

Understanding Huntington Disease

A Resource for Families

Reprinted 2008

Introduction

Huntington Disease

Huntington disease (HD) is an inherited progressive brain disease. Because it is hereditary, it is a family disease. Although all family members will not be affected physically, everyone will be affected emotionally, socially, and very often financially.

Traditionally, this condition was known as Huntington Chorea. The name Huntington is used as tribute to George Huntington, the American doctor who first reported his description of the disorder in 1872. Chorea is the Greek word for dance, which reflects the involuntary movements associated with HD. The term Huntington disease is now commonly used to refer to the illness. We have learned that some persons with HD display cognitive (a change in thinking abilities) or emotional change, rather than involuntary movements, in the earlier stages of the illness. All areas are affected as the disease progresses.

Huntington disease is now more widely known and understood, thanks in large part to the cooperative efforts of the scientific community and families, professionals and interested friends, working through voluntary associations like the Huntington Society of Canada (HSC), the Huntington Disease Society of America (HDSA) and similar organizations developing throughout the world.

Huntington Society of Canada

The Huntington Society of Canada is a national network of volunteers and professionals united in the fight against HD since 1973.

Our Mission:

The Huntington Society of Canada aspires to a world free from Huntington disease. The Society maximizes the quality of life of people living with HD by delivering services, enables others to understand the disease; and furthers research to slow and to prevent Huntington disease.

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- Delivering services;
- Enabling others to understand the disease and;
- Furthering research to slow and to prevent Huntington disease

International Huntington Association

The **INTERNATIONAL HUNTINGTON ASSOCIATION (IHA)** is a federation of national voluntary health agencies that share common concern for individuals with Huntington's Disease and their families. Each agency promotes lay and professional education, individual and family support, psycho-social, clinical and biomedical research, and ethical and legal considerations related to Huntington's Disease in its respective country.

Primary goals of the International Huntington Association

- to promote international collaboration in the search for a cure for HD
- to maintain close liaison with the research scientists, who form the Research Group on Huntington's Disease of the World Federation of Neurology
- to develop and share information and resources among member countries, to avoid duplication of effort - to assist in organizing and developing new and existing national HD organizations

- to cooperate with other voluntary health agencies and international health organizations
- to publish and distribute an international newsletter to all members and to representatives in those countries where no HD group is not yet organized.

Families with Huntington disease are not alone. They are part of an international network of caring and concerned families and professionals determined to overcome Huntington disease.



Huntington Society Logo

The logo has been designed as a symbolic head and upper torso representing the effect Huntington disease has on the cognitive and physical functions of a human being. Because the disease decreases a person's ability to function normally, a negative or reverse image of the figure was placed inside the original symbol. It is smaller in size to illustrate the reduced physical and/or cognitive abilities of the person with HD.

The logo is shown as the flower of a growing, vibrant plant, which recognizes the growth and development of the Huntington Society of Canada since its founding in 1973. In 1983, this logo was adopted by the International Huntington Association to represent Huntington Societies around the world.

Table of Contents

Introduction

Huntington disease	3
Huntington Society of Canada	3
Mission	4
International Huntington Association	4
Huntington Society Logo	5

Section I

Facts About Huntington Disease	8
Early Symptoms	8
Disease progression	9
Behavioural changes	11
Available treatment	13
Difficulty getting an accurate diagnosis	14
Entitled to a second opinion	14
How common is HD	15
Hereditary pattern	16
Genes	16
Predictive Testing	17
Prenatal Testing	19
Intermediate Alleles	19
Juvenile form of HD	20

Section II

Experiences of Family Members	21
The most difficult experiences	21
The most helpful experiences	23

Section III

Living with Huntington Disease	25
Coping with being at risk	25
Approaching marriage and parenthood	26
Living with increased risk	27
Coping with denial	28
Experience severe tension	30
Experiences role changes	31
Feeling isolated	31

Changes in sexual activity	32
Suicide	34
Change in lifestyle	36
Who to talk to	36
Counsellors	37
Genetic Counselling	38
Therapeutic Intervention	39
Communications & Swallowing Therapy	39
Physical Therapy	40
Occupational Therapy	41
Nutritional Therapy	41
Recreation (Therapy) Activities	41
Music Therapy	42
Other Therapy	42
Safety	43
Driving	43
Smoking	43
Medic-Alert Service	43
Planning	44
Care of the Caregiver	44
What can be done now	45
Reason for hope	47
For Further Information	48

Appendix A

Action Plan for Caregivers	49
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Appendix B

Legal Issues of Living with A Chronic Illness	51
Definition of Legal Terms	52
Power of Attorney	52
Enduring Power of Attorney	52
The Dependant Adults Act	53
Trustee Order	53
Guardianship Order	54
Advance Healthcare Directives	54

Section I

Facts About Huntington Disease

What are the early symptoms of HD?

Early symptoms may appear as slight physical, cognitive or emotional changes. Physical symptoms may initially consist of “nervous” activity, fidgeting, a twitching in the extremities, or excessive restlessness. The individual may notice a certain clumsiness, alterations in handwriting, or difficulty with normal daily physical skills such as driving. These initial motor symptoms will gradually develop into more marked involuntary movements such as jerking and twitching of the head, neck, arms and legs, which may interfere with walking, speaking and swallowing. There are exceptions to this. Sometimes people with HD have a minimum of difficulty with chorea (involuntary movements). Where chorea is present, the movements usually increase during voluntary effort, stress or excitement, tend to decrease during rest, and disappear entirely during sleep for many people.

In addition to the initial physical symptoms of HD, there are often very subtle cognitive signs as well. These may involve little more than a reduced ability to organize routine matters or to cope effectively with new situations. There may be a loss of short-term memory which according to Betty, a recently diagnosed individual, “may occur several times each day”. Work activities may become more time-consuming. Decision making and attention to details may be impaired. “Making choices that involve more than two items becomes very stressful”, reminds Betty.

Early emotional symptoms may be equally subtle. There may be an accentuation of certain aspects of the individual’s normal make-up such as more periods of depression, apathy, irritability, impulsiveness or there may be a change in personality. Rarely, a person may become delusional or unrealistically paranoid.

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 usual 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 or presymptomatic (those with an increased risk after testing) 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 and do not necessarily signify the onset of the disease.

From time to time, all of us are depressed, apathetic, irritable or impulsive. An at risk or presymptomatic person must not assume that every little outburst or bout of depression is the start of HD. Anyone who becomes concerned about changes in his/her physical, cognitive or emotional behaviour should consult a doctor familiar with the detection and treatment of Huntington and related neurological disorders.

The symptoms of HD most frequently appear between the ages of 30 and 45. If symptoms have not appeared by 45, the probability that a person will develop HD grows less with each year of life. Symptoms rarely appear in a person who is 70 or over. The juvenile form is discussed later in this booklet. It must be emphasized that there is much variation and not every person has all the difficulties mentioned here. While some individuals may have a great deal of difficulty with involuntary movements, others have very little. Similarly, some individuals may have marked emotional or cognitive difficulties and others very few.

How does the disease progress?

While research into HD is growing there has been relatively little meticulous research into the progressive development of the disorder in people with HD. One study of 247 American subjects with an average duration of illness of five years from the time they were first diagnosed found that 75 percent of the subjects were living at home and 61 percent were independent and self-reliant in the basic activities of daily living. While such figures are not conclusive, Dr. Ira Shoulson, Professor of Neurology, Pharmacology & Medicine, University of Rochester Medical Centre and co-workers state that “surprising functional capacities of these subjects suggest that the course of HD may not be as rapidly progressive or incapacitating as widely believed”.

Dr. Shoulson and his team have proposed adoption of a five-stage functional approach to coping with the disorder. Briefly, these stages are as follows:

1. **Early Stage** - a diagnosed individual 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 level of capacity. Self-management of daily affairs continues despite some incapacity.
3. **Late Intermediate Stage** - the person is no longer employable or can no longer manage household responsibilities. Considerable assistance or supervision is required in handling daily 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 home care services or minimal professional assistance in a care facility. The Huntington Society of Canada is exploring a number of supportive housing arrangements.
5. **Advanced Stage** - the person requires complete support in activities of daily living. Professional nursing assistance is usually necessary in a long-term care facility.

This staging system is deliberately general and flexible to allow for the varied characteristics and progression of individual cases. How long someone may remain in the home environment is related to many circumstances, including behaviour manifestations, family resources and age of children.

Death usually occurs 15 to 25 years after onset of the disease. The person dies, not from the disease itself, but from complications such as pneumonia, heart failure or infection developing from the weakened condition of the body.

How do you explain the behavioural changes which occur?

The following excerpts from a paper by Dr. Jane Paulsen and Dawn Stoll-Fernandes, MS, in the Dept. of Psychiatry at the University of California at San Diego, help us to understand the behavioural changes.

“The behavioural changes of HD are largely a result of degeneration of select parts of the brain, specifically the *basal ganglia* and within that the *caudate nucleus*. The person with HD is undergoing specific progressive changes in the brain that directly affect body movements, feelings, thinking, perception of the world and behaviour in general. Only when this is understood and accepted can successful and sensitive management of behaviour take place.

“Although HD is commonly characterized by choreic movements, changes in several other behaviours occur as well. In fact, for some individuals and families these other behaviour changes are the primary disability, *not* the chorea. Neuropsychological research has been important in characterizing these behavioural changes as a result of brain changes secondary to the disease and not due to the personality of the individual with HD.”

Some of these behavioural changes include:

- difficulties with memory
- slowed thinking (for example, pauses during conversations, and increased time required to do previously routine tasks)
- difficulties in planning and organization (for example, following a recipe or prioritizing a list of tasks for the day)
- difficulty initiating actions and behaviours, which may result in family members and caregivers assuming the person is apathetic or lazy. (“He just sits in front of the TV all day long”)
- temper outbursts
- impaired ability to perceive body position in the environment (for example, how close your tie is to the soup or difficulty with directions)

Because people with HD remain aware of who they are and their environment, family and professional caregivers often feel frustrated when behavioural difficulties arise. There is sometimes a feeling that problem behaviours could be controlled if they just tried.

Betty, a kindergarten teacher, had no choice but to give up teaching because of HD symptoms. Betty helps us understand the frustration she feels because of the behavioural changes she is experiencing:

“From inside me I’m screaming, ‘My cognitive skills are disappearing—short-term memory—making connections, recalling sequence of numbers or thoughts, making choices etc. Help—somebody, quick! Where are the scientific studies about cognitive losses in HD and how they affect behaviour? relationships?’

“It’s those changes in skills that are causing changes in what I say, think, and in how I behave! It is not my will to change; it is not my person that’s changing; it’s not my commitment that’s changing, it’s not my faith in my Creator nor my faith in my human sojourners that I’m changing. But, there are ‘cracks’ appearing in all of the above.

“You can see when a flag is at half mast. You can’t see which of my cognitive skills are at half mast or non existent. I, the person, am not at half mast. I am still wholly worthy—each person is.”

Nor is the person with HD often aware of when their cognitive skills are at “half mast” to help out the caregiver. Refer also to section on denial.

A more comprehensive treatment of behavioural changes in HD is included in a separate publication *Understanding behaviour in Huntington disease: a practical guide* produced by the Huntington Society of Canada.

What forms of treatment are available for HD?

As yet there is no effective treatment for the condition, but some drugs are now available which are useful in reducing the severity of the abnormal movements. Other drugs are helpful in controlling the depression, moodiness, anxiety and irritability that can occur. The effectiveness of these drugs varies from person to person.

A family doctor will likely refer an individual to a neurologist for diagnosis, treatment and at least an annual or more frequent follow up, depending on each individual situation. The family doctor may then continue to monitor the person.

Antidepressants may be the most useful category of drugs, treating depression, irritability and apathy. Medications are most helpful when an affected person's symptoms interfere with activities of daily living. For those with milder symptoms, medications may not be appropriate. Medications to suppress movements may have short-term and long-term side effects and interfere with activities such as walking and swallowing.

The mainstay of care is providing accurate and comprehensive information in a supportive and caring environment. Betty, in the earlier stages of HD, finds the neuropsychologist she sees every 3 or 4 weeks to consider ways of handling the changes in cognitive skills, to be extremely helpful. Her caregiver team includes her family, family physician, neuropsychologist, physiotherapist, the Huntington Society of Canada Resource Centre Director and her neurologist. "I feel very strongly about this—treatment and drugs are not synonymous, although drugs may be part of treatment sometimes" states Betty emphatically.

More information about medical management is available from *A Physician's Guide to the Management of Huntington Disease*. Contact the National office or visit the website to obtain a copy. The Huntington Society of Canada encourages every family to see that its family physician has a copy.

Sometimes it is difficult to tell if a particular symptom such as apathy or incontinence is a sign of the disease process or a reaction to medication. Fatigue or restlessness, withdrawal or hyperexcitability may also occur as drug side effects. It is best to consult a doctor about any questions in these regards. Monitoring of medication for side effects is important.

It has often been observed that persons with HD are too heavily medicated. However, some medication may help with behaviour difficulties and allow the person to remain in the home environment for a considerably longer period of time than would otherwise be possible. Each situation must be considered individually and will require an assessment of the wishes of the person and immediate family members.

Current treatment strategies offer ways to improve symptoms, but do not alter the underlying disease process. Research is underway to develop ways of slowing or stopping the degenerative process.

Why is it that some families have had great difficulty getting an accurate diagnosis?

Until recently, no test was available to tell if a person showing possible signs and symptoms of HD was indeed carrying the gene that causes HD. In addition, a variety of subtle early symptoms may also be present in other conditions. A physician, very often a neurologist, diagnoses HD by evaluating clinical signs and symptoms and obtaining a family history. It is extremely important for family members to give all relevant information to the doctor who is taking a family history, as this is a vital part of the diagnostic process. Be sure to mention any relatives who have experienced mental problems, or been diagnosed as having other neurological or psychiatric conditions, e.g. Parkinson's disease, schizophrenia.

With our current knowledge, the physician may, in cases where HD is suspected or where there is no, or an inconclusive, family history, request confirmatory testing to see if the gene that causes HD is present.

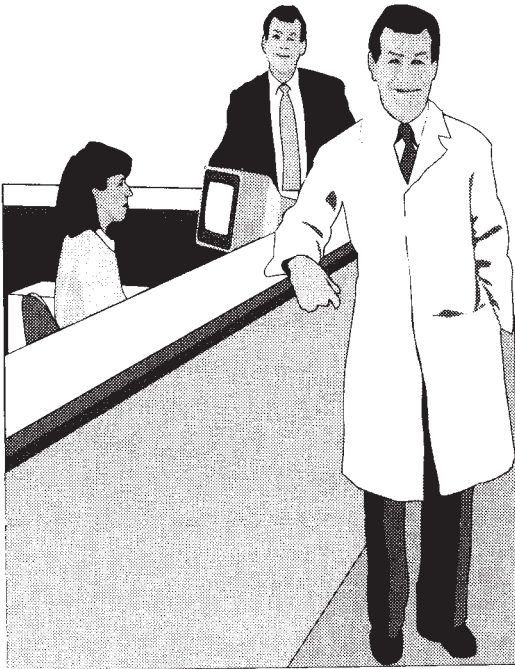
Am I entitled to a second opinion?

You most definitely are. If your family physician is not willing to refer you to a neurologist or a genetics clinic, speak to another physician, or be in touch with the nearest Huntington Society of Canada contact person. It is certainly legitimate to get an opinion from a second neurologist as well. Some physicians are not very familiar with Huntington disease and may not be able to offer enough information or support.

How common is Huntington disease?

For several reasons, it is difficult to predict accurately the prevalence of the disease. Some patients may be diagnosed as having another disease. At death, the cause may be listed as pneumonia or heart failure, rather than HD. In addition, many people conceal the fact that HD is present in their families to avoid discrimination or fear of rejection.

Scientists tell us that, to the best of their knowledge, about 1 in 10,000 persons in North America possess the gene that causes HD and therefore either have or will develop the disease. However, the partner and children of every person with HD and individual at risk must also live with this illness. As a result of this ripple effect, it is estimated that 1 in 1,000 Canadians is directly affected by the problem.



What is the hereditary pattern of the disease?

Huntington disease affects both males and females, and can be passed from one generation to the next by either the mother or father.

The gene that causes HD is called a dominant one. This means that each child of a parent with HD has a 50% risk of having inherited the gene which causes HD.

Sometimes a parent may die before the age when his or her HD symptoms would have appeared. This gives the wrong impression that the illness has skipped a generation. Although each child has a 50:50 chance of escaping or inheriting the disease from an HD parent, in some families all children may inherit it, and in others no children may inherit it. It's like flipping a coin—there's an equal chance to get heads or tails, regardless of the result of the last toss. It's the same chance for each child regardless of whether or not any of his/her brothers or sisters are affected.

Genes

In each cell of our body we have 46 chromosomes which are arranged in 23 pairs. One chromosome from each pair comes from our mother, and the other from our father. Chromosomes are made up of small units of genetic material called *genes*. The gene for HD is located on chromosome number 4.

Genes are made up of deoxyribonucleic acid, or *DNA*. DNA molecules consist of chains of four small elements called bases. There are four bases known as A (adenine), T (thymine), G (guanine) and C (cytosine). Their order constitutes a code which determines the type of protein that the particular gene produces. Any change in the sequence of bases may cause a problem in the functioning of that protein.

The gene causing HD has been shown to have a region in which three of the bases (CAG) are repeated many times (Figure 1).

Figure 1

The CAG repeat sequence with 5 repeats:

/ C-A-G / C-A-G / C-A-G / C-A-G / C-A-G /

The publication Genetic Testing for Huntington Disease indicates that the normal gene containing 35 or fewer CAG repeats, while the disease-causing version has 36 or more repeats. A gene with 36-39 repeats falls into a “reduced penetrance” range, which may or may not be associated with the onset of HD symptoms. Further advances in genetic studies of HD may provide additional or reviews information in the future.

The individual with a “positive” test result means that the individual has inherited DNA changes associated with HD. The CAG repeat on one gene is expanded, usually 36 or more repeats. Except in rare cases, the other gene has a normal number of repeats.

It is extremely important to point out that the actual number of repeats that is called the intermediate range is going to vary from laboratory to laboratory and is becoming more clearly defined with ongoing research.

The actual numbers stated here are used simply for purposes of explanation, and have no individual significance. The repeat size only indicates what category an individual is in, i.e., will not develop HD, intermediate range, or will develop HD. In most situations it does not tell an individual about the onset or course of the illness.

Because our genes come in pairs, each person will have a certain number of repeats in one gene and another number of repeats in the other gene. The repeat sizes can be the same (such as 18 in both genes) or different (such as 18 repeats in one gene and 42 in the other). Each parent passes one of their genes on to their child. Analyzing the number of repeats in this region on each gene can be done on very small samples of DNA, which can be obtained most easily from blood samples.

A more comprehensive explanation with regards to Genetic Testing is included in a separate publication produced by the Huntington Society of Canada.

How can predictive testing for HD be done?

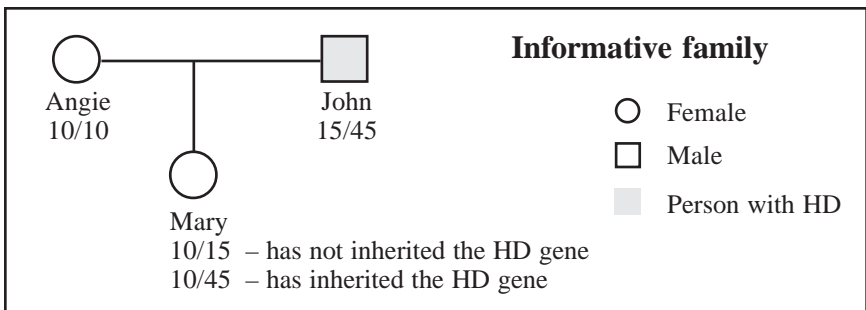
In 1983, genetic markers closely linked to the HD gene were identified. The discovery of additional genetic markers led to the development of predictive testing programs for HD. This indirect

approach to testing using only genetic markers resulted in some inaccuracy. In March 1993, the gene that causes Huntington disease was identified. This means that individuals at risk for HD can now be directly tested to “predict” who will develop this disease.

The following example (Figure 2) illustrates how predictive testing may be done in a family. Mary has requested predictive testing. Her father, John, has Huntington disease. A sample of her father’s DNA is analyzed and it is determined that he has 15 repeats on one chromosome and 45 repeats on the other. The fact that he has a gene with greater than 40 repeats supports the clinical diagnosis of HD. Mary’s DNA can then be assessed. If she has received 10 repeats and 45 repeats, the 10 must have come from her mother and the 45 repeats must have been inherited from her father. This indicates that she has also inherited the gene that causes HD. If, on the other hand, she has inherited 10 repeats and 15 repeats, she received the gene containing the 15 repeats from her father. This is not the gene causing HD and she, thus, will not develop HD.

Predictive testing is most informative and accurate when blood from an affected family member is available. However, testing would still have been possible for Mary in this example, if blood from either parent was unavailable.

Figure 2



Predictive testing provides virtually a 100% certain answer, as to whether Mary has or has not inherited the gene causing H. It does not, however, say anything about her current health with respect to Huntington disease.

In a small number of families, predictive testing may not be possible. This may occur if the clinical diagnosis in the family is HD, but none of the affected individuals in the family demonstrates the increased number of repeats found in the HD gene. This is most likely because the family has been misdiagnosed with HD but potentially could be because there is another change in this gene causing HD that we do not yet know how to detect. Another reason why testing could be uninformative is that the at risk person may inherit a gene that has a repeat size between 36 and 39. As stated earlier, some of these individuals may still receive results after the laboratory has had the opportunity to do some further testing or to obtain more blood samples.

The decision about whether or not to take the predictive test is a deeply personal one. For some individuals this test will provide much desired information about their future. Others will choose not to undergo testing at this time.

Prenatal Testing

The same kind of prediction described above can be used in prenatal testing (testing of a pregnancy). Details about prenatal testing and further information about predictive testing is provided in a separate Predictive Testing pamphlet available through the Huntington Society of Canada.

People interested in learning more about predictive or prenatal testing may wish to obtain a copy of this pamphlet and/or contact the nearest Genetic Counselling Centre to explore the personal, emotional and technical issues in more detail.

Intermediate Alleles

Until recently scientists believed that Huntington disease did not arise spontaneously in families. However, in recent years we have learned that Huntington disease can arise in a small percentage of families in which there is no obvious family history of this disorder.

In these families in which Huntington disease has arisen spontaneously, we now know that this has arisen on a chromosome which has an intermediate range of repeats, said to be between 36 –

39. These may be referred to as intermediate alleles or “reduced penetrance”. The CAG repeat size has then increased **on transmission from the parent in the intermediate range**, so that the child is in the Huntington disease range.

Individuals with an intermediate number of CAG repeats will likely not develop HD themselves, but they are at risk for expansion of the gene to the HD range when it is passed down to their children. The likelihood that this expansion will occur is significantly increased when it is transmitted from a male. This means that the children and the siblings of an individual with an intermediate number of repeats are at risk for HD.

Further research into the meaning and cause of intermediate alleles and their ramifications for families continues.

To summarize, the discovery in 1993 of the gene that causes HD has not changed our understanding of the transmission and manifestation of HD in families with a clear family history of HD. What we have learned though, is that in a small percentage of families, there appears to be what might be considered a new mutation or an intermediate allele which predisposes future generations to developing HD.

What about the Juvenile (childhood) form of HD?

Although rare, HD does occasionally appear in children. Physical symptoms of the juvenile form may be rigidity and limited muscle movement. Convulsions occur in about half the cases. Research indicates that in cases where children develop the illness, the father is usually the affected parent. The Huntington Society of Canada has published a booklet entitled *Living with Juvenile Huntington Disease* for use by families and professionals.

Section II

Experiences of Family Members

Family members share their thoughts and feelings about dealing with Huntington disease:

The most difficult experiences in coping with HD:

Partners

- the **initial** reaction when my partner was diagnosed and we did not know where to go for help and information; the **secondary** reaction when I realized our marriage relationship would change drastically, and then to have these changes occur, in almost every aspect. As the disease advances it is important to continually reassess the coping role and to arrange daily “time to think” periods.
- the realization that in addition to my partner with HD, our children may develop HD, and a concern for the stability of their marriages, should this happen.
- having to watch a loved one deteriorate slowly and cope with HD.
- my partner’s personality changes, aggressiveness, temper outbursts, swearing, and accusations which appeared as the disease developed, were very hurtful after years of happiness and raising a family together. Before he entered the nursing home, I was afraid to leave him when this behaviour erupted, for fear he would harm himself.
- the long-term loneliness and feelings of hopelessness, as the person advances with HD.

At risk

- dealing with the anger and hostility of the person with HD toward close relatives who wish to offer help and assistance; getting an unwilling person to co-operate.

- when I was younger, to be patient with the family member with HD.
- dealing with caregivers who don't seem to know or care about the needs of persons with HD and the family, and doctors who seem more concerned about making the patient look normal, even if the medication interferes drastically with cognitive functioning.
- not knowing if I will get HD, and feeling if I do, that there is no future for which to strive.
- the brutal truth that if I develop HD, my appearance may be revolting to others, plus a great fear of loneliness and the realization I could be separated from those I love most.
- the feelings of guilt/anger I have toward my parent for having HD.
- being afraid and aware of every physical and mental "sign".
- deciding whether or not to have children.

Individuals with HD

- having to give up my favourite job, teaching.
- totally denying my diagnosis for the first year and trying to deal with the depression and anger related to it.
- coping with misunderstandings about HD and people's assumptions that I am drunk.
- being dependent on others.
- fearing that the disease will mask my personhood and that I'll be viewed by professionals, family and friends as an HD person, not a *person* with HD

Individuals with increased risk after testing

- waiting for the results of the test
- telling my family and friends that I have increased risk
- deciding whom to tell amongst my co-workers and acquaintances
- dealing with people who think I already have symptoms

The most helpful experiences in coping with HD:

Partners

- the support of family members
- finding an “understanding” family physician; the loving concern and emotional support given by our family physician.
- the support of a “small” circle of personal and business friends.
- the concern, support, and empathy of health professionals in various programs e.g. day care, long-term care, public health, social work, ...
- help from the Huntington Society of Canada, e.g. books and pamphlets, meetings, and the mutual support and friendships provided by its members—in particular, a caring people who share in a common goal of helping others.

At risk

- dealing realistically with the information I have; dealing with my at risk status from a healthy viewpoint (panicking is futile), and interacting positively with the Huntington Society of Canada.
- having someone with whom I can talk and share my feelings and anxieties.
- through the existence of the Huntington Society of Canada **I know I am not alone**, and that many people are working on our behalf.
- finding a facility where my relative with HD receives good care.
- learning about all the scientists involved in research to find the cause, improved diagnostic procedures and treatments, and a cure for HD, as well as the health professionals investigating new therapies and ways of coping.

Individuals with HD

- the development of positive thinking and a positive attitude. Norman Vincent Peale’s book, *The Power of Positive Thinking* has been very helpful and has led me to a spiritual awareness. The opportunity to express and work through my anger and denial

about having HD with a counsellor has been a terrific help. Daily exercise through walking is vital to my well-being. I find it helpful to talk with other people with HD and also to have time alone to think and sort things out.

- having an understanding spouse who recognizes the changes that HD brings and is willing to make changes with me in our relationship and lifestyle.
- having grown children who “pitch in” with cooking and groceries and who join in for fun times together like cross country skiing and canoeing.
- having a caregiver team of family and professionals.

Individuals with increased risk after testing

- the support of my friends and family
- learning to live my life “in the moment”. Whenever I start worrying about the future, my getting HD, I intentionally draw my focus back to the present. I find a great amount of peace in this and now use this problem solving skill in all areas of my life.
- getting tested—for me, knowing is far better than not knowing.
- through the Huntington Society of Canada, meeting and interacting with people with similar experiences; they have become my extended HD family.

Section III

Living with Huntington Disease

How do people cope with being at risk?

Studies of people at risk have shown that individuals react to this challenge in a wide variety of ways. Even though there is an equal chance that the person will escape the disease as there is that s/he will develop it, some people allow the odds to master them and live their lives with dread, denial, fear, emotional disarray and gloom. Unnecessarily, though understandably, they make their situation much worse than it might be.

Others 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 neither 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 wished away. A parent who carries the gene but who ignores the disorder or represses discussion of it still has a 50:50 chance of passing it on to children.

Still others manage the risk in a reasonable way and keep it in perspective when making choices.

Many at risk people seem to fantasize 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 symptoms of Huntington. They ignore the years of productive and self-reliant living which are at their disposal even if they do have the disease.

The availability of predictive testing has added an additional pressure to those at risk. The vast majority have chosen not to be tested, one major factor being the current unavailability of satisfactory treatment. Others view testing as an opportunity, even though it is frightening. In any case, the availability of testing requires each individual to at least make a decision one way or the other.

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 in life and a deep understanding of certain aspects 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.

How do at risk persons approach marriage, partnerships and parenthood?

The hereditary nature of HD, combined with the problem of late onset and the fact that testing is a very personal decision, make contemplation of marriage and the prospect of starting a family particularly difficult. When a person who is at risk is contemplating marriage, then it is important that the partner be told the facts. However, many at risk individuals have already established families before they learn about HD, or fully realize the hereditary nature of it. Some who fully understand the disorder will consider all of the options carefully and then make their decision to have children. Others at risk decide not to have children of their own, in order to avoid the possibility of passing the disease on to another generation. Through genetic counselling the full implications of the genetic characteristics of HD can be discussed and all the alternatives available (e.g. adoption, artificial insemination, predictive and prenatal testing, having a child without testing) can be considered. In other words, know all the facts and alternatives before making decisions appropriate to your individual situation.



How do people live with increased risk after testing?

The following is a personal account of one such person.

“Finding out you have the gene for HD is another step in a disappointing journey. This journey usually starts when your parent or family member is diagnosed with HD. You soon realize that you are at a 50/50 risk for HD and it is overwhelming emotionally. If you then pursue predictive testing, you may receive a positive test result, confirming your worst fear that you too have the HD gene. It feels like all you get is bad news, and the other door, the door with the good news, has just slammed shut on you. There you stand, knowing you cannot avoid the demise of HD (unless you die first, of course). Your route is set, and you easily imagine the diagnosis of HD in the future to be the last in a series of disappointing and very unpleasant revelations.

This is the HD movie playing in your life but largely in your mind. Sometimes it wreaks havoc. The burden can feel overwhelming, and the fear can be immobilizing. The future is very frightening. How will you cope when you are actually diagnosed? Will people still want you? The last leg of the journey is a focus in your mind now, and your dreams are filled with scary scenarios. When it happens, you hope you will be able to adjust. You hope you can accept the change in the script, like you have had to do so many times before.

Gradually the initial shock wears off and you are less and less consumed by the movie, but the images are always there, particularly in your mind. Much of the time your life carries on very normally, and there can be long stretches where HD plays only a minor part. You have friends and family, and there are lots of choices to make about how to live your life. You may find a reluctance to “waste” time and everyday experiences are as rich as ever if not more so. When that “other door” closes on you, and there is the certainty you have the

gene, a lot of things become very clear. Expect an immediate reevaluation of all that you do. Fortunately, the emotional effort that goes into being at risk is also dispensed with, because you need to focus on your priorities now. It is liberating to finally get rid of the uncertainty! Ironically, the dreaded news can give new meaning and direction to your life.

*Knowing you will get this disease makes the intervening years more precious. The present is enriched and you are motivated to fill your time to the maximum with wonderful experiences and relationships. As unwelcome as the bad news is, you still know perfectly well there are many people in far worse circumstances. A reality check is helpful. It is also helpful when you remember that biologically, nothing has been changed. You are the same person as before, but **armed** with some information that you can use to your advantage! You may even watch as HD becomes treatable or curable!*

Living with a positive test result for HD may turn out to be the most rewarding stage in your life, in the final viewing. A bit of fear and motivation may force you to take control over the things in your life that you can change. It may help you let go of the things that you cannot change.”

How does one cope with denial?

Denial on the part of the person with HD 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. Denial can be a very difficult behaviour for the family to deal with, particularly if safety issues, such as continuing to drive the car when it is no longer safe, are involved.

In many cases, denial in persons with HD is due to brain dysfunction. For example, when neurons die, feedback loops become disconnected so that information cannot travel from one part of the brain to another. As a consequence, many people with HD suffer from anosognosia (lack of self-awareness) and they don't recognize

their disabilities. That is, they are unable to evaluate their own behaviour. It is like looking down at yourself and not being able to “see” what you are doing, saying, or feeling. There is rarely any benefit in someone with HD accepting his or her illness and all of its consequences. Therefore, we encourage family and staff members to accept the “denial” as a common symptom of HD. In these cases, avoid interpreting “denial” or noncompliance with therapy or nursing care as intentional—it may be an honest response to a confusing situation.

In some cases, the person’s acceptance of the diagnosis can be improved, depending on who does the explaining. The explanation of an understanding partner or relative may be easier for the patient to take than the more clinical analysis of a physician. Families may be helped by recognizing a five-step coping process we all may experience. These steps are not necessarily consecutive and may be concurrent.

- 1. Denial:** The 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.
- 2. Anxiety:** In response to the fears of what could happen, the person suffers headaches, fatigue, insomnia, irritability, and will require support.
- 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 medical personnel. There may be a great deal of angry behaviour. Resentment builds up between family members, particularly between the person and the partner. Counselling can help to channel the guilt and redirect the anger in constructive ways.
- 4. Depression:** This is a critical phase, necessary for eventual readjustment. With acceptance, the person begins trying new behaviour, planning for that other way of life.
- 5. Stability** (a period of acceptance and adjustment to the diagnosis): A person “cycles” back and forth among these phases, but with family and professional support and reinforcement, the person with HD can be helped to *live with HD*.

As mentioned previously, moodiness and irritability can make life very difficult for the children and partner. The first and probably most difficult step is to face the truth—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. It may be very helpful for children to realize that it is the illness, and not them, causing a parent's outbursts. The Huntington Society of Canada has helpful articles on the needs/reactions of children.

Relatives of an at risk individual who is having problems, or of a diagnosed person, may also deny the existence of difficulties and may blame the partner. Denial or secrecy may be chosen because they are not ready to accept the diagnosis or the fear of discrimination in such areas as employment, promotion, adoption, insurance or social acceptance.

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.

Do families living with HD experience severe tension at times?

Some of the symptoms of the disease, particularly denial, moodiness, irritability, personality change, apathy, impaired judgment, or violent outbursts at the slightest provocation, can make life very difficult for the family. The isolation that families experience causes tension. Persons with HD may need help with their legal and social affairs, or may require more care than can be given by the family in the home. Unfortunately, the person is not always in a position to realize when this does occur and tense family relationships result.

From the other perspective, a person with HD reminds us that “forgetting to do things several times each day is stressful, as well as being able to do only one thing at a time, and then much more slowly than in the past. Keep tables and counters cleared, put books and shoes in their proper places, as clutter and mess cause anger. The loss of organization skills leaves the person with HD unable to clean up the mess, resulting in stress and feelings of hopelessness.”

Sharing these stressful situations may be extremely helpful to discover management strategies. Your family doctor may involve the public health nurse, a social worker, psychiatrist, or a community agency. The Huntington Society's of Canada Resource Centre Directors, chapters and area reps are there to help. Other families who have handled these situations are often willing to share how they coped and offer understanding and support, either individually or in support/discussion groups organized by the Huntington Society of Canada.

Do persons with HD and their partners experience role changes?

Most definitely. Jobs and/or tasks normally carried out by a person with HD may become too difficult for them, requiring their partner to step in. This role reversal can be very stressful for both the person with HD and the partner. Over time, the marriage relationship will become drastically altered, and the person with HD will be less of a friend, companion and lover. This adds personal grief to a complex situation, and both the partner and the individual with HD may need professional support to deal with these changes. Another possibility is a couples- group, in which one partner has been recently diagnosed. We are aware of such a group which meets monthly with four couples, where changes and strategies to cope are shared openly. Another suggestion, coming from a relationship where one partner is at risk, is to begin sharing responsibilities/tasks from the time you learn about HD, in case HD should develop later.

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 interacting with someone who has involuntary movements and/or slurred speech, especially if they have not been fully informed about the condition. The person with HD may not like to go out socially or shop because of embarrassment about movements, people who stare, etc.

A person with HD states “our family and friends are wonderful. I’ve told them it’s easier for me if we all move after two hours of visiting. Also, to extend a visit, I sit in a rocking chair and crochet afghans. By keeping my fingers moving (rather than sitting and watching them move) and by rocking, my spouse and I can join in the fun of a small group of friends for an extra two hours. Otherwise I would need to go for a quick-paced two mile walk to relieve the building pressure associated with HD. Also, instead of playing card games that involve memory, e.g. *Rook* or *Hearts*, our friends have introduced us to games like *Skipbo* and *Rummy*, so that I can play too”.

Family members experiencing changes in the behaviour of the person with HD may not realize that this does occur in HD. Feeling that it is unique to their family, they may not wish to share it, or try to handle it on their own and further isolate themselves.

The educational materials available through the Huntington Society of Canada can help family and friends understand what’s happening. Family members are encouraged to seek assistance and support from friends, family, clubs, social groups, church, synagogue, and/or professionals, e.g. social workers, counsellors, therapists, community nurses, clergy. Don’t be afraid to ask for help. The discussion/support groups mentioned previously can also be beneficial.

How does one adjust to changes in sexual activity?

“I have a shrewd suspicion that there are very few people in this world, if they are totally honest, who have a perfect sex life,” states Shirley Dalby, former Director, Huntington Disease Society in the United Kingdom. “And even if they do, would their



partner or partners feel the same? The media, in all their guises, constantly tell us what a good time we all ought to be having, and raise expectations of ourselves and our partners. But, however open society may appear to be on sexual matters, most people feel that their own experiences are completely private; to discuss the intimate details with an outsider is embarrassing and disloyal to your partner.

“Sexual problems with HD need to be seen in the context of what is common, what the sexual relationship was like before onset, and what specific difficulties HD brings. People’s sexual needs and activities are very variable in such aspects as frequency, techniques, numbers of partners and reaching of orgasm. In a mutually satisfying partnership people need to accommodate each other so that some kind of consensus is arrived at. This may change over time. Problems arise when mutual agreement can’t be reached or when changes take place. The changes can be emotional and/or physical. Relationships deteriorate or improve, boredom sets in, somebody is ill, children circumscribe sexual activity to time and place and may even emotionally replace the partner. And some people have a problem in finding a partner at all.”

Common sexual problems are loss of desire, inability to have sex, and conflicting desires and needs. Impotence in men is common in the late stages, but there appear to be no physical reasons for it early on. Medications can contribute to impotence or loss of desire. This may be discussed with the physician and an alternative schedule or medication considered.

A general loss of desire, in men and women, may be due to depression and emotional problems. “It can also be a grief reaction to feelings of loss of self,” says Ms. Dalby, “fear of what they will become, or a low self image as a sexually desirable man or woman. The person may be anticipating rejection by their partner and may withdraw to save their personal pride—it is better to reject than be rejected.

“Some people deliberately withdraw from sex to give their partner permission to find someone else in the hope that they will still go on caring in other ways,” she states. “This is difficult for the partner who wants to be loving and caring, but feels rejected and inadequate.”

Another type of problem is one of unmet or excessive demands. Loss of inhibition can lead to inappropriate and over-activity regarding frequency, place and time. This can be an enormous embarrassment to partners and make them feel constantly harassed.

There may, in fact, be a complete loss of physical and emotional attraction. Physical, cognitive and emotional deterioration can lead to emotional distance. Physical movements, particularly facial ones,

can be disturbing. Partners often do not want sex at all, and most certainly do not want to cope with increased demands. Ms. Dalby confirms that “some women have described the experience as marital rape. Some have talked about the feeling of being attacked, with the result that they have withdrawn more than strictly necessary to prevent further attacks. To some extent the partner’s feelings will depend on the previous sexual relationship. If it were already difficult then it would be likely to become more so, but if it were good, there will also be feelings of loss, sorrow and guilt.”

In the words of a spouse “it is often very hard to respond sexually after a long day of guiding and helping the person with HD (more like a mother-child relationship). To switch to a sexy bed partner just doesn’t seem to fit into the picture at this stage of the game”.

There is likely to be sorrow, guilt and fear on both sides, and not only over the sexual aspects. Discussion may be difficult as it will be feared that being open about feelings may be hurtful and dangerous. Dr. Neal Ranen, a psychiatrist at the Johns Hopkins University School of Medicine, notes that “the spouse should be asked about this when the person with HD is not present. Wives in particular may be quite distressed or fearful of their affected husbands who can become aggressive if their sexual demands are thwarted, but they may be afraid to mention it in the presence of their husbands.” Says Shirley Dalby, “Partners do need to be reassured as to the normality of their feelings and supported in their actions. Outsiders are sometimes quick to criticize things they don’t understand. It is also important to note that nobody should suffer sexual harassment or violence—for their own sake and for their ability to continue caring physically and/or emotionally for the person with HD.”

Is suicide more likely to occur in persons who have Huntington disease?

To respond to this question, we have used excerpts from a presentation to members of the Huntington Society of Canada by Dr. Allen Rubin. Dr. Rubin is a psychiatrist and neurologist at the University of Medicine and Dentistry of New Jersey, Camden, NJ.

Dr. Rubin comments that “Talking about troubles does not make them happen. The same principle holds for suicide. In fact, often talking about suicide can uncover solutions that can be preferred routes of response to what seem to be intolerable situations. Conflicting surveys have suggested, on the one hand, that HD suicides occur primarily in early illness, while another study comes to the opposite conclusion that suicides have occurred primarily in advanced illness. From my own experience I am not certain suicide is more common in persons with HD who have the opportunity to benefit from a response to their need. I conjecture that the higher numbers that were reported in the past reflect numbers of individuals detached from a supportive and potentially therapeutic community or from appropriate recognition and professional care.

“In early illness, individuals might be caught in a trap in which they have the need for control, but yet have a pessimistic vision of their own future. They suffer a loss of their own identity and productivity and an inability to get things done the way that they want to. The intervention is to find one wall of the trap that can be opened. Often this is sufficient to treat the suicidal moment. People generally will move quickly to any solution which frees them from the trap. An individual’s morbid pessimism may arise from an unrealistic vision of their own future. That can be challenged. Optimistic or pleasurable alternatives can be created. Role and identity problems can be examined. An individual might be able to find some valued part of himself that can be recruited and revived. A small intervention that allows a person to feel good about himself can be very potent.

“Although medicine can make a dramatic difference to a suicidal person, there are times when a medicine would be an incomplete treatment.” Dr. Rubin offers an example: “An individual might experience a catastrophic reaction when the future comes crashing in. Combined with impulsivity and depression, this makes for a very dangerous moment. It is especially dangerous if the individual does not have sustaining pleasures or pleasurable resources. The solution may be to find something that individual can grab on to which creates some pleasure again—a self-cuing prop, a set of steps to follow—to protect the vulnerable person from the sense of being totally overwhelmed. If you can divide the suicidal moment into its parts,

defining each wall of the ‘trap’ that is felt, the individual can protect himself from a sense of global chaos. In late illness, people with HD may become exhausted, isolated, and dependent; they may attack their own sense of what’s left of themselves. Here, personal relationships are critically sustaining. We work on each part of this problem, giving more help, more comfort and attention, making new friends. We work on every part of the problem we can detect.”

From quite a different perspective, some people will feel that suicide or euthanasia are reasonable considerations, as long as such a decision is made before judgment is impaired and not in an emotional moment or crisis.

Does a diagnosis of HD mean that a person should immediately change his/her lifestyle?

Definitely not. The person with the disease should be encouraged to carry on normally for as long as possible. If s/he is having difficulties with work, it is often better that the person gets 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 partner with HD to also carry on normally for as long as possible, although it may be necessary for another member of the family or homecare staff to assist with some of the chores.

One person living with HD, who loved to cook and bake at the same time, was burning most things because he forgot to stir them. This happened until he realized “I can focus on only one thing at a time. Now I cook only one casserole or one soup at one time, instead of the 5 or 6 recipes I used to have on the go simultaneously. We also plan to go out for a meal each week as I find it stressful organizing now.”

Who to talk to

Sometimes a family with Huntington disease will work together with a team of health professionals to receive the best care. A

neurologist, a psychologist or 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 within their own circle of friends aided by only technical professional help.

Health professionals vary also in their preferences to work individually or with a team. 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 should 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 local chapters of the Huntington Society of Canada 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.

Counsellors

Unfortunately, some health professionals, particularly those who may deal only rarely with HD, sometimes communicate little more than a sense of hopelessness and futility because of the irreversible nature of the disease. The diagnosis is made and the person with HD and family are frequently left to cope with the future as best they can. They may leave the office in a state of shock or depression with no provision for a follow-up to assist in adjusting to the new reality or to help them cope with the personal, medical, financial and legal problems which may arise.

The Huntington Society of Canada has opened HD Resource Centres with trained staff to assist family members and offer consultation and information to professionals. The Society also employs several support (social) workers to assist families on a short-term intervention basis. Community and health professionals are also becoming much more familiar with the special needs of persons with HD and their families.

The availability of counselling is essential for 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 toward helping persons with HD and their families retain maximum productivity and acceptance while coping with what is admittedly a difficult and challenging situation.

A diagnosis of Huntington disease invariably deeply affects the person and family, as well as a wide circle of friends, work associates, neighbours, and others in the community. As with any serious diagnosis, the person with HD and the 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 a diagnosis has finally been made, thoughts of "Why me?"—all of these feelings and many others are **common** and **normal** reactions.

Feelings may come and go, change and return, and may last many years, as noted under the discussion of denial. 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. Some counsellors are specially trained to help people deal with depression and grief and can be of great help to families dealing with Huntington disease. As a wife of a person with Huntington disease said, "The counsellor made it safe to cry". Families may also find it useful to consult marriage counsellors to work out new adjustments and roles for marriage partners. Often the initial symptoms of the illness have placed a strain that neither partner could understand until after the diagnosis was made.

Genetic Counselling

Because HD is hereditary, a genetic counsellor may be very helpful. Genetic counselling has developed in response to the increasing number of diseases recognized as hereditary. Some genetic counsellors, because of training and experience, will not only inform families about the disease and its pattern of inheritance, but also explain 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 the disease can have. The counsellor may assist with the difficult problems involved in telling others (partner, siblings, and children) about the disease. Genetic counsellors and social workers often work together to ensure that persons with HD and their families receive all the services and benefits to which they are entitled. Genetic counsellors will also make referrals to other professionals for more in-depth counselling or support.

Therapeutic Interventions

This is a fairly new concept for the person with HD. For years, because HD has been categorized as a progressive degenerative disease, it was believed that therapy would not be of much assistance. The quality of life of the person with HD and family may be greatly enhanced by the following therapies. If these are not available in your area, hopefully you can travel to a centre where they are, and learn strategies which can be practised and used in the home environment.

Communication & Swallowing Therapy

Speech and language therapy can help the individual maintain communication skills for as long as possible. A qualified speech/language pathologist will be helpful in assessing the individual's speech (the ability to talk and be understood) and language (the ability to use language to express thought and ideas) skills. Assessment/therapy will be helpful in providing strategies so that the individual will be able to speak as clearly as possible and to verbally express his/her thoughts and ideas. It is important that the individual with HD clearly explain to family and caregivers his/her care needs and wishes before speaking becomes too difficult. When speaking does become too difficult, prior knowledge of the individual's lifestyle, personality and care preferences will enhance his/her quality of care and greatly facilitate communication between the person and those providing care.

Difficulty swallowing and an increased risk of choking are also part of the disease process. A qualified speech language pathologist

will assess swallowing difficulties and suggest management strategies. Change in position during eating, varied food textures, and adaptive equipment have all been beneficial in maintaining independent eating for as long as possible. You can contact a speech/language pathologist through your family doctor, Homecare Program, local hospital or your insurance company.

In many locations you may require a referral from your family physician to a speech/language pathologist.

The Huntington Society of Canada has produced booklets and videos which outline management strategies for communication and swallowing difficulties. Please see www.huntingtonsociety.ca or contact the Huntington Society of Canada to learn how to obtain copies.

Physical Therapy

The physiotherapy goals and interventions for people with HD may vary as the disease progresses. In the early stages the goal is to improve or maintain the client's functional level within the limits imposed by the disease process and to promote safe mobility for as long as possible. In the later stages the goal is to prevent or delay the complications of immobility/bed rest such as joint contractures and muscle weakness.

The physiotherapist will assess areas such as balance, coordination, strength, flexibility, and the ability to transfer, walk and participate in one's self-care. Exercises and compensatory strategies may be recommended. The physiotherapist will assess the need for walkers, canes, etc. to maintain safe and independent walking and train the client in their use. The physiotherapist in cooperation with the occupational therapist will also determine if there is a need for specialized seating to achieve a secure upright sitting posture and prescribe the necessary equipment. Seating becomes increasingly important in the later stages of the disease as more and more of the day is spent in a sitting position.



Physiotherapists may be located through your Homecare Program, your family doctor, Homecare Program, local hospital or your insurance company. There also may be outpatient physiotherapy clinics in your area. You may require a referral from your family physician.

Occupational Therapy

Occupational therapy primarily deals with independence in activities of daily living. Assistance from an occupational therapist may help to maintain or retrain such basic activities as cooking, dressing, eating, bathing, grooming, (smoking), shopping and the use of public transportation which are all geared to keeping the patient independent as long as possible. The occupational therapist may be able to make other suggestions for daily living such as using adapted equipment or making environmental modifications to the home. For example the use of grab bars in the bathroom may help to prevent falls. You can contact an occupational therapist through your family doctor, Homecare Program, local hospital or your insurance company.

Nutritional Therapy

Good nutrition helps to maintain a healthy physical state and to prevent secondary illness. Additional calories, possibly up to 6000/day for men and 4000/day for women, are required to prevent weight loss as the disease progresses. Plan food intake carefully, after assessing the individual's needs (be sure to include necessary foods from the four food groups essential to good nutrition). Your hospital nutrition services, your family doctor, Homecare Program, or local hospital can likely supply a copy of Canada's Food Guide to Healthy Eating. Qualified nutritionists and dietitians may be available in through your family doctor, Homecare Programs, or through a private clinic for counselling. Ask your physician for a referral.

Recreation (Therapy) Activities

Many persons with HD are able to participate in activities outside the home, although they may be hesitant to get involved. It may take a good bit of encouragement. Therapeutic work and recreation centre's

give individuals the opportunity to pursue hobbies and interests and to meet new people. Participation lessens the person's dependence on the family and provides the family with temporary respite from care giving. The recreation departments of some cities and municipal areas have special recreational and social programs for individuals with disabilities, and some hospitals run recreational programs and/or offer day hospital programs. Your family doctor, occupational therapist, your nearest Community Information Centre and your local Huntington Society of Canada may know of programs and activities. Local transportation services for persons with disabilities often provide transportation to these activities at a reduced fare.

Horticulture therapy, offered at some hospitals and rehabilitation centre's involves caring for plants. It has been beneficial to some persons with HD as they can, with minimal care and effort, see growth and response to their efforts.

Music Therapy

Music therapy can be defined as the clinical use of music as a therapeutic intervention for persons who have special needs, using music as a tool, music therapist's work with individuals to achieve specific objectives. The goal is not to cure or prolong a client's life in the medical sense, but rather to develop an individual's potential in a specific area and improve their quality of life.

Grieving, losses and meaning in life are some of the issues dealt with in music therapy at the HD day activity program in Vancouver.

In quite a different setting, staff of long-term care facilities use well selected music, reflecting the individual's personal preferences, to enrich the life of the person with HD, and to help relax and calm the individual during stressful times.

Other Therapies

Various other therapies are being explored such as Art Therapy, Pet Therapy, Relaxation Therapy, and various discussion groups to reflect the interests of various individuals (Music Appreciation, Bible Study, etc.)

Safety

Driving

A family member has suggested that whenever possible, it is wise for the person with HD and his/her partner or caregiver to work out a cooperative method for dealing with safety and medical issues

As soon as indications of deteriorating driving ability appear, the person with HD must stop driving. If the person denies that a driving problem exists and absolutely refuses to stop, your family physician should be consulted. Local law may require the physician to report a person with HD to the appropriate government authority. Cooperation with an objective driving assessment may provide important information to see if a change in driving status is warranted. If the person refuses to take a test and continues to drive, the appropriate government authority or local police may have to be contacted to request that the individual have a driving test. This can prove very awkward for the family. One possibility is to investigate if this can be done anonymously in your area, if other approaches fail.

Smoking

Because of the fire hazards involved, smoking often becomes a serious concern for the person's family. There are a number of options available from smoke cessation patches as well as a Smoker's Robot may also prove useful. Consult your family doctor, or other professional to recommend an appropriate device or program.

Medic-Alert Service



A Medic-Alert bracelet or necklet may be worn to indicate that the individual has a medical problem. This may be important for individuals with involuntary movements which are sometimes seen by the police as drunkenness. For further information contact the Huntington Society of Canada, www.huntingtonsociety.ca

A person may also obtain a wallet-size Medical Identification card from the Huntington Society of Canada. The card includes the person's name and address, whom to contact in an emergency, and a

statement about the symptoms of HD. This card may be especially desirable, if involuntary movements cause skin injury with a bracelet or necklet.

Planning

All of us need to consider long-range financial and legal planning; we need to know what our options are. This is particularly true for someone who is diagnosed. It is also important for anyone who has received increased risk after testing. It is wise to discuss long term legal arrangements with a lawyer. Legal Aid services may offer legal advice to qualifying applicants. Topics may include giving a power of attorney, perhaps changing one or both wills depending on their provisions, changing ownership of property such as the house and cottage, making bank accounts and safety deposit boxes available to the other partner and so on.

This can be very upsetting to the person with HD and must be handled carefully. One person with HD, a successful accountant, mentioned that he had given up his job and his driver's license. Then came the question of giving a power of attorney, which was most disturbing, as it represented the last bit of control he had, and he was being asked by his wife to do this. She was anxious to make arrangements while her husband could still take part in the planning. After the husband and wife discussed this with an objective third party, these arrangements were made with the family lawyer.

As provincial laws vary, check with your lawyer to make appropriate arrangements for your family. As useful background information, please see Appendix B, Facing the Future: Legal Issues of Living with a Chronic Illness.

Care of the Caregiver

The primary caregiver, whether it be the partner, or someone else, has an important duty to take care of him/herself—physically and mentally, for one's own sake and that of the person with HD. Be sure to arrange times for recreation and socialization and talks with friends and relatives. Don't be trapped in your home 24 hours a day. **You must take care of yourself; this is not being selfish.** You are encouraged to contact the local authority which provides home support

services to see what services are available to assist you with personal care of your family member, homemaker duties, etc. Respite care in your home or in local care facility can also provide a much needed break for you.

At a time when it is necessary to choose a care facility outside the home, feel free to seek objective advice and assistance from professionals. As there may be long waiting lists, don't wait until the situation is critical to begin making enquiries. Many communities now have placement coordination services to assist families. Local contacts of the Huntington Society of Canada can also be helpful. This is a most difficult decision to make. The partner or family member responsible must learn to live without guilt and realize that placement is in everyone's best interest.

You are reminded to set realistic goals—to undertake only as much as you can accomplish and give precedence to things that are really important. Many HD families have learned that it is best to take one day at a time. Check the Action Plan for caregivers included as Appendix A.

Is there anything I can do now?

- If you are part of a family at risk for HD, provide other family members with information about the disease and encourage them to join and support the HSC. Refer them to the Caregiver's Guide to Understanding Huntington Disease. Be sure your doctor is also well informed and has a copy of *A Physician's Guide to the Management of Huntington Disease*. The Society will also be happy to send you further information, including its list of Information & Education Materials Available.
- Be open within the family about the effects of HD and the hereditary risks. Encourage families living with HD to tell their children about the disease. Tragedies are many when children are not informed. It is no kindness to "protect" children from this knowledge. Local contact persons of the Society will assist you to locate where genetic counselling is available.
- Consider appropriate future planning (for example, legal, financial, health, life and disability insurance, tax breaks, etc.).
In Section III, Planning

- Encourage family members with the disease or at risk to consider donation of brain tissue to the Canadian Brain Tissue Bank at time of death. This will greatly assist new research efforts. It is critical to make arrangements in advance. To obtain additional information and appropriate consent forms, contact the HSC or one of its local contacts.
- Be willing to participate in research when the opportunity is presented. This will help further the understanding of HD and the search for effective treatment. Opportunities are announced in *In The Know*, the e-bulletin available bi-monthly, and through *Horizon*, the newsletter of the Huntington Society of Canada.
- Support the Huntington Society of Canada by volunteering at the local and/or national level. The Society needs volunteers with a variety of interests, skills and time available.
- If you are able, support the Huntington Society of Canada with a donation. Your contribution will be used to support the programs we offer to HD families across Canada. Many people who find it difficult to make a donation now are choosing to support the Society through their wills and/or life insurance policies. After providing for family and friends, making a gift to the Society by will or life insurance provides a meaningful and lasting contribution to the fight against Huntington disease. Planned gifts help ensure the long-term funding needed to advance our ongoing support of research, family services and education programs. The Society's National Office can provide more information on how to make a planned gift.

In the words of Dr. Nancy Wexler, American scientist, HD advocate and HD family member, speaking at an HSC annual conference: "Individual people can make an impact, and when you're in a group like this all across the country you make a bigger impact, and when you're international you make an even bigger impact. There's absolutely no question that the Huntington movement we're all part of is considered one of the very most successful throughout the entire world. And what are we? Just individuals getting together and saying 'this disease cannot get me down; I'm going to conquer this thing'. And we're going to do it soon. We're going to do it in the lifetime of ourselves and our children."

Is there reason for hope?

Most definitely! In addition to the work of the Huntington Society of Canada in Research, Education, and Individual/Family Services, similar organizations in many other countries are pushing forward in these same areas. These efforts complement and encourage the work of professionals and scientists in many countries, including the World Federation of Neurology Research Group in HD, an international network of scientists dedicated to increased knowledge, understanding and treatment of Huntington disease. A major breakthrough occurred in 1993 with the discovery of the gene associated with Huntington disease. In 1993 the Huntington Study Group was formed, a consortium of scientific investigators from academic and research centre's in several countries, whose goal is to find improved treatment through a planned series of drug trials and basic science research. Also important is the increasing information available to doctors, nurses and other professionals, which improves the level of care offered to persons with HD and their families.

While much remains to be done, it is important to note that more has been learned and accomplished in the past two decades than in the time since Dr. George Huntington first described the condition in 1872. There is light at the end of the tunnel.

Please join us and work with us. You can make a difference.

For Further Information

The Huntington Society of Canada has a wide range of publications and videos available on Huntington disease and many aspects of care management. To order copies or to obtain a complete list of Information & Education Materials Available, please contact:

www.huntingtonsociety.ca

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Appendix A

Action Plan for Caregivers

by Dorothy Orr, R.S.W.

Caregiver Support Counsellor for Caregivers of people with dementia. Adapted for Caregivers coping with any disease.

1. Get help early—*counselling*, assistance with care giving duties, etc.
2. Involve your family from the beginning by sharing your concerns with them.
3. Access all the information you can about the disease and educate yourself as much as possible about its progression.
4. Have an awareness of the losses to come, such as incontinence, inability to dress, etc., so they are not totally unexpected.
5. Recognize the hidden grief component of your anger, anxiety, guilt and depression. Expect adaptation, but not resolution, of your grief.
6. Appreciate your grief and seek out someone who understands it.
7. Recognize the signs of denial: for example, you insist, “I don’t need any help.” “Nothing’s wrong. Everything’s okay.” “The doctor has made a mistake—she doesn’t have (the disease.)” “She’s fine today, so she’s getting better.” “No, we don’t need power of attorney.” “Placement in a nursing home is not an option; I’m keeping her at home.”
8. Acknowledge your right to feel emotionally off-balance.
9. Learn to “let go” from the start and share your care giving burden with others. Your loved one **can** survive a few hours without you.
10. Forgive yourself for not being perfect.
11. Stop trying to be perfect: caring for someone with a chronic illness means your world has been turned upside down and you will probably have to compromise some of your personal standards of housekeeping, etc.

12. Join a support group early.
13. Take care of yourself—physically and emotionally. Have regular check-ups. Get as much rest and respite as possible. Eat well-balanced meals. Give yourself time to cry. Don't be afraid to acknowledge your feelings of anger, anxiety, helplessness, guilt and despair.
14. Hang on to your sense of Self. Keep up your regular activities as much as possible to help preserve your identity.
15. Take one day at a time, but don't neglect to plan for the future. Good planning can include getting a power of attorney, accessing community care early and filling out placement papers.
16. Be kind to yourself. Remember you are experiencing normal reactions to abnormal circumstances.
17. Learn how to communicate differently with your loved one if cognitive and language abilities decline. Good communication strategies help to avoid frustration.
18. Make sure your family doctor is someone who is willing to listen and understand.
19. Accept yourself for being human; even if you "lose it" sometimes, give yourself a pat on the back for doing the best you can.
20. Follow the action plan to help avoid caregiver burnout.

Appendix B

Facing the Future: Legal Issues of Living with A Chronic Illness

Our thanks to Gina Rohs, R.N., Movement Disorders Clinic, University of Calgary Medical Clinic, her colleague Susan Calne RN, Neurodegenerative Disorders Centre, University of British Columbia and The Parkinson Foundation of Canada for permission to reprint this material. While very definitely based on legislation in Alberta, it provides background information that will be useful as you plan for the future. It includes information about a living will, also known as an Advance Healthcare Directive. In Ontario it is called a Power of Attorney for Personal Care. Several provinces in Canada have or are currently looking at legislation in this regard.

Legal issues for consideration by individuals with Parkinson's disease or related disorders and their families.

This information is intended to help individuals with Parkinson's disease or related disorders, and their families, become familiar with some legal issues which may arise at some point during the course of the disease. This information will be of interest to the general public as well as the affected family.

When an illness strikes, the attention of the individual and caregivers turns to learning to cope with the presenting problems of the disease. Future care needs and financial protection are not often planned for in advance. While it may not be necessary to have these plans for the future completed in the early stages of the illness it is important to begin to form an awareness of legal options.

The degenerative nature of Parkinson's disease and related disorders is somewhat predictive of a slowly progressive decline in physical and sometimes mental abilities. As the disease progresses caregivers may find they have concerns about the individual's safety and security as well as his ability to make appropriate decisions related to finances and healthcare issues. The caregivers need to know how the legal system can be of assistance.

The knowledge of some of the legal issues and options available can assist individuals and families to discuss these important subjects and to make decisions well in advance, preventing a crisis situation in the future.

Definition of Legal Terms

The terms explained below are examples of legal options currently available in many areas. Please remember that this information cannot replace individual legal advice, nor can it describe specifics as each province or territory will have different rules. You are encouraged to discuss these matters with the legal counsel in your province.

Power of Attorney

Power of Attorney allows an individual to appoint another person to act on his instruction regarding financial matters and/or personal care, *providing he is mentally competent to make that decision.*

Enduring Power of Attorney

Enduring Power of Attorney enables a person to appoint another person who will be empowered to act on his behalf on financial matters *if or when* he becomes incapacitated and unable to make this decision. *The person must understand the nature and the effect of the document at the time of signing it.*

Enduring Power of Attorney:

1. allows the person to choose who will be his attorney for financial matters.
2. can be legislated to take effect immediately and continuing in the event that the person becomes incapacitated or can take effect only if he becomes incapacitated.
3. expects the attorney to keep accurate accounts of all transactions.
4. eliminates the expense of obtaining then renewing an order for Trusteeship, under the Dependent Adults Act after the person is incapacitated.
5. does *not* allow the attorney to make decisions regarding personal care needs when the person becomes incapacitated.

6. can be cancelled at any time provided the person is mentally capable of understanding what he is doing.

The Dependent Adults Act

The Dependents Act, in Alberta, provides protection of the person who is over 18 years of age who is unable to manage his estate or financial affairs or unable to physically care for himself. Trusteeship and Guardianship orders are the two type of protection under this act (Please ask legal counsel in your province about similar legislation)

Trustee Order

The Trusteeship Order is a legal order under the Dependent Adult Act which appoints a Trustee to manage financial affairs of a person who is over 18 years old and who is mentally incapacitated.

Trusteeship

Trusteeship is initiated by the caregiver, interested family members, or in some cases by the Office of the Public Trustee.

1. An application is made to the court to appoint a suitable Trustee who will act in the best interests of the dependent adult, in estate or financial matters. The immediate caregiver or a close family member is always considered to be the most appropriate candidate to act as Trustee.
2. The appointed Trustee must adhere to the accounting requirements.
3. The Trustee Order must be renewed periodically.
4. Trusteeship is more costly than Enduring Power of Attorney.
5. Joint bank accounts and ownership could eliminate the cost of Trusteeship for those persons who might become mentally incapacitated later in life.
6. A person may need only Trusteeship and not Guardianship or visa versa.
7. The responsibilities of the Trustee cease at the moment of the individual's death.

Guardianship Order

A Guardianship Order is a legal order under the Dependent Adults Act which appoints a legal Guardian to make decisions for a person over the age of 18 when he is unable to physically care for himself.

Guardianship

1. Guardianship is usually initiated by the caregiver, interested family members or, in some cases, by the Office of the Public Guardian, often at the request of the administrator of the care facility.
2. An application is made to the court to appoint a suitable guardian who will act in the best interests of the dependent adult in matters related to decisions about health care and where or with whom he will live.
3. The immediate caregiver or a close family member is always considered to be the most appropriate candidate to act as guardian.
4. The guardianship order can be quite wide ranging depending on the abilities of the dependent adult.
5. The guardianship order must be renewed periodically and is expensive.
6. The responsibilities of the guardian cease at the moment of death.

Advance Healthcare Directives

(The Living Will)

Legislation is being developed to give legality to decisions made in advance concerning the individual's future healthcare. This legislation is the Advance Healthcare Directive, more commonly referred to as the Living Will. Speak to the legal counsel in your province regarding this legislation.

Advance Healthcare Directive

- The Advance Healthcare Directive will enable the individual to make some decisions about his future healthcare.
- The individual may be allowed to choose, in advance the person who will be legally responsible to make healthcare decisions on his behalf when he is unable to.
- The individual may also be allowed to state who he does not want to act as his healthcare agent.
- Instructions regarding the types of medical treatment the individual would or would not want in certain circumstances could be made clear.
- The individual would maintain his autonomy over healthcare matters related to himself even in the event that he might become mentally unable to make these decisions at some future time.

At time of writing there is a need for clarity in this area. If a Healthcare Directive (Living Will) has been made there is no guarantee that it will be honoured. It seems to depend on the willingness of the healthcare personnel involved to respect the individual's wishes.



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