Huntington’s Disease: Symptoms And Stages

There are many diseases out in the world that are affecting people’s lives in different ways. They can either be infectious or non-infectious disease. A non-infectious disease would be Huntington’s disease which is inherited. It causes a severe breakdown of nerve cells of the brain. The parts of the brain that get damage are the basal ganglia, cerebral cortex the frontal and temporal lobes, ventricles, and caudate nuclei. This disease is most common in people with European ancestry and it’s is less common in people with Japanese, Chinese, and African ancestry. About 3 out of 7 per 100,000 people that have European genes are affected by Huntington's disease . A person can experience symptoms at the age of 30 to 50 years of age and they can also come at the early age of 20 which is referred to juvenile HD. According to statistics more than 30,000 Americans have Huntington’s disease. The reason that this disease is developed in a human is the mutation in the gene of a protein named huntingtin which causes cytosine, adenine, and guanine building blocks of DNA to replicate more times than it should. A child with a parent that has HD has a 50/50 possibility of inheriting the disease. Now a person who does not inherit HD can not get the disease and can’t pass it onto future generations.

George Huntington, American physician from Long Island, New York was the one to contribute to the explanation of HD which was named after him in 1872. Huntington wrote a paper called On Chorea which was later published in the Medical and Surgical Reporter of Philadelphia and became known for Huntington's Chorea. Huntington Chorea h evolved through the years and is even known today as Huntington's Disease. In the year of 1933 a research group found the gene of HD, because of this discovery it is possible to diagnose a person through the use of their blood and tissue samples.

Huntington’s disease occurs from the mutation in the gene that is located on chromosome 4. Every person has this gene and has the CAG repeat sequence. When someone has HD their CAG sequence has a high number of repeats which is a result of protein that has a long polyglutamine sequence. The regular huntingtin gene contains 17 to 20 repetitions of the code of its total of 3,100. The number of repeats that adults with HD have is more than 40 and for people who have juvenile HD usually have more than 60 CAG repeats. Depending if one has a big number of triplet repeats the more it is likely to develop HD at an early stage of life. A father passing the gene to his child might extend the gene even more, starting the disease at an early age. For those who contain 27 to 35 repeats don’t develop Huntington’s disease but there is a slight possibility that their children will get the disorder.

The symptoms of this disease varies for each person but HD does cause psychiatric, cognitive, and movement problems. Depression is an example of a psychiatric effect that Huntington's disease causes. It occurs because of the injury the brain goes through and changes in the functions of the brain. Other psychiatric disorders are obsessive-compulsive disorder, mania, and bipolar disorder. Some signs include feeling irritability, sadness, social withdraw, insomnia, fatigue, and frequent thoughts of suicide. For cognitive disorders people may experience difficulty organizing or focusing on tasks, people may tend to get stuck on a thought or on an action. When someone is talking they may forget what they just said and not be able to come up with a response as fast as a person who doesn’t have HD can. People also are not aware of their own behavior and are slow at processing thoughts and have difficulty learning new

information. Huntington’s disease affects ones movement ability which can be involuntary movement problems and damage involuntary movements. This includes involuntary jerking or writhing movements, muscle problems like rigidity, slow eye movements, difficulty with posture and balance, and having a hard time with speech and swallowing. Once a person starts having symptoms of Huntington’s disease the duration can be from ten to thirty years. According to the Huntington’s Disease Society of America “Huntington’s Disease manifests as a triad of motor, cognitive, and psychiatric symptoms which begin insidiously and progress over many years, until the death of the individual. The average length of survival after clinical diagnosis is typically 10-20 years, but some people have lived thirty or forty years. Late-stage HD may last up to a decade or more.” Voluntary movements are really impactful on an individual's ability to work, act, and communicate.

The symptoms for younger people who have Juvenile Huntington's disease are a little different than those of adults. Juvenile HD is not as common as adult HD itself. People have behavioral changes like losing academics or physical skills, a decrease in school performance, and behavioral problems. They also suffer in physical changes that include contracted muscles that affect their walking ability, changes in motor skills like their handwriting, seizures, and involuntary shaking. Weight loss is another factor that is affected because of HD When a young person has to go through these symptoms they may experience anger, sadness, and fear, leading to aggressiveness. Children who inherit Juvenile Huntington’s disease get their sequence repeats from their fathers however they can inherit it from their mothers even though it is rare. The life span for individuals with Juvenile HD is no more than 10 to 15 years after they begin experiencing symptoms. This affects young person daily life from attending school to having a job and even their chance of creating a family in fear of passing the gene of HD on to them.

There are five stages to Huntington’s disease progression. The first stage is considered the early stage, from when it starts it can last up to about eight years. During this stage the person has been diagnosed with HD but can function at home and in the workplace. They still have independence and can do activities own their own like managing their finances, home responsibilities, eating on their own, getting dressed, bathing, and so much more other things. Patients usually don’t experience movement problems but can experience cognitive symptoms as well as psychiatric symptoms. The second stage is the early intermediate stage. It can last up to three to thirteen years from the first day of HD onset. The individual can still function at their workplace but not to the same ability they first began with when they did not have HD. They are somewhat able to do daily life activities but have some difficulties which require them to have some help from others. However, at this stage, some may be incapable to work and need a lot of assistance in one certain activity but are fully capable to complete other activities. Involuntary movement in the body which is called Chorea becomes present in this stage. In the third stage the, late intermediate stage can last up to five and sixteen years. An individual can’t maintain house responsibilities or work any longer. They will also need significant help for finances, responsibilities in their home and activities they have in their daily lives. Their thinking ability weakens and some psychiatric symptoms become more prominent like anxiety, irritation, and they become more impulsive. Dementia is also is a disorder that can get this far into the disease. Not only does the psychiatric worsen but so does cognitive symptoms. The second to last stage is called the early advanced stage which lasts from nine to twenty-one years. They are no longer independent and can still stay living in their homes with the help of family members or with professional help. Facilities home can provide them better assistance in their needs and daily activities. They will require a lot help in all their activities and be aware of what

they have to do. The last stage, the fifth stage, is called the advanced stage. The duration for this stage is eleven to twenty-six years. They need 100% help from a nurse and Chorea gets toned down. Although Chorea lessens Parkinsonism intensifies which causes slowness, unusual posture, stiffness, and grinding of the teeth. There is a more probability of accidents like falling and as it states in Huntington's Disease News " Speech can become difficult at this stage and the patient may go through periods of confusion and scream. The ability to swallow also can worsen, and there can be extreme fluctuations in blood pressure and temperature."

People with Huntington's disease die fifteen to twenty years after being diagnosed and they die because of a complication of this disease. These life-threatening complications are pneumonia and heart disease. Huntington's disease patients have a bigger chance of choking causing respiratory problems, they can get gastrointestinal problems like cancer in the pancreas. As stated in the excerpt titled Complications of Huntington's Disease "As a result of these movements, the epiglottis a flap that acts as a valve in our throat, prevents food from entering the airway. People with HD often lack this coordination, and food will accidentally enter the respiratory tract, leading to choking. Moreover, when food particles manage to get into the trachea (the "wind pipe" leading to the lungs), instead of the esophagus (the "food pipe" leading to the stomach), the lungs can become infected and cause what is known as aspiration pneumonia." As stated this is one complication that leads to the death of a person with Huntington's disease. One can prevent this from happening by adjusting the diet of the patient to a make sure they have a more appropriate intake of the right food. A study that took place in the years of 1952 until 1979 in Victoria, Australia showed that 51% of patients that had HD died from getting pneumonia.

There is no cure for this that can backtrack or slow down the symptoms to this disease, however, there is the medication that helps with symptoms. Some of these medications are Tetrabenazine which is accepted by the Food and Drug Administration, treats involuntary jerking movements or chorea. Side effects do come with this medication and they are feeling sad, losing interest in relationships with others, either sleeping more or less than one should, feeling angry, and weight loss. With these side effects the patient should contact a doctor. Medication that controls movements and hallucinations are clonazepam, haloperidol, and clozapine. Depression is one of the symptoms that Huntington's disease cause, so to ease that there is fluoxetine (Prozac), sertraline (Zoloft), nortriptyline (Pamelor). Many patients with HD experience mood phases and to lessen that they can take mood- stabilizing drugs.

To help a patients speaking ability there is a choice of doing speech therapy as well as other types of therapy to help with their different necessities. Psychotherapy helps the individual to be able to control their aggressive behavior and they are demonstrated strategies to communicate with others. Speech therapy helps better their speech and exercises the muscles that have to do with the mouth and throat. For physical therapy the therapist helps the patient gain strength in their muscles which improves their ability to walk and their balance. They also provide them with techniques on posture and how to properly use a walker or a wheelchair. Lastly, occupational therapy can help the patient with HD, their family or caregivers. They teach them about the equipment they can use to help assist the patient.

There are six different types of ways a person with HD can get tested to see if they carry the gene for it. The first one is the neurological examination were a neurologist asks a series of questions and does tests to be able to examine their abilities. They test their movements, their senses, and their feelings. Another test is the neuropsychological testing that has to do with the

patients memory. Mentality, and reasoning. The next one will be the psychiatric evaluation which is in relation with behavior, emotions, and their thinking. Brain imaging and function is another form of testing that show the structure of the brain and how it is functioning. It can be an MRI or a CT scan. They will show how the brain has changed and how it has been affected by Huntington’s disease. There is genetic counseling to test out their genetics if they have the defective gene. Lastly there is predictive genetic testing for those whose family members have had this disease but they themselves don’t show any signs, symptoms of having HD. Some people prefer to do this type of testing so they know what is to come and know whether they have the disease so they are able to decide if they want to have children and start a family knowing the risks there could be.