



HUNTINGTON'S DISEASE

Huntington's NSW

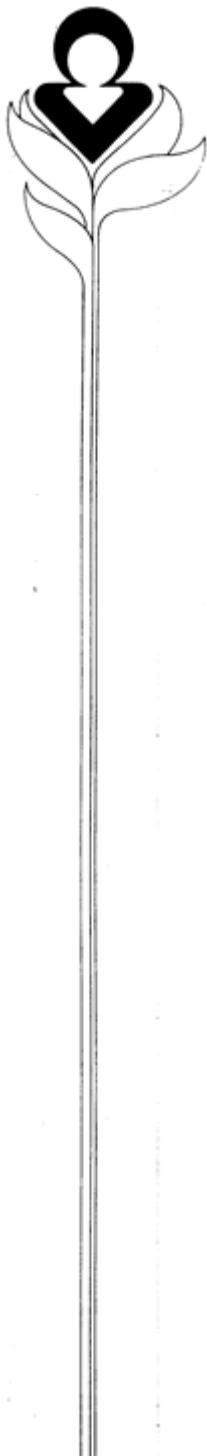
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THE HUNTINGTON SYMBOL

The logo represents the upper torso of an individual, since Huntington's Disease may affect both mental and physical functions. The smaller image inside reflects the reduced physical and/or mental capabilities of an affected person.

The symbol is shown as the flower of a growing, vibrant plant to recognise the growth and development of the work of Huntington's Disease Associations throughout the world.

It is also a sign of hope – a sign that the work and achievements of the past decade are continuing.

AUSTRALIAN HUNTINGTON'S DISEASE ASSOCIATIONS

There is a Huntington's Disease Association in each of the six states of Australia. They were established to develop support and education programs to help people affected by Huntington's Disease; to assist their families to understand and cope; to improve community awareness of Huntington's Disease; and to give families a strong single voice.

For more information please contact the Association in your state:

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HUNTINGTON'S DISEASE

INTRODUCTION

This booklet is designed to help both patients and families understand and cope with the challenge of living with Huntington's Disease (HD). It is intended to provide an overview of HD, rather than cover all the issues in great depth. Because each HD family is unique and because HD manifests itself in a variety of ways, not all the medical symptoms, social implications, difficulties and problems outlined in this publication will be true for all families. All members of an HD family are encouraged to seek the services offered by an HD clinic or an understanding medical practitioner who will provide on-going support, advice, counselling and encouragement.

Huntington's Disease is an inherited, neurological disorder usually characterised by involuntary movement, and often by intellectual impairment and emotional and behavioural problems. HD is usually a "late onset" disease with symptoms generally appearing when the individual reaches his or her mid-thirties or forties. However, in rare instances, the illness may come on in childhood or as late as the seventies.

Although in recent years there has been improvement in treatment, acceleration in research and a better understanding of HD and the needs of the HD family, this disease is still progressive and as yet there is no cure. The gene for HD was isolated in 1993.

Because HD is inherited, it is a family disease. Even though all members of the family will not get HD, everyone is affected emotionally, socially and often financially.



HISTORY AND PREVALENCE

Huntington's Disease was first recognised as an inherited disease in 1872 when a 22 year old American doctor, George Huntington, read a paper 'On Chorea' to a medical academy in Middleport, Ohio. His paper was subsequently 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 a group of dances. It was applied to a number of so called dancing disorders which sprang up in the Middle Ages. In those days individuals with the involuntary muscle jerks and twitches characteristic of HD were often thought to be possessed by devils.

Today we more commonly use the term Huntington's Disease rather than Huntington's Chorea as 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 the physical symptoms, especially in the earlier stages of the illness.

Throughout the eighteenth and nineteenth centuries, chronic adult hereditary chorea was poorly understood. While the gene which causes the disease was probably no less common than it is today, the disorder itself seemed rarer because fewer carriers lived long enough to manifest symptoms. A study of England and Wales compared life expectancy in 1841 of 40.2 years for men and 42.2 years for women with 68.4 and 74.7 years respectively in 1966. Thus in the nineteenth century most carriers could not expect to live long enough for the HD gene to have much effect on them. Now that we live longer, the abnormal gene is given more time to express itself.

The hereditary nature of HD makes a knowledge of family history helpful for correct diagnosis. In the 1930's an American researcher (Vessie) was able to trace almost 1000 cases spanning 12 generations back to three individuals who emigrated from England to America in 1632.

It is difficult to predict accurately, for various reasons, the prevalence of HD. Some people may be incorrectly diagnosed as having another disease, some may conceal the fact that HD is present in their families to avoid discrimination or embarrassment, some may simply be unaware that there is a history of HD in their family because of family breakup, early death or adoption.

The HD prevalence rate in most Western countries is estimated at between five and seven per 100,000 (Folstein 1989). In Australia this would mean that about 1200 people now suffer from the disease and approximately five times that number are at risk (Conneally 1984). There are a few isolated populations of western European origin where the prevalence of HD is unusually high. For example in 1949 Dr Charles Brothers, Director of Mental Hygiene, traced all 86 known cases in Tasmania to a woman of French Huguenot stock who emigrated to England in the 1840's. This woman had thirteen children, many of whom later developed HD. The prevalence of HD for Tasmania is now estimated at 12.1 per 100,000 of population (Pridmore 1990).

In 1999 Dr Elizabeth McCusker and her colleagues at Westmead Hospital undertook a prevalence study in NSW based on the 1996 census. This study concluded that the prevalence for NSW was 6.28 per 100,000. This translates to 378 people with a diagnosis of HD in NSW although this may be an underestimate of the actual number of people with HD as some may be affected but not diagnosed.

A very high concentration of HD has also been found in the Lake Maracaibo region of Venezuela. The origin of the family has not been precisely documented but it is thought that the HD gene was introduced by European immigrants or sailors. The prevalence of HD in this population is estimated at 700 per 100,000 (Avila-Giron 1973). It was the study of this extended family and another large HD family in the U.S.A. that led to the discovery in 1983 of the genetic marker for HD on chromosome 4, and subsequently the isolation of the gene for HD in 1993.

FACTS AND IMPACT

Huntington's Disease is caused by the gradual destruction of brain cells, particularly in those parts of the brain known as the basal ganglia and the cerebral cortex. By some mechanism yet unknown, the causative dominant gene, which for years has remained inactive, begins to take its toll. Central nervous system neurons (brain cells) begin to die. Unfortunately they cannot be replaced.

Neurons come in many shapes and sizes, but they may be generally likened in function to a human hand with many thread like fingers called dendrites and a long thin thumb called the axon. A nerve signal in the form of an electrical impulse starts in the dendrite and speeds to the axon where it causes the release of a neurotransmitter which acts across specialised contacts between cells called synapses. In effect, the neuron acts as a tiny

secreting gland, with either excitatory or inhibitory effect on adjacent cells. When excitation exceeds inhibition, a potential for action is created.

In HD this complex neurological system slowly begins to go awry. The gradual destruction of brain cells causes progressive manifestation of symptoms which are somewhat similar (though usually more pronounced) to the normal process of aging. This is also why it is difficult to get at the root cause of HD. It is not entirely unlike trying to find out why all of us grow old and die. On the other hand, because of its very complexity, research into the cause of HD offers much in terms of “carry over” value for related neurological disorders and for understanding the normal process of growing old.

What are the early symptoms of HD?

The first symptoms of the actual presence of HD may be reflected in slight physical, intellectual or emotional impairment. Physical symptoms may be initially demonstrated in the forms of “nervous” activity, fidgeting, a twitching in the extremities, or excessive restlessness. The individual may notice a clumsiness, alterations in handwriting, or difficulty with normal daily physical skills like driving. If the disorder is actually present, these motor systems will gradually develop into the more marked involuntary movements – jerks and twitches of the head, neck, arms and legs which may lead to difficulties in walking, speech and swallowing. There are exceptions to this. Sometimes people with HD have a minimum of difficulty with involuntary movements and may not be aware of them. In addition, drugs have been developed which help control the involuntary action of the muscles. Where chorea is present, the movements tend to increase during voluntary effort, stress or excitement, decrease during rest, and may disappear entirely during sleep.

In addition to the physical symptoms of HD there are often very subtle intellectual signs as well. These may involve little more than a reduced ability to organise routine matters or to cope effectively with novel situations. There may be a loss of recent memory. All of these signs add to the concerns of the person living at risk of HD because on an “off day” all of us experience such things. It is quite normal for healthy people to be somewhat clumsy, or a bit fidgety when anxious or under stress, or to twitch or jerk when dropping off to sleep. At risk individuals are well advised not to worry too much about the occasional stumble or forgetting a phone number as these experiences happen to all of us.

Early emotional symptoms are usually subtle. There may be an acceleration of certain aspects of the individual’s normal make up – periods

of depression, apathy, irritability, impulsiveness. Here again, from time to time, all of us are depressed, apathetic, irritable or impulsive. A person at risk of HD must not assume that every emotional outburst or bout of depression is an inevitable sign of the onset of Huntington's Disease. Any person who becomes concerned about changes in their physical, intellectual or emotional behaviour and is known to be at risk for HD should consult a doctor who is familiar with the diagnosis and treatment of Huntington's Disease and related neurological disorders.

Later Onset of HD

Approximately 25% of persons affected by Huntington's Disease develop physical symptoms (usually chorea) after the age of 60 years. In this situation, symptoms may progress more slowly and are usually less severe than onset in mid-life. Also, the onset of symptoms is likely to have a less disruptive effect on the person's life and that of their family; the person may have retired (or be close to retiring) and children will have reached their late teens or adulthood. The family may also be more financially secure. The person will probably be able to continue his or her usual retirement activities for many years, and may not require full nursing care until quite an old age.

Juvenile Onset

A small percentage of people with HD manifest a form of the disease characterised by muscular rigidity rather than chorea. This is called the Westphal variant of the disorder. In a small minority of cases the disease can also appear in children. It is worth noticing that there are at least 50 different conditions where involuntary muscle movements may be present. Seeing a doctor experienced in the diagnosis and treatment of neurological disorders is the best way to resolve questions if you fear that HD may be present.

How does HD progress?

Huntington's Disease is a progressive illness, although the rate of progression varies from individual to individual. Dr Ira Shoulson and his team from Rochester, U.S.A. believe that "the surprising functional capacities of these patients suggest that the course of HD may not be as rapidly progressive or incapacitating as is widely believed". In the absence of any standardised assessment of physical, intellectual or emotional incapacity among people with HD, Dr Shoulson and his co-workers have proposed the adoption of a five-stage functional approach to coping with HD. Briefly the stages are as follows:

- 1 **Early Stage** - a person diagnosed as having HD who remains fully functional in both his or her family unit and at work.
- 2 **Early Intermediate Stage** - the person remains employable but at a lower capacity. Self management of daily affairs continues despite some incapacity.
- 3 **Late Intermediate Stage** - the person with HD is no longer employable or can no longer manage household responsibilities. Considerable assistance or supervision is required in handling daily financial affairs. Activities of daily living may be marginally impaired but individuals usually require only slight assistance in such functions.
- 4 **Early Advanced Stage** - the person is no longer independent in activities of daily living but is still able to live within the home environment supported by family or professional assistance.
- 5 **Advanced Stage** - the person with HD requires complete support in activities of daily living and professional nursing assistance is usually necessary.

This staging system is deliberately general and flexible to allow for the varied characteristics and progression of individual cases. It does however indicate how long many people with HD may remain independent and self reliant within the family unit and home environment, related of course to such circumstances as family resources and the age of children.

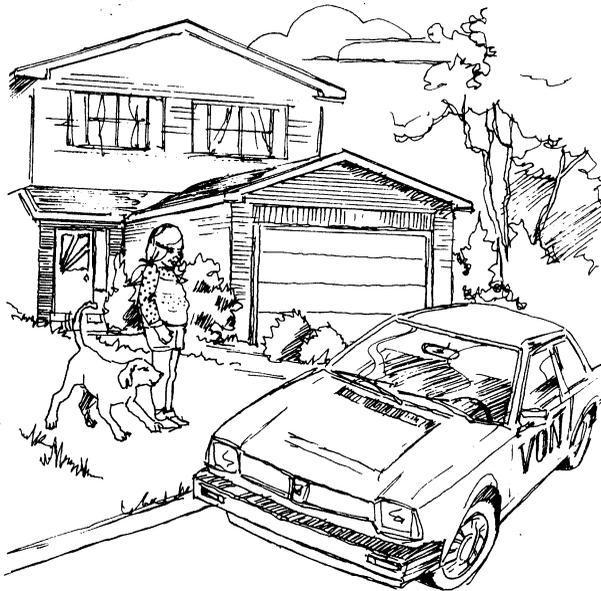
Death usually occurs some 15 to 20 years after the initial onset of symptoms. The cause of death is not the disease itself, but complications such as pneumonia, heart failure, or infection developing from the weakened condition of the body.

What is the hereditary pattern of HD?

Huntington's Disease is passed from one generation to the next because of a defect associated with one of the many genes each one of us inherits from our parents. The gene that causes HD is called a dominant gene. This means that if either parent has this gene, each son and daughter has a 50/50 chance of escaping or inheriting HD and is said to be "at risk". The fifty percent at risk factor does not mean that exactly half the children will get the disease in a family where the HD gene is known to be present. Each individual child stands a 50/50 chance at the moment of conception of inheriting the normal 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. The prospects are similar to the flip of a coin. If you flip a coin six times, you may or may not get three heads and three tails. The combination may be six heads and no tails, or four tails and two heads, and so on. Any combination is possible. But if you flip the coin many times, the average of heads and tails will be about equal. So it is with HD. Each person faces his or her own fifty percent risk irrespective of whether any of his or her brothers or sisters are affected or not. However when large numbers of families containing many children are studied, the proportion one in two (50/50) affected is regularly seen.

Huntington's Disease does not appear in one generation, skip the next, then reappear in a third or subsequent generation. In other words, if a parent who carries the gene for HD has a child who escapes the disease, then the child cannot pass the risk on to his or her children. From time to time, HD may **appear** to skip a generation. Consider for example, a theoretical case where a grandfather had the disease, his son apparently escaped but the son's daughter eventually developed the symptoms of HD. Rather than having skipped a generation we know that the defective gene was also present in the son but he died, perhaps in an accident or from a heart attack before symptoms appeared. Sometimes symptoms can appear relatively late in life.



Predictive Test

In the past a person known to be at risk of HD had to live a full and normal lifespan without showing any sign of the disease before his or her children could rest assured that they and their children were free from risk. However, there have been important and exciting discoveries concerning the genetics of HD. These discoveries now enable people who are at risk of HD to learn whether or not they have inherited the HD gene before symptoms appear.

Predictive testing for HD is a very personal decision which should be considered carefully. The implications for each individual are wide ranging and varied. Consequently, testing is carried out through a program which includes appropriate counselling.

It is important for the person at risk for HD to consider very carefully whether or not this test is right for them. Simply because the procedure is technically possible does not mean that it should be used. Each person must assess his or her own unique circumstances in deciding whether or not to have the test.

If you have just learned that you are at risk for HD, you may be tempted to rush off and have the test straight away. It is important that you are prepared for the possibility of learning that you have the gene for HD. Consider that, if being at risk is upsetting, learning that you have the HD gene may be much more disturbing. Taking time to adjust to the idea of being at risk before proceeding with the test is advisable.

If any person at risk of HD is interested in finding out more about the predictive test, it is important that he or she talk to a clinical geneticist, genetic counsellor or social worker who is familiar with the predictive test for Huntington's Disease.

Prenatal Testing

It is also possible to have a prenatal form of the predictive test for HD. The purpose of prenatal testing is to screen the foetus in order to avoid passing on the HD gene to the next generation. It is important to understand that prenatal testing involves the possibility of termination of the pregnancy if the foetus is found to be at high risk of having the gene for HD. It is very important that a lot of careful thought be given about prenatal testing and pregnancy termination, **well in advance** of a pregnancy. Once a pregnancy has occurred, there is very little time to absorb the information about the prenatal test and to make important decisions.

Again it is most important that anyone interested in the prenatal test for HD consult a clinical geneticist, genetic counsellor or a social worker involved in a predictive testing program for more in-depth information and preferably before a pregnancy occurs.

A new form of prenatal testing has been developed which avoids the need for termination of a pregnancy in which the foetus has inherited the HD gene. This form of testing is called pre-implantation genetic diagnosis (PGD). PGD involves the creation of embryos in vitro (i.e. in a test tube) which are then tested to see if they have the HD mutation. Only embryos which do not have the HD mutation are then replaced in the female partner where it is hoped that this will lead to a successful pregnancy.

More information about the PGD procedure can be obtained by consulting a clinical geneticist, genetic counsellor or social worker involved in a predictive testing program.

How do others cope with being at risk?

Studies of people at risk have shown that individuals react 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 with dread, denial and fear. Understandably at times, this struggle makes their situation seem much worse but at other times, they can live with it.

Others respond to the 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 doesn't co-operate with this attitude of denial. It cannot be washed away. A parent who carries the gene but who ignores the disorder, or avoids discussion of it still has a 50/50 chance of passing it on to children.

Some at risk people seem to fantasise the period following diagnosis to be a prolonged and unproductive wait on death row. They mistakenly perceive the end of their lives to be, not at the moment of death, but at the moment when they are diagnosed as having HD. They ignore the years of productive, self-reliant living which are ahead of them, even if they do have the disease.

In contrast there are those at risk who work through the facts about HD and learn to accept and cope with their situation rather than ignore or deny it.

These people live active and productive lives based on a realistic appraisal of the circumstances facing them. They learn to cope constructively with the 50/50 genetic possibility, determined that even if they get HD they will retain their productivity and self reliance for as long as possible. There is some evidence that this determined and positive attitude, especially when supported by loving friends and relatives may have some ameliorating effects upon the disease itself. Certainly it reduces the spectre of the disease and guarantees that at risk individuals will not ruin their lives fretting over a disorder from which they may never suffer. If the worst happens and the disease does appear, a determined and positive attitude helps the individual to deal with it more effectively.



Many individuals living at risk cope quite well without periods of excessive depression or withdrawal. Generally speaking those people who cope best are those who have known about HD from childhood and have discussed it freely and openly with relatives and close friends. Out of this open approach can come triumph rather than tragedy. There are numerous examples of couples who have very successful and happy marriages in full awareness that one is living at risk of eventually developing HD. Among these people there is often a remarkable appreciation for happiness and quality of life which many others take for granted. Out of possible adversity comes an enriched perspective and a whole new range of more satisfying values and relationships.

Dr Nancy Wexler, President of the Hereditary Disease Foundation in the U.S.A., has noted that “every at risk individual is continually on the alert for any suspicious signs of the disorder”. These people often feel as if they are living on a time bomb. Normal muscular twitches, awkwardness, psychological lapses, forgetfulness are frequently misinterpreted as an early indication of HD, thus while getting HD is only a 50% risk, “many at risk make themselves 100% miserable worrying while they are healthy”. Family members and close friends who are not themselves in any danger may make things worse by constantly observing the individual at risk for telltale signs of the disease.

In her work with individuals at risk, Dr Wexler has found that one by-product of this watchfulness and anticipation was the certainty on the part of 75% of her at risk respondents that they would eventually become afflicted with the disease. The 50% risk of developing HD should also be regarded as 50% probability of **not** developing HD. Dr Wexler concludes that at risk people often translate the 50/50 risk into 100% certainty that they will or will not develop HD. Such convictions usually fluctuate from day to day, month to month.

People living at risk of developing HD often experience a whole series of phases in their attitude towards the disorder. One person who actually got HD recalled a period of two years thinking about it and experiencing “depression, withdrawal, quietness, secretiveness and ruminating about whether all the struggles are worthwhile”. Eventually he emerged from all that and plunged into a program of community action to combat HD – activity that continued at a lively pace for more than a decade after he had been diagnosed with Huntington’s.

Should people at risk have children?

The hereditary nature of HD, combined with the problem of late onset makes contemplation of marriage and the prospect of starting a family particularly difficult. When a person at risk is contemplating marriage, it is absolutely essential that the prospective partner is told the facts about HD. However, many at risk individuals 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 take the risk of passing it on to 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. Some couples may take advantage of predictive testing, either the adult form of the test or the prenatal test. Everyone has a responsibility to future generations. Through genetic counselling, the full implications of genetic characteristics of HD should be discussed and all the alternatives available (e.g. predictive and prenatal testing, donor programs, pre-implantation genetic diagnosis) should be considered. In other words, at risk people should know all the facts before making decisions appropriate to their individual situations.

Do other HD families experience severe tension at times?

Some of the symptoms of HD, particularly denial, moodiness, irritability, personality change or violent outbursts at the slightest provocation can make life very difficult for the family. HD people may need help with their legal and social affairs, or require more care than can be given by the family at home. Unfortunately the person with HD is not always in the position to realise when this does occur and tense family relationships result. It is wise, therefore, to consult the family doctor or the social worker at the HD clinic for advice. In addition, through the Huntington's Disease Association, other families who have handled these situations are often willing to share how they coped, and offer understanding and support.

How does one cope with denial?

Denial may take many forms such as refusing to see a doctor, saying there is nothing wrong, or seeking the opinions of many doctors and hoping for a different diagnosis. Relatives of an at risk individual who is having problems, or a diagnosed person, may also deny the existence of difficulties and blame the spouse, for example. Denial or secrecy may be chosen because of fear of discrimination in such areas as employment, promotion, insurance or social acceptance.

In some cases the person's ability to cope with the diagnosis can be made easier depending on who does the explaining. The explanation of an understanding spouse or relative may be easier for the patient to take than

the more clinical analysis of a doctor. On the other hand some people when initially diagnosed will take more notice of the family doctor, neurologist, psychiatrist or social worker, especially if they are members of a clinic specifically for HD.

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

- 1 **Denial:** The HD person appears stunned or dazed, refuses to accept the information and says “No, it couldn’t happen to me”. Sometimes the person accepts the diagnosis well, but later cannot recall what was said. A lack of insight is often due to changes in the brain, particularly the frontal lobe, so that the individual is not able to recognise the changes that are occurring.
- 2 **Anxiety:** In response to the fears of what could happen, the person suffers headaches, fatigue, insomnia, irritability and may need support and counselling or treatment to overcome these symptoms.
- 3 **Anger:** The person sees his/her misfortune as an 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 up between family members, particularly between patient and spouse. Counselling attempts to channel the guilt and redirect the anger in constructive ways.
- 4 **Depression:** This is a critical phase, necessary for eventual readjustment. Severe depression often responds well to treatment with medication and counselling. With acceptance, the person begins another way of life.
- 5 **Stability:** The HD person “cycles” back and forth between these phases but with family and professional support and reinforcement, the HD person can be helped to LIVE WITH HD.

As mentioned previously, moodiness and irritability can make life very difficult for the children and spouse. 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 towards it. The whole family must understand what is happening. For children to realise that it is an illness, and not them, which is causing a parent’s outbursts may be very helpful. Various articles on the **needs of children** are available on request from the HD Association.

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

Does a diagnosis of HD mean there has to be an immediate change of lifestyle?

Depending on the stage of illness when the diagnosis is made the person may be able to continue with his/her usual activities for some time without any adjustments. The person with HD should be encouraged to carry on normally for as long as possible. If he/she is having difficulties with work, it is better that the person negotiates a lighter or easier job rather than retire from work altogether. It can be argued that these adjustments will be made more easily if the person concerned has been allowed to take full advantage of all educational opportunities in early life. Similarly it is better for the affected housewife to continue with her domestic duties as long as possible, although it may be necessary for another member of the family to help with the shopping and some household chores. With support the person can continue to feel useful, even though somewhat incapacitated. This is better than feeling a burden, which often happens if a patient gives up work completely or if a housewife's duties are taken over entirely by someone else.

Is it common for families coping with HD to feel isolated?

Chronic illness, and HD is no exception, tends to isolate family members from people just when they need them most. Friends may not be comfortable dealing with someone who has involuntary movements and/or slurred speech, especially if they have not been fully informed about the condition. The patient may not like to go out socially or shop because of embarrassment about movements, people who stare, etc.

Family members should seek the assistance or support required from friends, family, clubs, social groups, church and/or professionals, eg community nurses, social workers, clergy. You may be shy and/or proud, but to reach out for support could prove positive.

Do HD people and their spouses experience role changes?

Most definitely. A husband who has not previously found himself involved in meal preparation or caring for children may find himself increasingly responsible for these activities. Wives who had not worked outside the home or managed the family finances may have to accept both new roles. This role reversal can be very stressful for both the patient and the spouse. Over time the marriage relationship will become drastically altered, and the HD person will be less of a friend, companion and lover. This adds

personal grief to a complex situation and both spouse and patient may need professional support to deal with these challenges.



How does one adjust to the change in sexual activity?

Problems related to sexual adjustment for people with a lengthy illness are of great concern both to the patient and the spouse. 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. Many spouses experience conflict in deciding how to meet sexual needs which are not being satisfied in the marriage. 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 or the HD clinic.

COPING

Who to talk to

Sometimes a family with HD will work together with a team of health professionals to receive the best care. A neurologist, a psychologist, a psychiatrist, a genetic counsellor, a social worker, and others might see the family at different times for different reasons. Some families prefer to see only one person for all their needs – medical and emotional. Other families can deal best with the problem with their own circle of family and friends and need only some professional help.

Each family must work out the best situation with the resources available to them. Families should keep in mind that their **physical and emotional needs are important and should be met. They should not be afraid to ask questions and make sure they understand the answers.** Families must also remember that needs vary over time and a solution for one stage of life may have to be re-examined at another. Members of the HD Association are also available to talk to families. Through their own personal experiences of coping with HD they are often in a position to make suggestions and offer much needed support.

Professional support and counselling

Unfortunately, some doctors who may deal only rarely with HD, often communicate little more than a sense of hopelessness and futility because of the irreversible nature of HD. This was particularly so in the past, before the establishment of HD clinics. At a time when the family most needs positive support, all they may get is a message of resignation and despair. Diagnosis is made and the patient and family are often frequently left to cope with the future as best they can. Too often the facts concerning HD are unloaded like a bombshell on a patient and/or the family. They may leave the office in a state of shock or depression with no provision for a follow-up appointment to assist in adjusting to the new reality or to help them cope with the personal, medical, financial and legal problems which may arise. Continuing and affirmative counselling is essential for most people who learn that HD is in the family. Ignorance, fear and depression often make HD worse than it need be. Counselling can go a long way towards helping patients and their families retain maximum productivity and acceptance while coping with what is admittedly a difficult and challenging situation.

A diagnosis of Huntington's Disease invariably affects the patient and family, as well as a wide circle of friends, work associates, neighbours and others in the community. As with any serious diagnosis, the patient and

family can expect to experience a variety of emotions, some even frightening or contradictory. Disbelief or denial, rage, despair, guilt, shame, sadness, loneliness, self-pity, envy of others good health, relief that diagnosis has finally been made, thoughts of “why me?” – all these feelings and many others are **frequent** and **normal** reactions.

Feelings may come and go, change and return, and may last many years. After the initial diagnosis, a period of mourning is to be expected in all family members. This is a natural healing process and people need to be allowed to express their feelings. Mourning can also be frightening. Counsellors, specially trained to help people with depression and grief, may be of great help to patients and families with Huntington’s Disease. Some families may also find it useful to consult a marriage counsellor to work out new adjustments and roles for marriage partners. Often the initial symptoms of the illness have placed a strain on the marriage that neither partner could understand until after the diagnosis was made.

Genetic counselling

Because HD is hereditary, genetic counselling can be very helpful. Some genetic counsellors, because of training and experience, will not only inform families about HD and its pattern of inheritance, but will also explain the predictive test and all options that are available in order to make informed decisions about childbearing or other concerns. Others may be willing to directly help families cope with the impact of the diagnosis and understand all the ramifications – emotional, medical, legal and financial – that HD can have. The counsellor may assist with the difficult problems involved in telling others (spouses, siblings and children) about HD.

Genetic counsellors and social workers often work together to ensure that patients and families receive all the services and benefits to which they are entitled. A family may visit a genetic counsellor once, or on several occasions, depending on the needs of the family.

RESEARCH

Scientists in various parts of the world are now at work in an effort to discover the precise cause of Huntington’s Disease. Improved forms of drug therapy are being tested and, ultimately a cure for HD will be found.

In 1983 Dr James Gusella and his team at Massachusetts General Hospital in Boston U.S.A. traced the inheritance of DNA markers in a large Venezuelan family studied by Dr Wexler. They established that the defective gene was located in chromosome 4. Since that time researchers

throughout the world have collaborated in pin-pointing the gene, with the aim of isolating it and understanding how its function is disturbed in people with HD.

In March 1993 it was announced that the HD Collaborative Research Group had found the gene for Huntington's Disease. This international collaboration led and co-ordinated by the Hereditary Disease Foundation consists of six research teams, four in America and two in Britain, who have shared their expertise, research and resources in order to achieve their goals.

A variation in the genetic code for a gene on chromosome 4 was found among people affected with HD. Normally the genetic code of this gene has three DNA bases, which are repeated a number of times. These are known as CAG repeats and there are three possible ranges of repeat numbers. When used as a predictive test, the normal range is **26 repeats** or less. The person within this range will not develop HD and cannot pass on the HD gene. If the number of CAG repeats is **40 or higher** the person is either affected with the HD or will develop HD and may pass it on to his or her children. The repeat range between **27 and 39** is at present regarded as "**intermediate**" meaning that with a CAG repeat number in this range the likelihood of the disease developing is difficult to determine, however the person's children may still be at risk.

For a better understanding of the intermediate range, especially if someone is considering the predictive test, the person concerned should discuss the matter with a geneticist, genetic counsellor or social worker experienced in HD.

Other diseases are now known to have a similar underlying fault or mutation but located on different chromosomes (Spinocerebellar ataxia, myotonic dystrophy, Fragile X syndrome).

Hundreds of HD and normal blood samples were studied to establish that this variation is specific to HD carriers and actually is responsible for HD. Since the isolation of the gene for HD, researchers have identified the expanded CAG code for an altered protein "huntingtin" which appears to adhere to other brain proteins including one involved in energy metabolism. This may be responsible for the nerve cell deterioration.

The discovery of the gene and protein is not a cure for HD but is a major milestone on the road to the cure. It must be emphasised that this development will not immediately provide a treatment for HD but with the

new understanding that it brings it should help efforts towards finding and assessing any new possible treatment.

The scientific community has worked very hard in finding the gene and the process was long and tedious and required great patience and perseverance. Nevertheless the job is not over until an effective treatment is found. The scientists working on HD are brilliant and passionately committed to finding a cure.

Recent research developments include the placing of the HD gene in animals. The animal models are already teaching scientists enormous amounts about the gene. The first transgenic mouse was created by Dr Gill Bates and her colleagues at Guy's Hospital, London. These mice have been extensively studied by an international group of investigators.

Australian researchers are undertaking a range of research projects as well as participating in international research collaborations, such as the Huntington Study Group. These collaborative projects, bringing together the intellectual and financial resources of a large number of research institutions, offer the best prospect for major progress in HD research.

What do I need now?

The presence of HD within any family creates problems and tension for everyone concerned. Each family handles these challenges in different ways. Some people cope better than others and this is often because those who handle the challenge well are quick to utilise the support services available to them. Most families experienced in dealing with HD cite the following as among the most helpful resources:

- The love and support of other family members; being able to share the load.
- The help of a close circle of personal and business friends.
- The services of an experienced neurologist, social worker, and other health professionals, preferably through an HD clinic.
- Membership of the HD Association which provides brochures and information, an informative newsletter, regular meetings, friendships, shared experiences, mutual support, holiday homes, at risk meetings, and other group activities.

- Self-help: dealing realistically, positively, and responsibly with my own situation with respect to HD and other members of my family.
- Not cutting myself off: having someone with whom I can talk and share my feelings.
- Knowing that I am not alone: many people are working to overcome the problems associated with HD.
- Knowing that I too can make a contribution to a collective campaign against HD through my membership of the HD Association and through my efforts to improve the quality of care for those who have HD.

Is there anything I can do now?

- Support the HD Association by becoming a member, through financial donations and by attending association activities and functions.
- If you are a member of an “at risk” family, provide other family members with information about HD and encourage them to join the HD Association. Be sure your doctor is well informed.



- Assist in education, one of our largest tasks, by distributing information which will make the general public more aware of HD, as well as other genetic and neurological diseases.
- Encourage family members to discuss HD and the predictive test openly and honestly. The Association will assist you to obtain appropriate genetic and family counselling.
- Make arrangements to have a family history taken. Again the Association will advise you on whom to contact.

Is there a reason for hope?

Yes. In addition to the work of the Huntington's Association, similar organisations in many countries around the world are pushing forward in the areas of education, patient and family support services, and research. These efforts complement and encourage the work of scientists in many countries, including the World Federation of Neurology Research Group on HD, an international network of scientists who meet every second year to consider the latest research findings. Another vital organisation is the Huntington Study Group which is an international consortium of scientific investigators from academic and research centres who are committed to the co-operative planning, implementation, analysis and reporting of controlled clinical trials and of other therapeutic research for HD. Also important is the increasing information available to doctors, nurses and other health professionals, which improves the level of care offered to patients and families.

In years past, Huntington's Disease demonstrated an understandable but unfortunate propensity to burden all those associated with it with a sense of absolute hopelessness. This feeling of despair made the challenges of living with HD even more difficult than they are already. Little was done to combat the disease because so many people, including scientists and doctors, were too quick to admit defeat.

Fortunately that attitude has changed. We now live in a revolutionary age for genetic research. A few years ago we were predicting scientific breakthroughs for Huntington's Disease. Now we are experiencing them. Equally important, HD has come out of the closet. Fewer and fewer people are making their lot even worse by denying the presence of the disease or attempting to hide from it. More and more people are making use of the scientific services available to them. This combination of scientific progress and an open, INFORMED AND FORTHRIGHT APPROACH BY FAMILIES HERALDS A NEW ERA IN THE TASK OF MANAGING Huntington's

Disease. As mentioned earlier in this booklet, the way ahead will not be easy. We are by no means yet within easy reach of the answers, but we will never return to a past of ignorance, denial, superstition and fear. The great task now is to continue to improve the quality of care and opportunities for those who presently have HD. At the same time each of us has an important responsibility to make what contribution we can to hasten the day when the words "Huntington's Disease" are added to the long list of diseases that man has learned to conquer.

It is better to light one candle than curse the darkness.

ADDITIONAL MATERIALS AVAILABLE

1. Each state association publishes a regular newsletter.
2. Various fact sheets on physiotherapy, occupational therapy, speech, swallowing, diet, nutrition, talking to children.
3. Information and personal stories about the predictive test.
4. Information on the services provided by each state association.
5. Information for medical practitioners on diagnostic testing and the management of Huntington's Disease.
6. *A Physician's Guide to the Management of Huntington Disease, 2nd Edition* by Adam Rosenblatt, MD, Neal G Ranen, MD, *et al.*
7. *Understanding Behaviour in Huntington Disease* by Jane S Paulsen, PhD.
8. *A Caregivers Handbook for Advanced Stage Huntington Disease* Ed. Jim Pollard, MA.

WEBSITES

Huntington's NSW	www.huntingtonsnsw.org.au
Huntington's Qld	www.huntingtonsqld.org.au
Huntington's Vic	www.huntingtonsvic.org.au
Huntington's WA	www.huntingtonswa.org.au
Huntington's SA & NT	www.huntingtonssant.org.au
Huntington's TAS	www.huntingtonstasmania.asn.au
International Huntington Association	www.huntington-assoc.com
Huntington's Youth Organization	en.hdyo.org
HD Buzz (research news)	en.hdbuzz.net
Huntington's Disease Society of America	hdsa.org
Huntington Society of Canada	www.huntingtonsociety.ca
Huntington's Disease Association (UK)	hda.org.uk
Huntington Study Group	huntingtonstudygroup.org
Hereditary Disease Foundation	hdfoundation.org

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