DOWNLOAD PDF HUNTINGTONS DISEASE

Chapter 1 : Huntington's disease - Symptoms and causes - Mayo Clinic

Huntington's disease (HD) is a progressive brain disorder caused by a defective gene. This disease causes changes in the central area of the brain, which affect movement, mood and thinking skills. Huntington's disease is a progressive brain disorder caused by a single defective gene on chromosome 4.

This test measures the electrical activity in your brain. Brain-imaging tests can also be used to detect physical changes in your brain. Magnetic resonance imaging MRI scans use magnetic fields to record brain images with a high level of detail. Computed tomography CT scans combine several X-rays to produce a cross-sectional image of your brain. Psychiatric Tests Your doctor might ask you to undergo a psychiatric evaluation. This evaluation checks your coping skills, emotional state, and behavioral patterns. A psychiatrist will also look for signs of impaired thinking. You may be tested for substance abuse to see if drugs might explain your symptoms. A genetic test can definitively diagnose this condition. Genetic testing may also help you decide whether or not to have children. Medications Medications can provide relief from some of your physical and psychiatric symptoms. The types and amounts of drugs needed will change as your condition progresses. Involuntary movements may be treated with tetrabenazine and antipsychotic drugs. Muscle rigidity and involuntary muscle contractions can be treated with diazepam. Depression and other psychiatric symptoms can be treated with antidepressants and mood-stabilizing drugs. Therapy Physical therapy can help improve your coordination, balance, and flexibility. With this training, your mobility is improved, and falls may be prevented. Occupational therapy can be used to evaluate your daily activities and recommend devices that help with: Speech therapists can also help with swallowing and eating problems. Psychotherapy can help you work through emotional and mental problems. It can also help you develop coping skills. There is no way to stop this disease from progressing. The rate of progression differs for each person and depends on the number of genetic repeats present in your genes. A lower number usually means that the disease will progress more slowly. The early-onset form generally progresses at a faster rate. People may live for only 10 to 15 years after the onset of symptoms.

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Chapter 2 : Huntington's Disease | HD | MedlinePlus

Read the accounts of people from all walks of life affected by Huntington's disease. Friends, family, caregivers, volunteers, and those afflicted with HD share their experiences and observations.

It is a devastating disease that causes damage to brain cells, or neurons. It happens when a faulty gene causes toxic proteins to collect in the brain. Another , or more people are at risk of developing the condition. The first signs normally appear between the ages of 30 and 50 years. The disease happens when a faulty gene makes an abnormal version of the huntingtin protein. Early symptoms may include mood swings, clumsiness, and unusual behavior. During the later stages of the disease, choking becomes a major concern. There is currently no cure, but medications may help relieve symptoms. It is an inherited disease that happens due to faulty genes. Toxic proteins collect in the brain and cause damage, leading to neurological symptoms. As parts of the brain deteriorate, this affects movement, behavior, and cognition. It becomes harder to walk, think, reason, swallow, and talk. Eventually, the person will need full-time care. The complications are usually fatal. There is currently no cure, but treatment can help with symptoms. Changes in mood and thinking can be an early sign. Signs and symptoms are most likely to appear between the ages of 30 and 50 years, but they can occur at any age. They tend to worsen over 10 to 20 years. The key symptoms include: In some people, depression occurs before motor skills are affected. Mood swings and unusual behavior are common early signs. Early signs and symptoms Early symptoms may not be recognized if HD has not previously occurred in the family. It can take a long time to reach a diagnosis. Initial signs and symptoms include: However, most people do these from time to time. The middle and later stages In time, symptoms become more severe. These include physical changes, loss of motion control, and emotional and cognitive changes. Physical changes difficulty speaking, including looking for words and slurring weight loss, leading to weakness difficulty eating and swallowing, as the muscles in the mouth and diaphragm may not work properly risk of choking, especially in the later stages uncontrollable movements There may be uncontrollable body movements, including: Eventually they may become slower as the muscles become more rigid. Emotional changes These may alternate rather than occurring consistently.

Chapter 3 : Huntington's disease - Wikipedia

Huntington disease is a progressive brain disorder that causes uncontrolled movements, emotional problems, and loss of thinking ability (cognition).. Adult-onset Huntington disease, the most common form of this disorder, usually appears in a person's thirties or forties.

Evidence of substance abuse Brain imaging and function Your doctor may order brain-imaging tests for assessing the structure or function of the brain. The imaging technologies may include magnetic resonance imaging MRI or computerised tomography CT scans that provide detailed images of brain structures. These tests can also be used to rule out other conditions that may be causing symptoms. Before undergoing such a test, the genetic counselor will explain the benefits and drawbacks of learning test results. Predictive genetic test A genetic test can be given to someone who has a family history of the disease but shows no signs or symptoms. This is called predictive testing. Some people may elect to do the test because they find it more stressful not knowing. Others may want to take the test before they make decisions about having children. Risks may include problems with insurability or future employment and the stresses of facing a fatal disease. In principle, federal laws exist that make it illegal to use genetic testing information to discriminate against people with genetic diseases. These tests are only performed after consultation with a genetic counselor. But medications can lessen some symptoms of movement and psychiatric disorders. And multiple interventions can help a person adapt to changes in his or her abilities for a certain amount of time. Medication management is likely to evolve over the course of the disease, depending on the overall treatment goals. Also, drugs to treat some symptoms may result in side effects that worsen other symptoms. Therefore, the treatment goals and plan will be regularly reviewed and updated. Medications for movement disorders Drugs to treat movement disorders include the following: A serious side effect is the risk of worsening or triggering depression or other psychiatric conditions. Other possible side effects include drowsiness, nausea and restlessness. Antipsychotic drugs, such as haloperidol Haldol and chlorpromazine, have a side effect of suppressing movements. Therefore, they may be beneficial in treating chorea. However, these drugs may worsen involuntary contractions dystonia and muscle rigidity. Other drugs, such as risperidone Risperdal and quetiapine Seroquel , may have fewer side effects but still should be used with caution, as they may also worsen symptoms. Other medications that may help suppress chorea include amantadine, levetiracetam Keppra, others and clonazepam Klonopin. It may also cause leg swelling and skin discoloration. Side effects of levetiracetam include nausea, stomach upset and mood swings. It also has a high risk of dependence and abuse. Medications for psychiatric disorders Medications to treat psychiatric disorders will vary depending on the disorders and symptoms. Possible treatments include the following: Antidepressants include such drugs as citalopram Celexa , escitalopram Lexapro , fluoxetine Prozac, Sarafem and sertraline Zoloft. These drugs may also have some effect on treating obsessive-compulsive disorder. Side effects may include nausea, diarrhea, drowsiness and low blood pressure. Antipsychotic drugs â€” such as quetiapine Seroquel , risperidone Risperdal and olanzapine Zyprexa â€” may suppress violent outbursts, agitation, and other symptoms of mood disorders or psychosis. However, these drugs may cause different movement disorders themselves. Mood-stabilizing drugs that can help prevent the highs and lows associated with bipolar disorder include anticonvulsants, such as valproate Depacon , carbamazepine Carbatrol, Epitol, Tegretol and lamotrigine Lamictal. Psychotherapy A psychotherapist â€” a psychiatrist, psychologist or clinical social worker â€” can provide talk therapy to help a person manage behavioral problems, develop coping strategies, manage expectations during progression of the disease and facilitate effective communication among family members. A speech therapist can help improve your ability to speak clearly or teach you to use communication devices â€” such as a board covered with pictures of everyday items and activities. Speech therapists can also address difficulties with muscles used in eating and swallowing. Physical therapy A physical therapist can teach you appropriate and safe exercises that enhance strength, flexibility, balance and coordination. These exercises can help maintain mobility as long as possible and may reduce the risk of falls. Instruction on appropriate posture and the use of supports to improve posture may help lessen the severity of some movement problems. When the use of a walker or wheelchair is

required, the physical therapist can provide instruction on appropriate use of the device and posture. Also, exercise regimens can be adapted to suit the new level of mobility. These strategies may include: As the disease progresses, the person will become more dependent on caregivers. A number of issues will need to be addressed, and strategies to cope with them will evolve. Eating and nutrition Factors regarding eating and nutrition include the following: Difficulty eating, higher caloric needs due to physical exertion or unknown metabolic problems may be the cause. To get adequate nutrition, more than three meals a day or the use of dietary supplements may be necessary. Difficulty with chewing, swallowing and fine motor skills can limit the amount of food you eat and increase the risk of choking. Problems may be minimized by removing distractions during a meal and selecting foods that are easier to eat. Utensils designed for people with limited fine motor skills and covered cups with straws or drinking spouts also can help. Local and state health or social service agencies may provide daytime care for people with the disease, meal assistance programs or respite for caregivers. Creating legal documents that define end-of-life care can be beneficial to everyone. They empower the person with the disease, and they may help family members avoid conflict late in the disease progression. Your doctor can offer advice on the benefits and drawbacks of care options at a time when all choices can be carefully considered. Matters that may need to be addressed include: Care in the advanced stages of the disease will likely require in-home nursing care or care in an assisted living facility or nursing home. Hospice services provide care at the end of life that helps a person approach death with as little discomfort as possible. This care also provides support and education to the family to help them understand the process of dying. For example, these directions might indicate whether or not the person wants life-sustaining interventions or aggressive treatment of an infection. These legal documents enable you to identify one or more people to make decisions on your behalf. You may create an advance directive for medical decisions or financial matters. A review of your symptoms, mental state, medical history and family medical history can all be important in the clinical assessment of a potential neurological disorder. What you can do Before your appointment, make a list that includes the following: This person can provide support and offer a different perspective on the effect of symptoms on your functional abilities. What to expect from your doctor Your doctor is likely to ask you a number of questions, including the following: When did you begin experiencing symptoms? Have your symptoms been continuous or intermittent? Has anyone in your family been diagnosed with another movement disorder or psychiatric disorder? Are you having trouble performing work, schoolwork or daily tasks? Has anyone in your family died young? Is anyone in your family in a nursing home? Is anyone in your family fidgety or moving all the time? Have you noticed a change in your general mood? Do you feel sad all of the time? Have you ever thought about suicide?

Chapter 4 : Overview | Huntington's Disease | blog.quintoapp.com

Huntington's disease is an inherited disease that causes the progressive breakdown (degeneration) of nerve cells in the brain. Huntington's disease has a broad impact on a person's functional abilities and usually results in movement, thinking (cognitive) and psychiatric disorders.

Regions of the brain have differing amounts and reliance on these types of neurons, and are affected accordingly. The remaining variation is attributed to environment and other genes that modify the mechanism of HD. In some cases the onset may be so late that symptoms are never noticed. Inheritance is independent of gender, and the phenotype does not skip generations. This probability is sex-independent. Individuals with both genes affected are rare. For some time HD was thought to be the only disease for which possession of a second mutated gene did not affect symptoms and progression, [32] but it has since been found that it can affect the phenotype and the rate of progression. Early damage is most evident in the striatum , but as the disease progresses, other areas of the brain are also more conspicuously affected. Early symptoms are attributable to functions of the striatum and its cortical connectionsâ€”namely control over movement, mood and higher cognitive function. Huntingtin HTT is expressed in all cells. The highest concentrations are found in the brain and testes , with moderate amounts in the liver , heart , and lungs. It interacts with proteins which are involved in transcription, cell signaling , and intracellular transporting. Caspase , an enzyme which plays a role in catalyzing apoptosis, is thought to be activated by the mutated gene through damaging the ubiquitin-protease system. It also acts as an anti-apoptotic agent preventing programmed cell death and controls the production of brain-derived neurotrophic factor , a protein which protects neurons and regulates their creation during neurogenesis. HTT also facilitates vesicular transport and synaptic transmission and controls neuronal gene transcription. Over time, the aggregates accumulate to form inclusion bodies within cells, ultimately interfering with neuron function. Inclusion bodies have been found in both the cell nucleus and cytoplasm. Mutant Huntingtin protein has been found to play a key role in mitochondrial dysfunction. Excitotoxins may cause damage to numerous cellular structures. Although glutamine is not found in excessively high amounts, it has been postulated that because of the increased vulnerability, even normal amounts glutamine can cause excitotoxins to be expressed. The most prominent early effects are in a part of the basal ganglia called the neostriatum , which is composed of the caudate nucleus and putamen. Their functions are not fully understood, but current theories propose that they are part of the cognitive executive system [18] and the motor circuit. To initiate a particular movement, the cerebral cortex sends a signal to the basal ganglia that causes the inhibition to be released. Damage to the basal ganglia can cause the release or reinstatement of the inhibitions to be erratic and uncontrolled, which results in an awkward start to motion or motions to be unintentionally initiated, or a motion to be halted before, or beyond, its intended completion. The accumulating damage to this area causes the characteristic erratic movements associated with HD. Thus, the glutamines on CBP interact directly with the increased numbers of glutamine on the HTT chain and CBP gets pulled away from its typical location next to the nucleus. Even before the onset of symptoms, genetic testing can confirm if an individual or embryo carries an expanded copy of the trinucleotide repeat in the HTT gene that causes the disease. Genetic counseling is available to provide advice and guidance throughout the testing procedure, and on the implications of a confirmed diagnosis. If these are abrupt and have random timing and distribution, they suggest a diagnosis of HD. Cognitive or behavioral symptoms are rarely the first symptoms diagnosed; they are usually only recognized in hindsight or when they develop further. Cerebral atrophy can be seen in the advanced stages of the disease. Functional neuroimaging techniques, such as functional magnetic resonance imaging fMRI and positron emission tomography PET , can show changes in brain activity before the onset of physical symptoms, but they are experimental tools, and are not used clinically. A positive result is not considered a diagnosis, since it may be obtained decades before the symptoms begin. However, a negative test means that the individual does not carry the expanded copy of the gene and will not develop HD. A person who tests positive for the disease will develop HD sometime within their lifetime, provided he or she lives long enough for the disease to appear. It may cause symptoms, usually

later in the adult life. It is not associated with symptomatic disease in the tested individual, but may expand upon further inheritance to give symptoms in offspring. Not associated with HD. It occurred at higher rates within personal relationships than health insurance or employment relations. This technique, where one or two cells are extracted from a typically 4- to 8-cell embryo and then tested for the genetic abnormality, can then be used to ensure embryos affected with HD genes are not implanted, and therefore any offspring will not inherit the disease. Some forms of preimplantation genetic diagnosisâ€"non-disclosure or exclusion testingâ€"allow at-risk people to have HD-free offspring without revealing their own parental genotype, giving no information about whether they themselves are destined to develop HD. In non-disclosure testing, only disease-free embryos are replaced in the uterus while the parental genotype and hence parental risk for HD are never disclosed. An amniocentesis can be performed if the pregnancy is further along, within 14â€"18 weeks. This procedure looks at the amniotic fluid surrounding the baby for indicators of the HD mutation. The parents can be counseled on their options, which include termination of pregnancy , and on the difficulties of a child with the identified gene. Other autosomal dominant diseases that can be misdiagnosed as HD are dentatorubral-pallidoluysian atrophy and neuroferritinopathy. There are also autosomal recessive disorders that resemble sporadic cases of HD. These include chorea acanthocytosis and pantothenate kinase-associated neurodegeneration. One X-linked disorder of this type is McLeod syndrome. This is a feeding tube, permanently attached through the abdomen into the stomach, which reduces the risk of aspirating food and provides better nutritional management. Physical therapists may implement fall risk assessment and prevention, as well as strengthening, stretching, and cardiovascular exercises. Walking aids may be prescribed as appropriate. Physical therapists also prescribe breathing exercises and airway clearance techniques with the development of respiratory problems. Participation in rehabilitation programs during early to middle stage of the disease may be beneficial as it translates into long term maintenance of motor and functional performance. Rehabilitation during the late stage aims to compensate for motor and functional losses. Genetic counseling benefits these individuals by updating their knowledge, seeking to dispel any unfounded beliefs that they may have, and helping them consider their future options and plans. Also covered is information concerning family planning choices, care management, and other considerations. A longer repeat results in an earlier age of onset and a faster progression of symptoms. The largest risk is pneumonia , which causes death in one third of those with HD. As the ability to synchronize movements deteriorates, difficulty clearing the lungs and an increased risk of aspirating food or drink both increase the risk of contracting pneumonia. The second greatest risk is heart disease , which causes almost a quarter of fatalities of those with HD. The rate of occurrence is highest in peoples of Western European descent, averaging around 7 per , people, and is lower in the rest of the world; e.

Chapter 5 : Huntington's disease: Symptoms, causes, and treatment

Huntington's disease (HD), also known as Huntington's chorea, is an inherited disorder that results in death of brain cells. The earliest symptoms are often subtle problems with mood or mental abilities.

Stages of hd The Scope of hd Who is at-risk? Today, there are approximately 30, symptomatic Americans and more than , at-risk of inheriting the disease. Symptoms usually appear between the ages of 30 to 50, and worsen over a 10 to 25 year period. Ultimately, the weakened individual succumbs to pneumonia, heart failure or other complications. Everyone has the gene that causes HD, but only those that inherit the expansion of the gene will develop HD and perhaps pass it on to each of their children. Every person who inherits the expanded HD gene will eventually develop the disease. Pictured above are the basal ganglia - a group of nerves cell clusters, called nuclei. These nuclei play a key role in movement and behavior control and are the parts of the brain most prominently affected in early HD. Stages of HD Although symptoms of HD vary from person to person, even within the same family, the progression of the disease can be roughly divided into three stages. Early stage HD usually includes subtle changes in coordination, perhaps some involuntary movements chorea , difficulty thinking through problems and often a depressed or irritable mood. Medications are often effective in treating depression or other emotional problems. The effects of the disease may make the person less able to work at their customary level and less functional in their regular activities at home. In the middle stage, the movement disorder may become more of a problem. Medication for chorea may be considered to provide relief from involuntary movements. Occupational and physical therapists may be needed to help maintain control of voluntary movements and to deal with changes in thinking and reasoning abilities. Diminished speech and difficulty swallowing may require help from a speech language pathologist. Ordinary activities will become harder to do. In the late stage, the person with HD is totally dependent on others for their care. Choking becomes a major concern. Chorea may be severe or it may cease. At this stage, the person with HD can no longer walk and will be unable to speak. However, he or she is generally still able to comprehend language and retains an awareness of family and friends. When a person with HD dies, it is typically from complications of the disease, such as choking or infection and not from the disease itself. In all stages of HD, weight loss can be an important complication that can correspond with worsening symptoms and should be countered by adjusting the diet and maintaining appetite. Within a family, multiple generations may have inherited the disease. Those at-risk may experience tremendous stress from the uncertainty and sense of responsibility. In the community, lack of knowledge about HD may keep friends and neighbors from offering social and emotional support to the family, fostering unnecessary isolation. HD affects both sexes and all races and ethnic groups around the world. If the child has not inherited this expanded gene, he or she will never develop the disease and cannot pass it on to their children. Inheritance is independent of gender. Many people see no benefit in knowing that they will someday develop the disease. Others want an end to uncertainty so that they can make informed choices about the future. The decision whether to test or not is intensely personal and there is no "right" answer. Testing procedures at these centers involves sessions with professionals who are knowledgeable about HD and the local services available. It may take several weeks to receive the results once the genetic test is complete. Genetic testing for children is typically prohibited before the age of 18, as the child may not understand the full implications of testing and may be vulnerable to pressure from others. However, a child under the age of 18 may be tested to confirm a diagnosis of juvenile onset HD after a thorough neurological exam. Prenatal Testing For families wishing to have a child who does not have the gene that causes HD, there are a few options. This can be done without informing the at-risk patient whether or not they have the gene that causes HD. If a woman is already pregnant, she can receive testing for the fetus with a chorionic villus biopsy at weeks or via amniocentesis at weeks. The symptoms of Juvenile HD JHD are somewhat different than adult onset HD and may include stiff or awkward walking, increased clumsiness or changes in speech. The ability to learn new information may decline and the child may lose skills they previous had. JHD typically progresses more rapidly than adult onset HD. In , researchers identified the gene that causes HD. Since then, research has moved quickly towards developing treatments and, ultimately, a cure.

HDSA supports the goals of clinical and basic research at leading research facilities globally. Clinical and observational trials are an important way you can help to sustain the momentum of HD research and move potential new therapies through the approval process. Visit the Research section of the HDSA website for more information and to find a trial in your area. There are opportunities for all HD family members â€" gene positive, at-risk, gene negative, and caregivers â€" to participate. Where to find help? With HDSA, no one fights alone.

Chapter 6 : Huntington's Disease Causes - Huntington's Disease News

A diagnosis of Huntington's disease may come as quite a shock. There's a lot to take in. But tapping into a support system, such as a social worker, therapist, or support group, can make the.

An exam of the nervous system will also be done. Other tests that may show signs of Huntington disease include: Treatment There is no cure for HD. There is no known way to stop the disease from getting worse. The goal of treatment is to slow the symptoms and help the person function for as long as possible. Medicines can be prescribed, depending on the symptoms. Dopamine blockers may help reduce abnormal behaviors and movements. Drugs such as amantadine and tetrabenazine are used to try to control extra movements. Depression and suicide are common among persons with HD. As the disease progresses, the person will need assistance and supervision, and may eventually need hour care. Support Groups These resources can provide more information on HD: People with HD usually die within 15 to 20 years. The cause of death is often infection. Suicide is also common. It is important to realize that HD affects people differently. The number of CAG repeats may determine the severity of symptoms. People with few repeats may have mild abnormal movements later in life and slow disease progression. Those with a large number of repeats may be severely affected at a young age. Prevention Genetic counseling is advised if there is a family history of HD. Experts also recommend genetic counseling for couples with a family history of this disease who are considering having children. Parkinson disease and other movement disorders. Nelson Textbook of Pediatrics. Review provided by VeriMed Healthcare Network.

Chapter 7 : Symptoms of Huntington's Disease - Huntington's Disease News

Huntington's disease is a hereditary condition in which your brain's nerve cells gradually break down. This affects your physical movements, emotions, and cognitive abilities.

Chapter 8 : Huntington's Disease | Stanford Health Care

Huntington's disease (HD) is a complex disorder that affect's a person's ability to feel, think, and move. Symptoms tend to worsen over time and the disease often runs in families. In people with one parent with HD, the chances of them developing it are

Chapter 9 : Huntington disease: MedlinePlus Medical Encyclopedia

Huntington's disease (HD) is an inherited disorder that causes brain cells, called neurons, to die in various areas of the brain, including those that help to control voluntary (intentional) movement.