

REWRITE FOR PATIENTS: About Huntington's Disease

Huntington's is a disease that changes the brain, and these changes affect how you move, think, and feel. For example, when you have Huntington's, your body may move without your say-so, in a jerky fashion (called "chorea"), and it can make it hard to walk, speak, swallow, or even think. It was named for George Huntington, a doctor who in the late 1800's noticed these symptoms in a group of his patients. Over time, these symptoms get worse.

You may know other people in your family who have Huntington's because it is a disease that is passed on from parent to child, in a gene. Every child of a person with Huntington's has a 50% chance of developing the disease. Genes are the tiny sets of instructions that help make our bodies turn out the way they do. For example, your genes make your eyes and hair whatever colors they are. Some genes make proteins, which are the materials that help form every part of our bodies—from our muscles and bones to the acid in our stomach that helps break down the food we eat into fuel.

Huntington's is caused by a single gene in your body, the *HTT* gene ("HTT" stands for huntingtin), that did not form the usual way. The standard Huntington gene makes the HTT protein, which has several jobs. These jobs include helping signals travel through your brain between your nerve cells (called "neurons"), which allow you to walk or talk. So when you think about walking, the neurons send messages to tell your leg muscles to move.

In people who have Huntington's, this one misformed gene makes an unusual version of the HTT protein called mHTT (the "m" stands for mutant—meaning it's not made the usual way). Because your body has this one mutant HTT gene, it makes the mHTT protein. Having mHTT build up in your body is bad for it, because mHTT damages your neurons over time, so they don't send signals the right way. This is why Huntington's changes how you move, think, and feel.

Although researchers are working hard to discover treatments that work, currently there are no medicines that can slow, stop, or reverse Huntington's disease, which eventually leads to death. Instead, the goals of treatment are to lessen how much you are affected by the symptoms of Huntington's and to improve your daily life so that you can do as much as possible.

ORIGINAL, PROVIDED COPY: Information About Huntington's Disease

HD is a progressive and fatal monogenic neurodegenerative disease of the whole brain. HD has devastating impact due to relentless deterioration in motor, cognitive and neuropsychiatric function.

Everyone who develops HD has the causative, autosomal-dominant CAG-expansion mutation in HTT, though CAG-repeat length varies and SNP profiles in HTT differ.

HD is currently understood to be caused primarily by chronic expression of toxic mHTT. This leads to a well-characterized pathology cascade, rather than reduced levels of normal Huntingtin protein.

Toxic mHTT build-up throughout the brain leads to the progressive degeneration of neurons with devastating effects on the lives of people impacted by HD.

Available treatments cannot slow, stop, or reverse the course of HD. The goals of disease management in people with HD are limited to reducing the burden of symptoms, maximizing function, and optimizing quality of life via a multidisciplinary approach.