

# Juvenile Huntington Disease

A Resource for Families,  
Health Professionals and  
Caregivers

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### Huntington Disease

Huntington disease (HD) is a hereditary brain disorder with devastating effects on both mind and body.

- One in every 1,000 Canadians is touched by HD—for example, as an affected individual, person at risk, friend, family member or caregiver.
- One in every 10,000 Canadians has Huntington disease.
- Every child of a person with HD has a 50% risk of inheriting the disease.

The symptoms of HD — including uncontrollable jerking movements and profound cognitive and emotional impairment — usually begin in the late 30s, although HD can occur in children as young as five, or in adults in their 70s.

Over its lengthy course, HD leads to incapacitation and death.

HD is a disease of families. No member is left untouched. Children, siblings, spouses and parents all face the difficult personal and financial decisions of caring for their loved ones.

There is still no cure for Huntington's, and there are no treatments which can prevent HD or slow it down. However, the accelerating pace of research is offering more hope than ever before that a solution will be discovered one day soon.

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### Huntington Society of Canada

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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# About Juvenile Huntington Disease

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## I. Overview

When Dr. George Huntington first identified Huntington disease in 1872, he stated that the disease “manifests itself only in adult life”. It is now known that Huntington disease can occur in individuals younger than five years of age. When HD appears in someone under the age of 20 years, the illness is recognized as “juvenile Huntington disease”.

Juvenile HD is characterized by a movement disorder which differs from that of affected adults. The earlier the disease onset, the more likely it is that the child will be very rigid, and the less likely it is that he or she will have the chorea (involuntary movements) seen in most adults. In 25-30% of cases, there is a tendency to epileptic seizures — something almost never seen in adults with HD. Juvenile HD also involves changes in behaviour and mental function.

Like its adult counterpart, juvenile HD remains incurable, and there are no treatments which can stop or slow the course of the disease. However, dramatic advances in research have given rise to tremendous optimism that new forms of therapy will soon be within sight.

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## II. The Genetic Basis of Juvenile HD

Huntington disease is caused by a genetic mutation called a “CAG repeat expansion” — this type of mutation was unknown until the 1990s.

The letters CAG refer to three of the four nucleotides which make up DNA — A (adenine), T (thymine), G (guanine), C (cytosine) — and HD is one of a group of neurological diseases caused by this type of gene mutation.

In HD, there is an expanded CAG repeat in a gene on chromosome 4. The function of this gene remains unknown. People who do not have HD have approximately 10-35 CAG repeats in this particular gene, while persons with HD usually have more than 40 repeats. A very small number of individuals with 36-39 repeats may or may not develop symptoms, or may live well into their 60s or 70s without developing symptoms of the disease.

Juvenile HD involves a larger number of repeats — 50, 60 or more — than is found in the adult-onset form of the disease. Generally, the more repeats, the earlier the disease tends to start, but it is not possible to predict the exact age of onset for any individual on the basis of the number of CAG repeats.

“Though the disease is unrelenting, progressive and debilitating, it is the laughter and happiness of the children and the courage of the families that I most recall.”

— Sally



CAG repeat length can change as a parent passes the abnormal HD gene to a child. Thus, a parent whose HD gene has 45 repeats can have a child with 44 or 48 or 52 repeats. The number of repeats tends to get bigger, although occasionally it can get smaller. Very large increases in CAG repeat length almost always come from an affected father, and juvenile HD is more likely to be seen in the children of HD fathers than HD mothers.

There may be other mutations in the HD gene which do not involve the CAG repeat, and there may be “modifier” genes which interact with the HD gene to influence the onset and course of the disease. Research is expected to shed more light on these and other issues in the years ahead.

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### III. First Symptoms

Early signs of juvenile Huntington disease include:

- Rigidity
- Slowness and stiffness
- Awkwardness in walking
- Diminished coordination and frequent falls
- Difficulty in speaking
- Choking and drooling
- Behavioural/personality changes
- Slowness in responding
- Variable/poor school performance
- Difficulty in learning new information

Chorea — the involuntary, dance-like movements which typify HD in adults — is typically mild and, in some children, is totally absent. Chorea tends to be more prominent in older children (those with onset at 15-18 years of age).

Behavioural and personality changes may be evident in juvenile HD — for example, parents may encounter extreme anger, screaming, and impulsiveness. In addition, affected children may be slow to respond, variable in their school performance, and have difficulty learning new information. Often parents and teachers remain unsure whether or not the child’s mental competence is affected or whether he/she dislikes school for social reasons.

Disease onset before the age of six years is extremely rare (probably less than 1% of cases), and almost all children with HD walk and talk at a normal age. Symptoms often appear so gradually that it is difficult to differentiate them from normal “growing pains”. Families, schools and physicians sometimes fail to recognize the progression of symptoms — a critical part of the diagnosis of juvenile HD.

“My son Keith was a happy-go-lucky, mischievous little boy who was very good in sports during his first few years at school. He won lots of ribbons on sports day. Then, when he entered Grade 3, he started having behavioural problems. He ran away from school a lot — we still don’t know why. He was about eight years old. I began to notice he wasn’t very coordinated at times, but I just thought it was a clumsy age he was going through.”

— Laura



“I took Keith to the doctor several times but was told that it was all in my imagination. Finally, I asked for an appointment with a neurologist because, by then, I knew there was something wrong with him. Thank God my mother was with me when we learned the news. I don’t know what I would have done without her support. Deep down I had known all along that he had the disease but to hear those words...it was hard to take. Keith was 13 years old when he was diagnosed. He took it really well, though. It seemed as though he knew all along.”

— Laura

How does one know what to look for? Rebecca, a woman whose husband and two children developed HD, says: “The way in which each person is affected by the disease process is different. But one thing they all had in common was that the initial symptoms represented a noticeable change. The children began exhibiting some coordination difficulty where they had been very physically agile before. They each began having a noticeably more difficult time learning and remembering new information. Personality changes, too. One became more taciturn, the other acted out and emerged as a very stubborn child who would persevere on the smallest detail. All of them began to withdraw as speech and coordination problems worsened. Each continued to be able to learn, but appeared to be learning at a slower pace than in the past. In each instance, the clue was that there was noticeable change.”

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#### IV. Diagnosis

Diagnosing juvenile Huntington disease can be extremely difficult. Juvenile HD is extremely rare, and few physicians will have encountered the disease before. This can lead to a great deal of time being spent eliminating other possibilities.

Compounding this problem, of course, the examination of children can itself be challenging, and if the child is frightened and will not walk or talk for the doctor, signs of early impairment may not be evident. The best physician may have to see the child several times before being confident that neurological symptoms are apparent.

A neurologist can usually determine that the child has a disorder affecting a portion of the brain called the basal ganglia and may suspect that it is progressive, but it can be difficult to distinguish HD from other diseases. Other hereditary diseases such as neuroacanthocytosis, dentatorubralpallidoluysian atrophy (DRPLA), the hereditary ataxias, Wilson’s disease, familial basal ganglia calcification, and Pelizaeus-Merzbacher disease can produce symptoms that resemble HD.

A complete and accurate family history is invaluable in evaluating a child with symptoms suggestive of Huntington disease, and can make the process of diagnosis much more straightforward. However, there are situations in which parents may not even be aware that HD is in the family; in other cases, an adopted child may be involved.

A number of tests may be used in conjunction with the presence or absence of a family history, and clinical presentation, to help clarify the diagnosis. Blood and urine tests may help to exclude other possible diagnoses—such as the chorea-acanthocytosis syndrome, Sydenham’s chorea, systemic lupus erythematosus, or other conditions — though they cannot strengthen the diagnosis of HD.



“My husband Peter and I were idealistic and eager to start a family. I knew that Peter’s mother was in a nursing home even though she was only in her 40s. It didn’t occur to me that she might have a hereditary disease that had been passed on to Peter and would be passed on to our daughter. Peter was 20 and, as far as I could see, completely healthy.”

— Brenda

Brain imaging studies (CAT, or computerized axial tomography; and MRI, or magnetic resonance imaging) may also help to eliminate other possibilities but are not yet very useful for diagnosing HD because their sensitivity lags behind the clinical onset of disease.

Ultimately, a genetic test may be used to confirm the diagnosis. Genetic testing of children is not undertaken lightly and is only pursued when there is clearly a progressive neurological disease strongly suggestive of Huntington disease. Genetic testing, of course, can only document whether the HD gene is normal or abnormal. It cannot tell whether a child’s symptoms are due to the onset of Huntington disease or not.

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## Living with Juvenile HD

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### I. Schooling

The choice of schools for children with HD is of critical importance, given that juvenile HD is so rare and can be so demanding. There are several possibilities: special classes in regular schools, classes for individuals with intellectual disabilities, schools for individuals with orthopaedic disabilities, and others. The most desirable situation is for the child to attend a school with an attached support unit offering physical, occupational and speech therapy.

Parents should visit the classes which are suggested by school counsellors and try to determine which best fits their child’s needs. It is unlikely that there will be other children with HD present, but a class of children with obvious motor disabilities, who are not severely intellectually impaired and can do some academic work, will often come closest to the needs of the child with HD.

“Ms. Knight was very helpful. On the first day of school, she introduced Jodie to the class, and explained to the other children that Jodie was a little different than they were. She didn’t walk quite as well or print her letters quite as neatly. The children responded very favourably and many became good friends with Jodie, helping her and playing with her at recess.”

— Brenda

The choices change as the needs of the child change. Rebecca started her children in traditional classes but moved them “into the aphasia (speech and language) programme for as long as that most closely suited their needs, and then on to a programme for the multi-disabilities. My daughter remained in the aphasia programme as long as possible — she seemed to need to ‘model’ after the more high-functioning students. My son moved on to the multi-disabilities programme much earlier on, so that he would have the experience of being one of the most capable one again, for a change. It’s best to be adaptable and use whichever setting most closely suits their needs at the time.”

An understanding teacher can create a tolerant atmosphere for a child with HD, so it is essential for the teacher to be made aware of the symptoms and problems of Huntington disease. Brenda remembers the positive impact Jodie’s teacher had.

Home schooling is generally not recommended. Not only is the child deprived of social contact, but constant and undue pressure is placed on the parent because the child is home 24 hours a day. Parents are emotionally attached to their affected children and are often unable to push them to do more. Children often respond very differently to teachers and therapists than to their parents, and the presence of other children can often motivate a child in a way that an adult alone cannot. Of course, there will come a time for most children when school attendance is increasingly difficult, and there are circumstances when 24-hour-a-day home care is best for that child. But, for as long as possible, school and peer contact help children even when they are too impaired for most academic work.

When full-time school attendance becomes too much for the affected child, consider reducing the amount of time spent at school rather than eliminating it altogether. Arrangements can be made to have the child attend each day for a shorter period of time. Later, alternating days spent at school and at home might be more manageable.

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## II. Physical and Occupational Therapy

Most physical and occupational therapists have never seen a case of juvenile HD and they may erroneously assume that the affected child will relentlessly decline and that, therefore, therapy is a waste of time. Nothing could be further from the truth.

The child with HD has obvious motor challenges but may learn new skills which improve the quality of his/her life. Although physical and occupational therapy are particularly helpful in the earlier stages of the illness, affected children continue to learn new skills for years after the diagnosis is made.



A physiotherapy programme for children with HD centres on maintaining the range of motion in the joints and independent mobility. Although most children with HD have stiffness, physical therapy and activity can help to prevent muscle contractures. Therapists often need reassurance from the physician that exercise is not dangerous — for instance, in terms of the risk of a seizure or an increase in chorea — and that therapy cannot be postponed until all seizures have been stopped, because that day may never come.

**Jodie’s parents found a creative way for her to get around on her own. She lost her balance and fell quite often. Twice she gashed her head and had to get stitches. “After the second time,” said her mother, “we bought her a hockey helmet to protect her head, as well as knee-pads because her knees were always bruised. It worked well and classmates became quite protective of her.”**

Some very hypertonic (stiff) children benefit from therapy in a pool. In the water, their movements are more free and they can do more. Pool therapy is not recommended if frequent daytime seizures are occurring.

It may be helpful for school and other programmes for the parent to have a note from the physician stating that physical therapy is recommended. One parent obtained, and kept, programming and funding for physical and occupational therapy for her son by emphasizing that the primary benefit of these therapies for children with HD “is prevention of further costly deterioration and complication, not simply maintenance. Medical treatment and funding should be geared to making a difference in quality of life.”

In the later stages of the disease, it is often necessary for a child to use a special reclining wheelchair with a custom fabricated postural support.

## Strategies for the Person with HD and Listener

- ï Use a reduced rate of speech.
- ï Repeat the message.
- ï Rephrase the message.
- ï Simplify the message.
- ï Spell part of the message.
- ï Identify key letters in the message.

From *Managing the Communication and Speech Difficulties Associated with Huntington Disease*,<sup>1</sup> by Estelle Klasner, available on request from the Huntington Society of Canada. This publication, although it does not focus on juvenile HD, may be helpful to families.

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## III. Speech Therapy

It is important to have a speech/language pathologist provide an initial communication assessment.

Speech difficulties can begin with mild slurring of words and occasional nasality. As the disease progresses, intelligibility deteriorates significantly, with increasingly imprecise consonants, irregular and distorted breathing patterns, and inadequate breath support for speech.

A speech/language pathologist can help to devise strategies which will maintain speech ability for as long as possible. At the appropriate point, he/she can also introduce new tools to facilitate ongoing communication abilities. Computers with special software packages and other computerized communications strategies can be of tremendous help at this point.

For example, there are devices which allow the user to “speak” a word or phrase by pressing squares on the surface. Specific words and phrases can be programmed into the computer according to the child’s needs — from “I need a drink” to “I need a hug.” As physical problems increase, the size of the squares can be increased for ease of use.

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## IV. Swallowing

The speech/language pathologist will also provide initial and ongoing assessment of swallowing difficulties in children with HD, but families need to pay special attention to mealtimes.

Long before children become unable to feed themselves, leaving them alone to eat is a bad idea because so little food actually gets down. They tend to lose interest or spill the food, resulting in weight loss. Difficulty sticking with a task and increasing problems with handling and swallowing food contribute to the problem. To complicate matters, most children with HD dislike being fed. An often successful solution to consuming more food is to have someone remain with the child while he/she eats and to inform the child that there is something interesting to do once the meal is over.

When Jodie was 14, Dr. Smith asked Jodie’s mother, Brenda, if she would like to see a dietician to help her plan the meals. “No thanks,” said Brenda, “I am a vegetarian and consider myself quite knowledgeable on nutrition.” She remembers her shock when Jodie weighed in at only 48 pounds at her next appointment. “Although small for her age, she was also very thin. I cannot believe I let her get to that point right before my eyes. She was still feeding herself, but obviously not doing very well at it.”

Brenda comments on the experience: “We had tried to let her be independent, but obviously there came a time when this was no longer possible.” She consulted a dietician who advised her to feed Jodie as much high-calorie and high-fat foods as possible — especially foods which Jodie liked and could easily feed herself. Brenda found that “a lot of foods were better with sauces, with cheese melted in or with cream stirred into them. And most things are mashed up somewhat, though not puréed.” She was able to bring Jodie’s weight up to 60 pounds, a gain of 25%.

In general, the more weight children with HD can retain, the better off they are; for example, they sleep better and they fluctuate less in moods and mental function. Some parents find high calorie, carbohydrate-rich supplements such as Ensure® and Sustacal® very helpful in preventing weight loss. These supplements are low in vegetable fibre and therefore may exacerbate constipation. It is healthier to deal with constipation by various juices, such as carrot juice and prune juice, than to rely on stool softeners and suppositories.

Most children with advanced HD have inadequate fluid intake but they do not ask for fluids. If there is an incontinence problem, a vicious cycle can develop by restricting fluids to prevent bed wetting, because inadequate fluid intake makes colds and minor illnesses much more severe.

Water and similarly thin liquids, even liquid medication, may be much more difficult for the child to swallow than milkshakes or thickened fluids. Rebecca was able to administer liquid and tablet/capsule medications by adding them to small amounts of applesauce. Most caregivers become quite expert in this area as the child’s illness progresses, and are often better judges of what the child can handle than is the physician. It is also important to remember that although swallowing problems are expected as the illness progresses, they are often made worse by sedative medications.

## Feeding Tips

- ï Eating should be slow and deliberate.
- ï Be sure the child is positioned properly.
- ï Choose food of appropriate texture and temperature.
- ï Learn the Heimlich manoeuvre.
- ï Pay close attention to any coughing or choking incidents, and discuss them with the health care team.
- ï Make sure the child takes small bites and sips.
- ï Alternate solids and liquids.
- ï Have the child *idry swallow* or *idouble swallow* between bites.
- ï Have the child sit up after eating.

*Adapted from A Caregiver’s Handbook for Advanced-Stage Huntington Disease, by Jim Pollard (Huntington Society of Canada, 1999). This publication, although it does not focus on juvenile HD, may be helpful to families. Also of interest is Managing Swallowing Difficulties Associated with Huntington Disease, by Estelle Klasner, available on request from the Huntington Society.*

**Feeding tubes and gastrostomies.** In the later stages of the disease, when the child can no longer swallow solids or liquids without some entering the lung and producing aspiration pneumonia, it may be appropriate to consider a nasogastric feeding tube, passed through the nose, or a gastrostomy — an artificial opening into the body which permits food to be run directly into the stomach. Intravenous feeding is not a practical long-term option.

Feeding tubes today are superior to those previously available which had to be put down for each meal. Soft, thin tubes can be passed over a removable guidewire and can remain in place for days and weeks. However, the presence of the tube in the airway causes some irritation and increase of respiratory secretions. Some children are visibly uncomfortable even with the softest tubes. Nasogastric feeding tubes are therefore only a short-term treatment.

It is easier to put food and pills into a gastrostomy than a feeding tube, and many children with HD live long enough that gastrostomy becomes an issue. This method may seem extreme to some families, but comes as a welcome relief to others who have struggled with feeding problems and malnutrition. Gastrostomies can malfunction, and infection of the gastrostomy site is a frequent (but treatable) complication.

Feeding tubes and gastrostomies can raise important quality of life questions which affected individuals and their families must weigh and discuss themselves, and with their physician. Decisions about feeding tubes and gastrostomies are complex, and there are no easy answers for families or physicians. For example, it may be difficult for families to accept that their child can no longer eat food by mouth. Likewise, gastrostomies can be perceived as artificially prolonging life, and it may be difficult for families to accept the prospect of a tube being inserted into the stomach. For the physician, there may be awareness that a gastrostomy does not remove the potential for aspiration pneumonia, because the individual's own body secretions, such as saliva, can still cause choking and aspiration. At the same time, there may be only limited familiarity with the extent to which feeding tubes might improve a child's condition. The complexities of decisions about feeding tubes and gastrostomies call for families and physicians to share information and insights, and to collaborate on reaching the right decision for the affected individual — ideally in advance.

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## V. Behaviour and Judgement

Behaviour and judgement are eventually impaired in almost all people with HD. “On a day-to-day basis, medical problems aside, some of the most difficult situations seemed to result from behaviour that was inappropriate to the circumstance,” remembers Rebecca.

“Most of the distressful behaviours and situations, for me and for the children, came from them being where expectations no longer suited their capabilities and needs. It was also frustrating when health care professionals were not understanding enough, or flexible enough, to allow the young person to express thoughts or emotions appropriate to his/her capabilities — they tended to impose their own expectations and react negatively when those expectations were not met.”

Adolescence is a complex time for anyone. For the child with HD, it is even more difficult and complex. For example, “the child/young person may, at once, like watching cartoons, playing with dolls, and have a ‘crush’ on someone, with all the attending ‘responsible relationship’ problems!”

Poor judgment about sexual behaviour may also be a problem, especially for more mobile adolescents. Families may find it difficult to believe that the young person has an organic impairment of judgment and cognition because he/she is not confused or disoriented. Marla remembers her teenage daughter as being “argumentative, impulsive and prone to mood swings. She also became promiscuous.” Families will often accept a superficial psychological explanation more readily than a more complex one. For example, “she got pregnant because she was angry with her parents” is more readily accepted than “she got pregnant because she couldn’t understand the consequences of her behaviour.”

Adolescents with HD who have behaviour problems often act impulsively. Their promises to do better in the future do not mean much because they are likely to act before thinking. This is often a difficult concept for families to accept. The pattern can be similar to that seen in alcohol and drug abusers. For example, the adolescent makes sincere promises about future behaviour which he/she cannot keep and would not make if he/she had a realistic notion about why people get upset with his/her behaviour. The ability to monitor one’s behaviour and know when one is breaking the rules is often deficient in children with HD.

“When the screaming episodes first started, it was very hard for me to handle,” remembers Brenda. “There was no reasoning with her. I tried to be consistent, withdrawing a privilege as punishment. Sometimes I got so upset that I would scream back at her to stop. Nothing worked. There were some good days when she was a joy to be with. On these days, I would say to her, ‘Isn’t it nice to be good? We’re both so much happier!’ She would agree, but the next bad day, things would be exactly the same. Eventually, I learned to just let her scream. There was nothing else I could do. I would go into her room and give her something to drink because she was so hot. Sometimes she would calm down and come out of her room, but often she didn’t want to. The nights were bad also. After the screaming in the daytime

## Solving Behaviour Problems in HD

- ï Identify the main problem.
- ï Gather information about the problem.
- ï Review possible causes of behaviours.
- ï Set a realistic goal.
- ï Be flexible and ready to try several strategies.
- ï Reassure all individuals involved.

*From Understanding Behaviour in Huntington Disease, by Dr. Jane Paulsen (Huntington Society of Canada, 1999). This publication, although it does not focus on juvenile HD, may be helpful to families.*



“Keith gets very stubborn and frustrated at times and thinks he should have his own way. We don’t give in to him all the time. My other sons get upset with him at times because he can be very demanding.”

— Laura

started, there was never a night when she would sleep right through. Often she called me four or five times a night. She would sit on the toilet, and not necessarily go. I’d put her back to bed, and an hour or two later, she’d call me again.

“I think that when the behaviour problems start, it would be better not to try to treat the child the way you would a normal child,” says Brenda. “It is no use trying to reason with her. It just doesn’t mean anything to the child to say that, for example, you will turn off the TV if she throws things. She can only think of the present. She is only involved with what is happening now, not what will happen if she throws things. I think this has to do with the fact that she has no concept of time. She can remember past events, but she doesn’t know what you mean when you say that the school bus is coming in five minutes and we have to hurry. Her mind is only aware of what she can see now.”

When Abigail refuses to get dressed in the morning, Claudette usually leaves the room and comes back a few minutes later. “I will do this until she accepts getting dressed. Sometimes I will ask her brother to go to her room and help her get dressed. I find that it is important to break the cycle when behaviour problems occur. Leaving the room or asking someone else to do the job will usually work.”

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## VI. Seizures

About 25-30% of children with HD have recurrent seizures called “epilepsy”. These seizures may overshadow other medical problems because they lead to restrictions on the child’s activities.

Seizures usually develop after the first obvious motor abnormalities appear. Seizures are usually generalized (grand mal) and may be associated with cyanosis (turning blue) and falls. They are very frightening to witness. A teacher or parent observing a seizure often believes that the child is dying. Teachers fear that they will need to perform CPR and they often say things like “this child cannot come to school until this problem has been eliminated.”

Controlling seizures means chronic administration of anticonvulsant drugs. While no particular drug is specific for seizures due to HD, the most common anticonvulsant drugs often have disappointing results in children with HD. The drugs most likely to be effective are valproic acid and benzodiazepines such as clonazepam. In addition to valproic acid, some patients take lorazepam immediately following a seizure to prevent further seizures from occurring close together. There are several newer anticonvulsant medications, including lamotrigine, gabapentin, and topiramate, which may be tried if the older medications are ineffective.

As their illness progresses, a few children with HD develop focal seizures (partial seizures which involve changes in movement or sensation without loss of consciousness). These children may respond to anticonvulsants other than those mentioned.

The goal is to control seizures without side-effects such as drowsiness or increased difficulty swallowing and walking. Unfortunately, some children with HD have both a strong tendency to seizures and poor tolerance of anticonvulsant medication. Some of these children can do more and will have a better life if lower than maximum doses of anticonvulsant medication are used. For example, we accept the fact that the child will have an occasional seizure knowing that maximum doses of medication will make the child function much less well in school or sleep excessively. Similarly, a child whose seizures are controlled and has an anticonvulsant drug level below the usual therapeutic range should not ordinarily have his/her dosage increased. The aim is to use the smallest dose of anticonvulsants which controls seizures because of the likelihood that sedative anticonvulsant side-effects will increase the disability due to HD.

Drug administration is also a concern. Adolescents are often ambivalent about their medication — it reminds them of their serious illness. They often say that they have taken their medication, knowing full well that they have not. If the drugs do not seem to be working, parents should ensure that they are actually swallowed. Only one person should have the responsibility of giving the patient's medication because the chance of forgetting or of administering the wrong dose is much greater when several people handle the medication.

As the disease progresses, the child's ability to tolerate anticonvulsants and sedative drugs usually decreases, so he/she may appear groggy and over-medicated even though the amount of medication used may be no greater than that which was well tolerated in the past. There is a very small chance of serious organ toxicity from anticonvulsants. If this is going to occur, it will almost always develop in the first six months of drug use and can be recognized and dealt with if the child is being monitored regularly. Changes in diet or activity may alter tolerance of medications.

Some children with seizures also have single jerks of the limb or trunk called "myoclonic jerks" or "myoclonus". A myoclonic jerk is not the same as a seizure and the family and the physician must be careful not to use the wrong term. A child with well-controlled seizures may still have myoclonus. If myoclonus is severe or frequent, it might be appropriate to treat it, but it is usually not necessary to treat myoclonus as vigorously as seizures are treated.

The physician will usually do best by trying to prevent or reduce the number of tonic-clonic (grand mal) seizures and ignoring the occurrence of jerks. Tonic-clonic seizures are major/dramatic seizures associated with loss of consciousness and alternating cycles of tonus (straightening out or stiffening) and clonus (flinging or jerking). If jerks cause falls, injuries, or interfere with food/fluid/medication intake, it is reasonable to try to reduce their frequency. This may require use of additional antimyoclonic drugs like valproate.

Families may notice an increased number of jerks or other warning signs in the days or hours preceding a tonic-clonic seizure. If experience with that particular child shows that it is possible to predict the occurrence of big seizures, there may be an opportunity to prevent them by using extra doses of medication. The family should not attempt this independently, however — it is very important that the treating physician know exactly what dose of what medication is being used, because over-medication can cause trouble. Therefore, it is important that seizure medication be given according to a strict schedule, at the same time of day, even on holidays. The child can take his/her early morning medicine and go back to sleep if no activity is planned.

Optimal therapy for seizures rests on a solid partnership between parents and physicians. Physicians must be aware that, as the disease progresses, and by necessity, families acquire a vast and intimate knowledge of HD and its effects on their child. Their insights and observations will be invaluable in tailoring therapy to fit individual needs.

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## VII. Other Drug Treatment

The general principle of drug therapy in HD is for the family and physician to work as a team, with both understanding why a drug is being used, what the potential benefits and side-effects are, when its effectiveness will be checked, and what to do if there are problems.

Many physicians have heard that neuroleptic (antipsychotic) drugs such as haloperidol and thiorazine are helpful for adults with HD. This view derives from early animal experiments which suggested that neuroleptics could reduce chorea, but which did not consider questions of alertness or learning ability. Because chorea is rarely of major importance in juvenile HD, neuroleptics may not be advisable because they may produce drowsiness, falls, swallowing difficulties caused by dry mouth, and increased rigidity. These drugs should only be continued if there is a clear improvement in overall function.

Newer drugs such as risperdal, olanzapine, and quetiapine may be considered because they are reported to be associated with fewer side-

effects. Tetrabenazine is an older drug (currently available through a special access programme in Canada) which can also be useful.

Muscle relaxants such as benzodiazepines, baclofen or tizanidine may help to reduce stiffness in the child with HD. Any benefit from reduced stiffness must always be balanced with the cost of increased drowsiness, difficulty swallowing, and trouble attending to schoolwork.

Many children with HD have trouble sleeping. Low doses of a benzodiazepine can be helpful, though there is much variation from one person to another, and in each individual over time. Parents and physicians should remember that the sudden discontinuation of the drug can prompt withdrawal symptoms including agitation, sleeplessness, and seizures.

“The doctor has prescribed a few different medications over the years. He explained that we were just trying them, that nothing could take away Jodie’s symptoms, but we could try to make them slightly better. Some medications were helpful in making her less stiff. One or two that we tried just zonked her out, to the point where she was really tired or unaware of her surroundings. The doctor always listened to my input. At my suggestion, we tried a lower dose, or a higher dose of certain medications, if I felt it was helping.”

— Brenda

Antidepressant drugs such as amitriptyline, desipramine, and fluoxetine are helpful in adults with HD, even without overt signs of depression. They have not been used as much in juvenile cases and their place in therapy is still uncertain. They reduce sleep disturbances in some patients, but may aggravate seizures, jerks, and trembling. While the rigidity and akinesia (lack of movement) shown by many juvenile patients reminds physicians of Parkinson disease (a disorder which is responsive to L-DOPA), use of DOPA or other dopamine-related drugs is rarely helpful.

Other behavioral or psychiatric symptoms may respond to a variety of medications, counselling, or environmental modification strategies. Anxiety, obsessive-compulsive disorder, sexual disinhibition, mania, and explosive behaviors are common and can be severe. Benzodiazepines such as alprazolam, lorazepam, or buspirone can be used to treat anxiety. Obsessive thinking or compulsions can respond to serotonin reuptake inhibitors such as fluvoxamine or to clomipramine. Aggressive physical or sexual behaviors may require a combined approach of behavioral modification and medications. Neuroleptics, beta blockers (such as propranolol), and anticonvulsants such as valproate, carbamazepine and gabapentin are sometimes used for control of aggressive, irritable, or manic behaviors.

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## VIII. Juvenile HD and Family Life

Just as it calls on families to cope with difficult physical and behavioural problems, so too does juvenile HD place everyone affected under tremendous emotional and financial pressures.

Rebecca summarizes some of these challenges. “My perception as a wife and mother,” she says, “reflects many things, not the least of which is sorrow. But HD inflicts an additional insult and burden for a parent of an affected child. It is unbearable enough for any parent to see a child endure a devastating, life-threatening illness, and to lose that child. But with HD, you do not have the other parent (who is usually your life-partner as well) to share in the painful, overwhelming experience. Instead, by the very nature of the disease, they are another part of the painful experience. The strain of caring for our two affected children, our ‘normal’ child and my HD-affected husband was tremendous, both physically and emotionally. It seemed that I was always torn between spending my time and energy caring for, and trying to meet the needs of, each of them. It was difficult to remember to include my needs in the equation.”

When one parent has HD, the other is often the sole wage earner. This combination of pressures can sometimes be felt in the form of intolerance, guilt and resignation. Brenda recalls, “One winter day, I took Jodie tobogganing. We had a few rides together and then I wanted to get my camera to get a shot of Jodie coming down the hill. I set her up at the top of the hill and showed her how to sit on the toboggan. I wanted her to wait until I went part way down the hill, so I could take her picture coming down. But she had trouble holding the toboggan and then sitting on it by herself. I remember yelling at her: ‘What’s the matter with you?’ This is one of the things I feel guilty about now. I know what the matter was. It wasn’t her fault. It was Huntington’s.”

At times, though, it is hard to separate the person from the disease. One young woman remembers growing up with HD in her family. “My father, brother and sister were all affected with Huntington disease and I fulfilled many roles: sister, daughter, individual at risk, and caregiver. I remember attending support group meetings and being confused as to which sub-group I should attend: at-risk or caregiver? My mother and I talked constantly about my feelings regarding HD. It was so helpful to have her as a sounding board and as a resource. A recurring feeling I had throughout my high school and early college years was one of resentment for having to take care of my family members. Of course, I often felt guilty for having these feelings because I loved them and did want to help them. But their care seemed to take away from my social and study time. The most difficult things I had to deal with most of the time were feelings of anger and even hate. My best way of dealing with these feelings was to remind myself that it was the disease I was angry with, not the person. Unfortunately, they often seemed one and the same. It is important to say that it was, and still is, my constant communication with my mother that helped me through it all.”

It is also very difficult for a young person to watch a sibling become progressively ill. Two brothers, 18 and 15 respectively, were unable to express their feelings. “I’m sure they sit there and wonder if some day they’ll be like that,” said their mother. “It’s very hard to live with this every single day. We take one day at a time, whether good or bad.”

Huntington disease puts a strain on friendships as well. “The disease extends over a long period of time, particularly when two generations are involved simultaneously. For the most part, my family and friends were supportive but they needed to pull away from time to time,” said Rebecca. “The intensive demands of HD tend to ‘burn out’ people. It causes them to run low on what they feel they can do or how much they can tolerate. It seems they need to draw away and gather strength for the next period of commitment. When friends and family cycled in and out of the picture, it often felt as though we were being deserted. It made support somewhat tenuous and unpredictable. Of course, I understood that need, but as the parent and wife, I did not have that option.”

Yet Carl J. Crosley, a paediatric neurologist who has treated children with HD, offers an important reminder: namely that families focus also on “education, growing up, and enjoyment. It takes much awareness and a delicate balance to not over-emphasize the purely medical aspects of their illness and ignore what they are really concerned about.”

Families should enjoy and celebrate all the happy moments and small victories life brings — a new skill, a good day at school, a joyous family get-together. The emphasis should be on *living* with juvenile HD.

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## Later Stages of the Disease

As the illness progresses, children with HD are capable of doing less. They may be difficult to live with because of sleep disturbances, mood swings, and general uncooperativeness. They may be receiving large amounts of medication, and have growing difficulty participating in family activities. Mobility decreases, while speech and swallowing problems grow more severe. There is a need for strategies to be adapted as the disease progresses. Ultimately, placement of an affected child in an appropriate long-term care facility will need to be considered.

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### I. Leisure and social time

Planning activities for the child in the later stages of Huntington disease is very challenging but essential to maintaining quality of life. For suggestions on activities that are appropriate, consult a therapeutic recreation specialist with expertise in working with children with disabilities. A plan can then be developed based on the individual interests and abilities of the child. The following activities are good examples:

- Visits with other children
- Visits with pets
- Talking books/short stories
- Music
- Daily calendar time. Set aside time each day to review the date (day and month) and the day's schedule. Take this time to highlight important occasions or special memories associated with the day.
- Colourful decorations. Use a large bulletin board to decorate the child's room according to the seasons and special holidays. This provides varied visual stimulation and a focus for conversation. It will also help the child keep track of time.
- Board games like bingo and Rumoli. Some adaptations may be required, such as replacing small tokens with large buttons or other household items.
- Card games. Card holders may be needed.
- "Rat-A-Tat Cat", a card game that facilitates interaction with others and encourages communication, thinking and jokes. (Available at <[www.gamewright.com](http://www.gamewright.com)> or 1-800-345-6665)

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## II. Institutionalization

Decisions about institutionalization are complex and emotionally draining for everyone concerned.

As the illness progresses, most families will need help in providing care. Visiting nurses and other caregivers can check that feedings are being handled properly, free up parents, siblings and relatives for other family responsibilities, or sometimes just give the primary caregivers a break. Respite programmes can also be of great value.

But at some point, the very difficult question of institutionalization will arise. Finding appropriate placement can be a tremendous challenge for families and members of the care team. In many cases, nursing homes are not a viable option, and would in any case have problems accommodating a juvenile. Families must also be prepared to continue to play a key role in caring for the affected child, even after institutionalization.

Brenda recalls Jodie's institutionalization as the toughest decision of her life. With summer holidays approaching, she realized that she would need a break during the two months that Jodie was home from school. She arranged for Jodie to go to the Children's Rehabilitation Centre for two weeks. During that time, Jodie's behaviour problems improved dramatically. Brenda hoped that this improved behaviour would continue when she came home after the summer. "No such luck. She went right back to the screaming episodes and the sleepless nights. Then I did a lot of thinking — about how she was wearing herself out screaming and about how happy she was when she was at the Rehab Centre. I love her so much and I didn't want to let her go. Sometimes we were very close and we had some good days that were very precious to me. But eventually I admitted to myself that she would be better off not staying at home anymore."

Jodie stayed at the Rehab Centre for two weeks every month, and at home for the other two weeks. "When she was at home for the two weeks, the negative behaviour continued. When she was at the Rehab Centre, she was much better. I knew that I had made the right decision." Only after eight months was it possible to find an appropriate permanent home in the community.

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## III. Death

Death is the inevitable end for all of us. It just seems more cruel and unfair for children with Huntington disease.

"Losing a child is one of life's deepest wounds," says Rebecca. "With HD, once the symptoms begin, you lose your child over and over

“I guess many of the feelings I experienced, and still experience, are common to any parent who has seen the life of a child end prematurely. I wonder what each of them would have grown to be, and the experiences we missed together. When death comes, it is both the deepest of sorrows and the greatest of releases.”

—Rebecca

again. I watched as my sweet children who had learned to walk, to feed themselves, to speak, to use the bathroom, to ride a bicycle — were reduced to accepting diapering, to learning to use a wheelchair while losing the ability to ride a bicycle, indeed even to walk. It was very painful to see the confusion on their faces when something that was once easily accomplished became a struggle for them. There was almost a look saying ‘Don’t I know how to do this? What’s wrong — why can’t I do it?’”

In trying to prepare her daughter for the inevitable end, Claudette approached the subject in an open and straightforward manner.

“I tried to demystify death as much as possible. I told Abigail that normally, parents die first. We all come into this world to accomplish a certain thing and, most of the time, it takes many years for people to understand their role in life. Some people don’t ever fulfil it, even if they live to be 100 years old. Sometimes though, a very special child comes into the world who can do her job on this earth in a shorter time.

“When her job is done, she will leave her physical body and will fly through the universe, making sure that beauty is always a part of our world. I told her that when her physical state is over she will still be part of my life because every time I see beauty, I will see her face. She is one of those very special children.

“In a way, children with Huntington’s are special. They have such an acceptance of life. We can learn so much from them. Of course, the agony of the situation is unbearable. The waiting for death is awful. But we try to focus on life. This is not the life I planned for her, but it’s the best life I can give her. Huntington disease has changed our lives, but we have grown tremendously because of it. There are lots of tears and lots of laughter, too.

“I asked people close to Abigail to tell her what she has meant to them,” Claudette said. “I wanted her to know that she has touched them and that she has done her job. I think that it is important to her to know that she will not be forgotten.” Claudette laughs with delight, remembering that Abigail “told her classmates that she would like to be remembered as the girl with the most beautiful smile in the world.”

“I told her that she will be at peace when she dies — that only her body is sick. Her mind and soul and heart are perfect and will live on. I will always hear her voice inside of my heart. When she heard that, Abigail told me she’ll play tricks on me with her voice. She has a very good sense of humour!

“She has given me unconditional love, which is so rare.” Claudette smiled. “A gift is a bonus, not something one needs, and I tell her often that she is my gift from Life.”



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— RM, October 2000

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