

What doctors want patients to know about Huntington's disease

AUG 1, 2025



Sara Berg, MS

News Editor

Huntington's disease, a rare but devastating genetic disorder, affects thousands of families each year, gradually robbing people of their movement, cognitive function and emotional stability. Often described as having [amyotrophic lateral sclerosis](#), [Parkinson's disease](#) and [Alzheimer's disease](#) simultaneously, this progressive illness carries a heavy emotional and physical toll, with symptoms typically emerging in adulthood. Knowing what to expect and how to prepare is key.

The AMA's What Doctors Want Patients to Know™ series gives physicians a platform to share what they want patients to understand about today's health care headlines.

In this installment, [Lisa Toran, MD](#), a neurologist who specializes in epilepsy at Confluence Health in Wenatchee, Washington, took time to discuss what patients need to know about Huntington's disease.

[Confluence Health](#) is part of the [AMA Health System Member Program](#), which provides enterprise solutions to equip leadership, physicians and care teams with resources to help drive the future of medicine.

It's a rare genetic disorder

"[Huntington's disease](#) is a genetic neurodegenerative disorder that causes progressive involuntary abnormal movements, dementia and psychiatric symptoms," said Dr. Toran, noting that about 41,000 Americans today are symptomatic and more than 200,000 are at risk of inheriting the disease. "The most common type of this disease is adult-onset

Huntington's disease, with symptoms usually starting in a person's 30s or 40s."



Lisa Toran, MD

One symptom experienced in adult-onset Huntington's disease is chorea, which causes involuntary, irregular or unpredictable muscle movements in the arms, legs and facial muscles, she explained, adding that it also includes "cognitive decline and psychiatric symptoms."

"The other less common type is juvenile Huntington's disease, which begins in childhood or adolescence, with symptoms including rigidity, bradykinesia, cognitive decline and often seizures," said Dr. Toran. "It progresses more rapidly than adult-onset Huntington's disease."

Additionally, "Huntington's disease is relatively rare," she said. "The prevalence of Huntington's disease globally is approximately three to seven per 100,000 people and does occur more commonly in people of European ancestry."

Huntington's disease is hereditary

"Huntington's disease is inherited from one parent to a child," said Dr. Toran. "It has an autosomal dominant pattern of inheritance, so only one copy of the altered gene in each cell will cause Huntington's disease."

“Individuals with a family history of Huntington’s are at risk of developing the disease,” she said. “The autosomal dominant pattern of inheritance means that each child of a parent who has Huntington’s disease has a 50% chance of inheriting the disease.”

The cause is a repeat gene

Huntington’s disease “is caused by an expanded CAG—cytosine, adenine and guanine—trinucleotide repeat in the huntingtin gene on chromosome 4, which leads to the production of a mutation huntingtin protein,” Dr. Toran said. “This abnormal protein causes progressive breakdown or degeneration in some areas of the brain.”

“The number of CAG repeats correlates with the onset and severity of the disease, with larger expansions usually leading to earlier onset and more severe symptoms,” she said.

Genetic testing is key

“Huntington’s disease is diagnosed through a combination of clinical evaluation, family history and genetic testing,” said Dr. Toran. “In the clinical evaluation, a physician will evaluate for typical symptoms such as chorea, cognitive decline and psychiatric disturbances.”

“Most patients will present with a family history of Huntington’s disease,” she added. But “the definitive diagnosis of Huntington’s disease is made by genetic testing to identify an expansion of 36 or more CAG trinucleotide repeats in the huntingtin gene on chromosome 4.”

“With genetic testing you can find out if you have Huntington’s disease before clinical symptoms appear,” Dr. Toran said. “If there are more than 40 CAG repeats in the huntingtin gene then it is predictable that the individual will go on to have symptoms of Huntington’s disease.”

“It is important to personally consider if one wants to know this information ahead of time before genetic testing is sent,” she said, noting that “it can be helpful for personal decisions regarding reproduction, financial matters and career planning.”

Dr. Toran added that “if you have Huntington’s disease and are planning to have children, then your risk of having a child with Huntington’s disease is 50% for each child.” She noted that “it is possible to do in vitro fertilization with genetic testing to make sure you don’t pass Huntington’s disease down to your future children.

“Alternatively, if a child is naturally conceived, prenatal diagnosis ... is possible,” Dr. Toran added. “A procedure, either chorionic villus sampling, performed at 10–13 weeks pregnant, or amniocentesis—performed at 15–20 weeks pregnant—can determine if the baby has inherited Huntington’s disease.”

Symptoms fall into three categories

“First there are motor symptoms, which can involve chorea, or involuntary jerking movements, dystonia—sustained muscle contractions causing twisting or repetitive movements or abnormal postures—bradykinesia or slowness, rigidity, slurred speech and difficulty with eye movements,” said Dr. Toran.

“There are also cognitive symptoms, such as impaired memory, planning, judgment and decision making,” she said. “And lastly there are psychiatric symptoms, which include [depression](#), personality changes and psychosis.”

“The first symptoms that appear in Huntington’s disease are usually subtle changes in mood, coordination and cognition, such as clumsiness, mild depression, irritability and difficulty concentrating,” Dr. Toran noted.

Chorea worsens as disease progresses

With Huntington’s disease, chorea “involves abnormal involuntary movements that are nonrepetitive and arrhythmic,” said Dr. Toran, “and can affect the entire body or just parts of the body such as the limbs, face or trunk.”

“Typically, these movements can start off mild and worsen as the disease progresses,” she said. “These movements are typically present continuously while awake and can be exacerbated by stress.”

Complications develop later

“Difficulty swallowing and speaking are common in later stages, leading to nutritional deficiencies and aspiration pneumonia,” said Dr. Toran. “[Pneumonia](#), due to aspiration, is a leading cause of death in this disease.”

Additionally, with Huntington’s disease, “weight loss is common,” she said. Meanwhile, “there is an increased risk of falls and injury from falls, such as bone fractures.”

Then, “as the disease becomes more advanced, patients develop cognitive decline and then dementia,” Dr. Toran said.

There is no cure

While “there is no cure for Huntington’s disease, there are a few medications that can help improve symptoms,” said Dr. Toran. “For example, tetrabenazine is recommended as a first-line pharmacologic treatment for Huntington’s disease chorea.”

Also, “it is important to monitor and treat psychiatric disturbances such as depression, psychosis and aggression, which are commonly managed with psychotropic drugs such as antidepressants, antipsychotics and mood stabilizers,” she said. “Without a cure, supportive care is essential, with a focus on optimizing the patient’s quality of life.”

“This includes nutritional support to prevent weight loss and aspiration, physical therapy to maintain mobility and [prevent falls](#), occupational therapy to assist with daily living activities, and speech therapy to address dysphagia and communication difficulties,” Dr. Toran added.

Medications do have side effects

“There are several medications that can help with chorea in Huntington’s disease, but they do come with a risk of possible side effects, so should be reserved for chorea that is at least moderate or severe and interfering with quality of life,” said Dr. Toran. For example,

“tetraabenazine can cause depression, suicidality and parkinsonism.

“Deutetabenazine and valbenazine are similar medications but are more convenient as they are dosed less frequently and may have fewer side effects,” she added. “Antipsychotic medications, such as risperidone, olanzapine and aripiprazole, are an alternative option that can help with chorea as well as psychiatric symptoms. Side effects with these medications in some patients include parkinsonism, weight gain and sedation.”

“Benzodiazepines, such as clonazepam or lorazepam, can be used in the short term to decrease severe episodes of chorea, but they are not useful or well tolerated as long-term therapy,” Dr. Toran said.

Care requires a multidisciplinary team

“Since Huntington’s disease is a complex, multifaceted disease, patients should have a multidisciplinary team to help them to address the physical and psychologic needs that arise in this disease,” Dr. Toran said. “Ideally the team would include a neurologist, psychiatrist, geneticist, social worker, physical therapist, occupation therapist, speech pathologist and nutritionist. ?

“It is important to have a primary care doctor to continue to monitor any other health issues that arise,” she added.

It progresses to severe disability

“Progression of Huntington’s disease leads to severe disability, with individuals eventually becoming dependent on others for care and losing the ability to speak, walk and swallow,” Dr. Toran said. “The disease itself isn’t fatal but makes everyday activities more difficult over time, such as walking and swallowing, and leads to complications that can cause death, such as aspiration pneumonia.”

“The median survival time after onset is approximately 15 to 20 years. The actual survival time varies from person to person,” she said.

Take care of yourself

While there is no cure for Huntington's disease, there are things you can do to take care of yourself and improve your quality of life. For example, "you can get regular exercise and eat healthy foods," said Dr. Toran. "Aerobic exercises, strength training, walking, balance training and fine motor exercises can improve motor functions and quality of life."

If you are having weight loss, you may need to increase how many calories you eat in a day, as the movements with chorea can burn more calories," she said. Meanwhile, "alcohol and tobacco use should be avoided."

Additionally, "support groups can help for social and emotional support," Dr. Toran said, noting "you can research caregiver options either in the home or in a nursing home in case that is needed in the future as well."

Care givers must take care too

"Caring for a family member with Huntington's disease can be challenging," said Dr. Toran. "Though it can be rewarding to help a loved one in need, caregiver burnout is especially prevalent when caring for someone with Huntington's disease because of the cognitive impairment and behavioral changes that can occur in the disease."

"Isolation and loneliness as a caregiver for someone with Huntington's disease is a very common experience. For any caregiver it is extremely important to remember that caring for oneself is important too," she said. "Focusing on maintaining external support networks and relationships or hobbies can be helpful."

Additionally, "reaching out for support networks such as through the Huntington's Disease Society of America or local social workers or case managers to investigate local care resources can be helpful tools for caregivers," Dr. Toran said.

Prepare for the future

“You can also plan for the future,” said Dr. Toran. “You should choose an individual who can help or make medical decisions for you if you have significant cognitive decline and are no longer able to make your own decisions.”

Additionally, “you should create a medical advanced directive and communicate this information with this chosen person so they know what your wishes are in case you become sick and are not able to make your own decisions,” she said. “These two actions should be taken early in the course of the disease.”