Huntington's Disease: Causes And Treatment

Huntington’s Disease was first discovered in 1872, in Long Island by American Doctor, George Huntington (3). Before the discovery of the disease, it was believed that the symptoms of the disease were signs of the devil possessing one’s body. Huntington’s Disease is divided into two subtypes: Adult-onset Huntington's disease, in which people develop symptoms in their mid-40s and 50s, and Juvenile Huntington's disease, which is a rare form where children and teenagers are affected by it.

Huntington’s Disease is a fatal genetic disorder that is autosomal dominant. This means that only one copy of this gene is needed to develop this gene. A child inherits two copies of every gene, with the exception of genes on sex chromosomes, one copy from each parent. This means that a parent has the possibility of passing along the defective gene or a healthy gene. This, unfortunately, means that each child in the family has a 50% chance of inheriting the defective gene that causes Huntington’s Disease (5).

It is estimated that one in every 10,000 persons, around 30,000 people in the US have Huntington's disease. Juvenile Huntington's occurs in approximately 16 percent of all cases (6). It also affects an estimated 3 to 7 per 100,000 people of European ancestry. Fortunately or unfortunately, Huntington’s disease has not been shown to be prevalent in any particular population. All races, ethnic groups, and both sexes are affected by it.

Huntington’s Disease is caused by an inherited defect in a gene. This gene is called the HTT gene. This gene is located on the short (p) arm of chromosome 4 at position 16.3. The HTT gene function is to provide instructions to make the huntingtin protein. The function of this protein is not yet known. Genetic defects can be caused by mutations. Mutations in the HTT gene involves a DNA segment known as a CAG trinucleotide repeat. Normally, the CAG triplet (glutamine) repeats around 10-35 times in a row, but for people with Huntington’s disease, the triplet is repeated 36 times or more (1). As it gets passed down from generation to generation, the size of the CAG trinucleotide may increase. The abnormally long protein is cut into smaller, toxic fragments that bind together and accumulate in neurons, disrupting their normal functions. The death of these neurons in certain areas of the brain is what leads to the signs and symptoms of the disease.

Unlike many other genetic disorders such as cystic fibrosis and achondroplasia, the symptoms of Huntington’s disease usually do not develop or show in babies or children. In most cases, symptoms start showing between ages 30 to 50 (2). Because of this, many people do not know until symptoms have developed that they have the disease unless they have been genetically tested. Symptoms of Huntington’s disease include movement, cognitive, and psychiatric disorders. Examples of movement disorders are involuntary jerking, rigidity or muscle contractures, abnormal eye movements, and impaired posture and balance. Cognitive disorders include lack of awareness of one's own behaviors and abilities, slowness in processing thoughts, and lack of impulse control (4). Psychiatric disorders include social withdrawal, insomnia, and frequent thoughts of death. People may also develop OCD and bipolar disorder as a result of this disease. Typically, the disease’s development can be divided into three stages: early, middle, and late. In the early stages, the symptoms are very minimal. People in

the early stages are able to complete their daily routines without a hassle and seem fine. In the middle stage, involuntary movements become more noticeable and may need assistance in completing their tasks. Finally, in the late stage, people with the disease are in need of complete care. The symptoms go as far as the inability to speak, swallow food, or walk. It is around this time where most people with the disease die, usually as a result of malnutrition, pneumonia, or heart failure.

Because of how limiting the disease is on people, they have to change their lifestyle to cope and survive. People diagnosed with Huntington's Disease can help to reduce the impact of their symptoms by staying socially, physically and mentally active. Movement issues are often managed by medications such as Xenazine, and with physical therapy (7). Non-slip mats and handrails are often helpful as they help to prevent falls. People with this disease require special diets with high-calorie foods because of their constant movement (7). At later stages, patients may have changes in control of the throat muscles can cause difficulties in swallowing food, so they transition to softer foods. In worse cases, people need to use tube feeders for nutrition consumption.

Unfortunately, as of now, there are no known cures for Huntington's disease. Drugs and physical therapy can only help manage some symptoms. The most people can do is take predictive genetic tests to see if their children will be born with the disease.