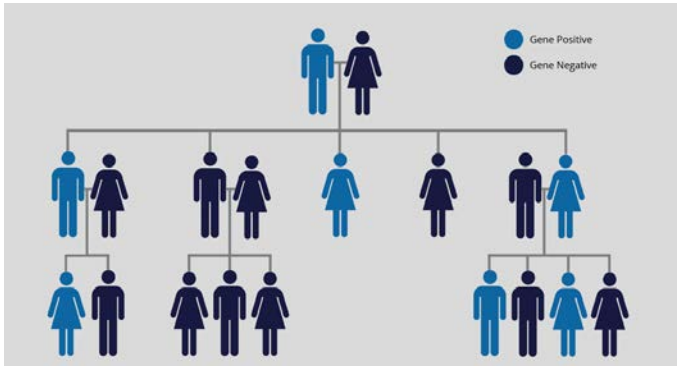
From the Experts: HD 101

What is Huntington's Disease (HD)?

HD is a genetic neurodegenerative disorder. That might be confusing so let's break it down. Genetic means inherited (one generation to the next). Neuro relates to brain. Degenerative means to get worse over time and disorder means illness or disease. So, HD is a brain disease that gets passed down from one family member to the other and worsens over time.

What Causes HD?



Our bodies are made up of millions of cells. The cells contain genes (there are about 25,000 of them). The genes are instructions for cells to make something called proteins. Proteins allow the cells to grow and have different functions. We all have a gene that makes a protein call Huntingtin (it sounds similar but this is different than Huntington). We know our bodies need this good protein to survive. Sometimes there is a change, called a mutation, which causes this gene to act differently. Instead of producing a protein our body needs, this gene produces a protein that is bad for cells (Huntington).

Each of us has 23 pairs of chromosomes. We get one set of chromosomes from each parent. Each person with HD has one chromosome that is normal and one that carries the HD gene. The parent with HD can pass on either the normal gene OR the gene that is positive for HD. So it is only a matter of chance who will have the gene for HD and who will not. Each child of a person with HD has a 50% chance of inheriting HD.

A person will be positive for HD if they have inherited the "mutated" gene from the parent with HD. A person will be negative for HD if they inherited the normal gene from the parent with HD. If a person does not inherit the gene, then they will never get HD. And if they don't have HD, they can't pass it on to their children.

In the picture above, the top row shows a father in light blue who has the HD gene (this is called being gene positive) and a mother in dark blue who is gene negative. The next row shows they have had five children. You can tell which are their children because there is a

vertical line directly to the child. So, in this case this couple had two boys and three girls. Starting from the left side the first male is gene positive. He then had two children, a boy and a girl. The girl on the third row is gene positive. The boy is gene negative. The next child on line two is a male who is gene negative. He had three children. But since he is gene negative, and his partner doesn't have HD all three of their children would be gene negative (none of them are at-risk for HD). The third child on line two is a female who is gene positive. In this case she does not have any children. The fourth child is also a female. She is in dark blue and is gene negative. She will not get HD and if she has children, they will not be at-risk for HD. The fifth child is also a female. She is gene positive. She had four children, two boys and two girls. In her case one boy and one girl are gene positive and the other boy and girl are gene negative.

Symptoms of HD

HD can cause three types of symptoms. We teach people to remember the symptoms with the letter M. The three symptoms are:

- Mood
- Memory
- Movements

People with HD can have one, two, or all three of the symptoms of HD. People with HD in the same family can even have different symptoms from each other.

When people have mood changes, their personality can change. They may become depressed or become anxious. They can become irritable, or they can become withdrawn from others. They can become upset if their routine changes. They can have mood swings, where they feel happy one minute and then very sad the next. It is important to recognize that they might not know they are having mood changes.

When people have memory difficulties they might have difficulty concentrating. They may have a hard time remembering things. They have a hard time planning or thinking ahead. They can forget how to do things that were once easy for them to do. They might have difficulty waiting. They might not know they are having trouble remembering things.

People with HD sometimes have involuntary movements (movements they can't control) called chorea or dystonia. These movements might look like the person with HD is drunk. They might have difficulty talking or swallowing. They might drop things. They might have problems with their balance. They might not know they have involuntary movements.

When Does HD Start?

About 30,000 (1 in every 10,000) Americans have HD and another 300,000 other people are "at risk". These 300,000 people include children and grandchildren of people who have HD. People who will get HD have the gene from birth but normally the symptoms don't start until people are in their 30's or 40's. There have been cases when people start to get the symptoms of HD when they are much older. There is also a form of HD which affects young kids. It is called Juvenile-onset HD (or JoHD) or Pediatric HD (PHD). These are less common than adult onset and typically passed down from a father who has HD.

How Long Does HD Last?

Right now, every person who inherits the HD gene will eventually develop the disease. Once the symptoms start, the disease lasts about 25 years. This is difficult because HD develops gradually and slowly, and many times people don't know exactly when it starts.

How Do You Treat HD?

Currently, there is no cure for HD. People don't die of HD, but they get very sick and weak and die from infections that their bodies can't fight off. There are many medications that can help to make some of the symptoms better. But a person with HD needs to work with a healthcare team in order to get treatment. Sometimes if people with HD don't admit they are sick, they won't want to go for treatment. One way to cope with this is to teach family members how to "deal" with behaviors" people with HD may have. We will talk more about dealing with behavior problems in an upcoming article.

To schedule an appointment with Dr. Bonnie Hennig-Trestman, call **540-521-8306**. Learn more at **CarilionClinic.org/huntingtons-disease**.