease the burden.

You need to take care of yourself-not only for you but for the person you care for. If you are too drained, too exhausted or too disillusioned you are putting your own health at risk. Take time out to relax and to rest. Try to focus on the present and your daily achievements rather than stressing about a future which is beyond our control.

HEALTH PROFESSIONALS

A Primary Care Team (PCT) is a team of health professionals who work closely together to meet the needs of the people living in the community. They provide a single point of contact to the health system for the person including:

- GP and Practice Nurse
- Community Nursing Service - Public Health Nurse, Community Registered Nurse
- Occupational Therapist
- Physiotherapist
- Home Help/support staff

The Primary Care Team members also link with other community-based disciplines to ensure all health and social needs are provided for.

These include:

- Speech and Language Therapy
- Dieticians
- Mental Health Services
- Counsellor/Psychologist
- Podiatry
- Social work
- Dental
- Ophthalmic Services

Your GP or Public Health Nurse should provide support in accessing other health professionals.

The Public Health Nurse can give advice and support on many aspects of daily living such as providing nursing care (e.g. help with bathing, dressing, skin and basic care). She/he can advise the family on nutrition, incontinence or support referrals to other professionals.

Occupational Therapist (O.T)

Will provide advice on aids to daily living and can assess what aids or even structural alterations (e.g. home extensions) may be needed.

Physiotherapist

Physiotherapy can help reduce balance and co-ordination difficulties and may also increase muscle power and endurance allowing greater independence for the individual.

Speech Therapist

Speech therapists can advise on methods of maintaining communication skills and can assess swallowing.

Mental Health Services

Seek a referral if the individual has behavioural or psychological problems.

Social Workers

Social workers can offer information and advice to help you get the most appropriate community service. They can assist with accommodation needs both for housing or residential care and assist with complex welfare rights or legal issues. They can set up and monitor family support services if you are in crisis or difficulty.

Home Help

Home helps work with vulnerable people in the community who, through illness or disability, are in need of help with day to day tasks. A home help might visit for a couple of hours per day to help with light housework, shopping, or may provide more personal care such as help with dressing, bathing, etc. You can access this service through your Public Health Nurse.

The Home Care Package scheme

This scheme provides enhanced home care services for people who require additional and specialist support in a home care setting. These enhanced services may include nursing, allied therapy services, day care, respite care and additional home help support.

Community Welfare Officers

Community welfare officers work for the Department of Social Protection but provide clinics from HSE health centres. Community Welfare Officers administer the Supplementary Welfare Allowance Scheme. The types of payments made under this scheme include: weekly Supplementary Welfare Allowance, Rent Supplement, Mortgage Interest Supplement, Diet and Heating Supplements, Back to School Clothing and Footwear Scheme and exceptional needs payments for items such as buggies, clothing, funeral costs etc.

OTHER SERVICES

Meals-on-Wheels

This is a voluntary service available in most areas. The service is for people who cannot cook for themselves. If a special diet is required it should be requested (e.g. - a minced diet or a high calorie diet).

Contact your Local Health Office or public health nurse for more information.

Day Centres

Many voluntary organisations have day care centres, clubs and various facilities throughout the country which can be accessed by patients or carers. Ask your Citizens Information Centre or Health Centre for relevant information.

6 TIPS TO COPE

CARERS WELLBEING

Caring for a sick or elderly person can be very rewarding but it is also very demanding at times. Benefits include: knowing you are doing something meaningful, learning to live in the present and appreciate the simple things in life, re-thinking priorities and values, and an increased insight into your own strengths and limitations. Caring however can be physically, psychologically, emotionally and even financially draining. You will experience a certain amount of stress but it is essential to manage stress successfully and avoid burnout. Chronic exhaustion, significant weight gain or loss and frequent illnesses are physical signs of high stress levels. Frequent crying, frequent irritation, feelings of hopelessness and inadequacy and difficulty controlling ones temper are emotional signs.

In order to prevent being overwhelmed by stress you should:

- Look after your own physical health: eat nutritiously and get adequate exercise and rest
- Take time daily to relax by doing something you enjoy e.g: read something uplifting, listen to music you enjoy, take a walk or phone a friend etc
- Stay connected to friends and outside activities
- Avoid wasting time and energy on unimportant things
- Acknowledge when you need some help and ask for it
- Find at least one person close to you who will listen and understand
- Find out about respite facilities in your area
- Take things one day at a time

THE IMPORTANCE OF RATIONAL THINKING

Fear and panic are normal reactions to stress.

If you experience this it is important to STOP and ask yourself:

- What am I telling myself to make me feel this way?
- Do I really want to do this to myself?
- Do I really want to stay upset?

Relax or distract

- Do something physical, walk, talk, read or listen to music.

Question the negative belief

- What is the evidence for this?
- Is this always true?
- Has this been true in the past?
- What are the odds of this happening?
- What is the worst that could happen?
- What would I do if that happened?
- Am I looking at the whole picture?
- What would I say to a friend in this situation?

Worrying has no effect on solving problems, taking action does.

Contact HDAI or your GP if you need support.

FINANCIAL SUPPORT

Entitlements And Benefits

Your local Health Office, Social Welfare Office or Citizens Information Centre can provide you with relevant information on entitlements and benefits. Entitlements may include:

A **Medical Card** issued by the HSE allows the holder to receive certain health services free of charge. This is a means tested benefit but special consideration is given in the case of HD.

Invalidity Pension is a payment for people who are permanently incapable of work because of illness or incapacity and who satisfy the Pay Related Social Insurance (PRSI) contribution conditions.

Illness Benefit is a payment for people who cannot work due to illness and who satisfy PRSI contribution conditions.

Disability Allowance is a weekly allowance paid to people with a disability who are aged 16 or over and under age 66. You must pass a medical exam, a means test and be habitually resident in Ireland to get the allowance.

Carer's Allowance is a means-tested payment for carers who look after people in need of full-time care and attention on a full time basis. If you are in receipt of another social welfare payment you may qualify for a reduced rate of carer's allowance.

Carer's Benefit is a payment for people who have made social insurance contributions and who have recently left the workforce to look after somebody in need of full-time care and attention. You can get carer's benefit for a total of 2 years for each person being cared for. You may be eligible for carer's leave.

The Respite Care Grant is an annual payment for full-time carers. The payment is made regardless of the carer's means but is subject to certain conditions.

Domiciliary Care Allowance is a monthly payment for a severely disabled child who is under age 16 and needs full-time care and attention far beyond what is normally required by a child of the same age.

Mobility Allowance is a means-tested monthly allowance paid to people who are unable to walk and would benefit from a change in surroundings. For further information, contact your local office of the Health Service Executive.

Dietary Supplement - Any person who is receiving a Social Welfare or HSE payment and who has been prescribed a special diet as a result of a specified medical condition, and whose means are insufficient to meet his/her needs, may qualify for a diet supplement under the supplementary welfare allowance scheme.

Heating Supplement is a payment to help people who have extra heating needs with the cost of heating their home. For example, people that are ill or have a disability.

The **Bereavement Grant** is a social insurance benefit which is payable, subject to certain PRSI contribution conditions. The objective of the scheme is to provide financial assistance to alleviate funeral and other post-bereavement expenses by way of a once-off grant on the death of an insured person. The scheme covers both the insured person and their spouse and dependent children.

SOURCES OF INFORMATION

The Citizens Information Board provides information, advice and advocacy on a broad range of public and social services in an easy-to-understand way. Information is available via:

- Website: **www.citizensinformation.ie**
- Citizens Information Centres
- Citizens Information Phone Service 0761 07 4000.

It also funds and supports:

The Money Advice and Budgeting Service (MABS) - a free and confidential service for people with debt and money management problems. Their services include: help in dealing with debts and making out a budget, examining your income to make sure you are not missing out on any of your entitlements and contacting creditors on your behalf with offers of payment if you are not able to do it yourself.
Tel: 0761 07 2000.
Website: **www.mabs.ie**
Email: helpline@mabs.ie

The National Advocacy Service for people with disabilities. For more information see: **http://www.citizensinformationboard.ie/services/advocacy_services/**

The Department of Social Protection provides information on the relevant supports and services provided by the Department. There is a nationwide network of social welfare local and branch offices. For general enquiries: Telephone 01-8748444, Email info@welfare.ie
Website: **www.welfare.ie**

The Department of Health has overall responsibility for the development of health policy and for the planning of health services. Their website provides information and links to their services: **www.doh.ie**

Information is also available by phone: 01 635 4000 or by writing to Department of Health & Children, Hawkins House, Hawkins Street, Dublin 2.

OTHER VOLUNTARY ORGANISATIONS

The Irish Wheelchair Association works with, and provides services to, people with limited mobility.

Their services include:

Assisted Living Services, Resource & Outreach Centres (ROCs), Motoring Advice, Assessment And Tuition, Parking Permits, Transport, Wheelchair Sales, Rental & Repair and Respite & Holidays.

The Carers Association provides a number of services nationwide, to family Carers, which are aimed at helping to increase the quality of life for the Carer and the person receiving care at home. Their services include: Advocacy and Lobbying, Care Line, Home Respite Service, Information, Membership, Resource Centre, Support Groups and Training.

HOW YOU CAN HELP?

If you have time, energy and skills to contribute, or if you know of a friend or family member who has, please consider volunteering for HDAI. Perhaps you have governance, financial, planning or promotion skills to contribute to our board.

All queries are welcome.

Government funding assists with staffing and office costs while fundraising helps us to cover the cost of outreach, support meetings, carers and youth support, publications, information seminars and awareness.

Thank you to all our wonderful voluntary fundraisers and to those who support them. Our members and friends have been remarkable in participating in charity runs/walks/cycles/dances, line dancing, parachute jumps, school fundraisers, bag packing, charity swims, coffee mornings and social nights.

You have a direct impact on improving the quality of life for people with HD, those at risk, carers and family members.

If you would like information on making a donation or supporting a fundraiser please see our website or contact the HDAI office.

DONATIONS

€100.00 provides a therapeutic day for a person with HD.

€300.00 provides counselling for someone at risk.

You can support our voluntary fundraisers or make a donation online at:

www.huntingtons.ie

If you would like to donate by Cheque or Postal Order:

You can forward a cheque or postal order made payable to **The Huntington's Disease Association of Ireland** and send to:

HDAI,
Carmichael Centre,
North Brunswick Street,
Dublin 7.

Or by Direct Debit to:

Huntington's Disease Association of Ireland Account

Bank of Ireland, Smithfield, Dublin 7.

Account No: 54757711
Branch Code: 90-00-92

INFORMING OTHERS

Family carers have practical day to day experience of looking after someone with HD. They become experts in caring for their loved ones and knowing their likes and dislikes. If a person with HD is availing of respite care or needs continuing care remember to update relevant staff with appropriate information:

- Tips on managing communication
- Diet - food likes and dislikes, how much, how often
- Choking difficulties - useful strategies
- Is help required when eating
- Present emotional state - tips for managing emotional problems
- Degree of mobility - associated risks
- Degree of cognitive impairment and capacity - needs time to process?
- Degree of Incontinence - toilet routine, use of aids
- Sleep pattern
- Medication

Patient's name: _____

Next of kin: _____

Main carer: _____

Information: _____

_____

Communication: _____

_____

_____

_____

Diet: _____

_____

_____

_____

Choking difficulties: _____

_____

Eating: _____

_____

_____

_____

Emotions: _____

_____

Mobility: _____

_____

Cognition: _____

_____

Toilet routine: _____

_____

Sleep pattern: _____

_____

Medication: _____

_____

_____

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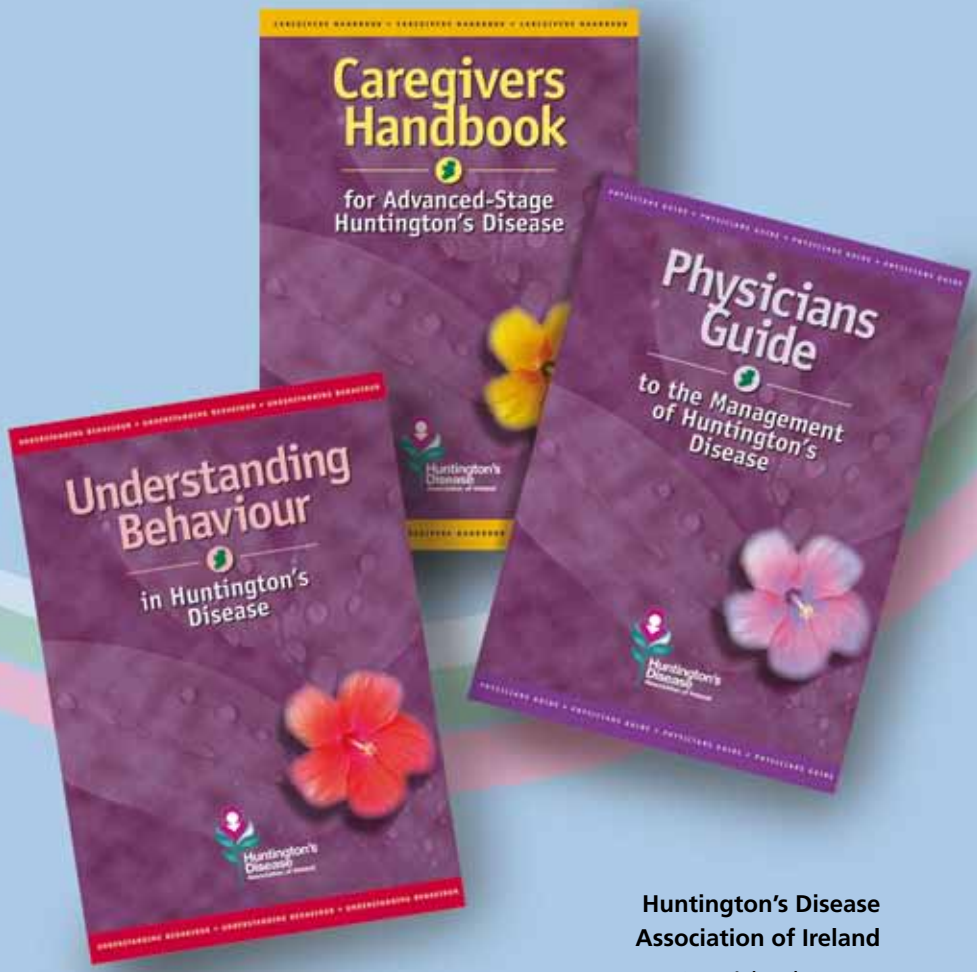
_____

sample list only



Huntington's Disease Association of Ireland

Carmichael Centre
North Brunswick Street
Dublin 7
Tel: 01 872 1303
FreeFone: 1800 393939
Email: info@huntingtons.ie
hdai@indigo.ie

www.huntingtons.ie

INFORMATION REQUEST

If you need further information on any issues mentioned in this booklet please contact:

Huntington's Disease Association of Ireland

Carmichael Centre
North Brunswick Street
Dublin 7
Tel: 01 872 1303
FreeFone: 1800 393939
Email: info@huntingtons.ie
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