



Pain Perception and Reaction in Huntington's Disease: Impact on Life

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DESCRIPTION

Huntington disease is rare, inherited disease that causes progressive breakdown of nerve cells in the brain. It affects movement, thinking, and emotions. Symptoms usually begin between 30 and 50 years of age, but can start at any age. There is no cure for the disease, but treatments can help manage some of the symptoms. This disease is caused by an inherited difference in a single gene called HTT, which is located on chromosome. This gene normally provides instructions for making a protein called huntingtin, which is involved in brain development and function. However, in people with Huntington's disease, the gene has a mutation that consists of a repeated pattern of three nucleotides Cytosine-Adenine-Guanine (CAG) in the DNA (Deoxyribonucleic acid) molecule. The more CAG repeats a person has, the more likely they are to develop the disease and the earlier it may start. The mutation causes the production of an abnormal form of huntingtin protein, which damages nerve cells in certain regions of the brain that control movement, thinking and mood. Huntington's disease is an autosomal dominant disorder, which means that a person only needs one copy of the mutated gene to develop the disease. If one parent has the gene, each child has a 50% chance of inheriting it.

It can be diagnosed by the combination of medical history, physical and neurological examination, brain imaging, and genetic testing. A doctor will ask questions about the person's symptoms, family history, and possible exposure to toxins or infections. The doctor will also check for signs of movement, cognitive, and psychiatric disorders, such as involuntary movements, impaired coordination, mood changes, and memory problems. Brain imaging, such as Computed Tomography (CT) scan or Magnetic Resonance Imaging (MRI), can show structural changes in the brain that are typical of Huntington's disease. Genetic testing can confirm the presence of the mutated Huntingtin (HTT) gene that causes the disease. However, genetic testing is not mandatory and should be done after consulting a

genetic counselor who can explain the benefits and risks of knowing one's gene status.

As brain imaging can help with the diagnosis of Huntington's disease by showing structural changes in brain that are typical of the condition. These changes include atrophy (shrinkage) of the caudate nucleus and the putamen, which are parts of the basal ganglia that control movement and cognition. The atrophy of these regions leads to enlargement of the frontal horns of the lateral ventricles, which are fluid-filled spaces in the brain. Brain imaging can also rule out other causes of symptoms that may mimic Huntington's disease.

CT scan and MRI are both accurate methods for diagnosing, but MRI has the advantage of having higher spatial and contrast resolution. Both techniques can show detailed images of the brain structure and reveal changes in the brain areas affected by Huntington's disease, such as the caudate nucleus and the putamen. These changes may not be visible in the early stages of the disease, but they become more evident as the disease progresses. This disease can be painful for many patients, but the pain may not be as severe or disabling as other symptoms of the disease.

The pain can affect the quality of life of Huntington's patients in various ways. Pain can cause physical discomfort, interfere with daily activities, reduce mobility and function, and increase the risk of complications such as falls and fractures. Pain can also affect the emotional and mental well-being of patients and their caregivers, causing distress, depression, anxiety, and isolation. However, pain may not be as severe or disabling as other symptoms of Huntington's disease, such as movement disorders, cognitive decline, and psychiatric problems. This may be because Huntington's disease affects the brain regions that are involved in sensing and responding to pain, leading to an abnormal perception and reaction to pain stimuli. Therefore, pain may pose a lower burden and have less impact on the quality of life of Huntington's patients than in the general population.

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