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Huntington's: Symptoms, Diagnosis

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Editorial Note

Huntington's disease is a neurodegenerative disorder characterized by uncontrolled jerking and writhing movements known as chorea, loss of thinking ability, and psychiatric problems. It is caused by a mutation of the huntingtin (HTT) gene, called a CAG trinucleotide repeat expansion because the segment of the gene containing the CAG portion of DNA repeats is longer than usual. The longer this repeat region is, the earlier the symptoms of the disease appear. They also tend to be more severe. Learn more about some of the symptoms of Huntington's disease below.

Symptoms

Behavioral changes

People with Huntington's disease not only experience physical changes, marked by a loss of movement control, but also cognitive and behavioral changes. The behavioral changes vary from patient to patient, but often include apathy, marked by a loss of motivation to start or finish activities. Patients may be irritable or depressed. They may lack inhibition and do or say things that one would normally find embarrassing.

Chorea

Chorea is the most common involuntary movement problem in Huntington's disease patients. It is characterized by involuntary jerking or writhing movements. It usually occurs during the early intermediate stage of the disease, several years after disease onset. Chorea diminishes at advanced stages of the disease, at which time dystonia (muscle spasms in the arms, head, or trunk) emerges.

Communication issues

Huntington's disease also affects a person's ability to communicate. A lot of patients with the disorder experience speech difficulties. Communication issues are more common in the later stages of the disease, although they also can be seen early on. Speech changes such as hoarseness in the voice, slurred words, the inability to control speech volume, and inappropriate pauses between words, are common in patients.

Delusions and hallucinations

Delusions and hallucinations are among the psychiatric symptoms of Huntington's disease. Delusions can be defined as false beliefs, while a hallucination is a sensory perception that is not real. Some environmental and behavioral adaptations can help manage delusions and hallucinations. If adaptions in day-to-day life are not sufficient, antipsychotic medications might be prescribed.

Feeding problems

Frequent complications associated with Huntington's disease include problems with eating and swallowing. Weight loss often is associated with the disease, so patients and their caregivers should seek professional advice to make sure they eat nutritious foods with a lot of calories that are easy to consume. A feeding tube may be an option for patients who are struggling with eating, drinking, and maintaining weight.

Other psychiatric issues

Huntington's disease damages the brain, leading directly to psychiatric problems. In addition, patients may experience depression and anxiety in reaction to their diagnosis and the disease itself. They can develop well-defined syndromes such as major depressive disorder or obsessive-compulsive disorder, as well as subtle changes in mood and personality including irritability or loss of interest. Most of these conditions can be treated with medication and behavioral interventions.

Sexual problems

Motor problems predominate in Huntington's disease, and it is common for patients to encounter issues with sexuality. Adults with this disease commonly face a loss of interest in sex. The behavioral changes that occur as part of the disease process can lead to a decreased libido. Less frequently, Huntington's disease may also cause an increased sex drive and inappropriate sexual behavior.

Neurological tests

A neurologist will interview the individual intensively to obtain the medical history and rule out other conditions. Tests of neurological and physical functions may review reflexes, balance, movement, muscle tone, hearing, walking, and mental status.

Genetic tests

The most effective and accurate method of testing for HD—called the direct genetic test—counts the number of CAG repeats in the HD gene, using DNA taken from a blood sample. The presence of 36 or more repeats supports a diagnosis of HD. A test result of 26 or fewer repeats rules out HD. A small percentage of individuals will have repeats in a borderline range.

Diagnostic imaging

In some cases, especially if a person's family history and genetic testing are inconclusive, the physician may recommend brain imaging, such as computed tomography (CT) or, more likely, magnetic resonance imaging (MRI). As the disease progresses, these scans

typically reveal shrinkage of the striatum and parts of the cortex, and enlargement of fluid-filled cavities within the brain called ventricles.

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