Huntington's disease: statistics, causes, diagnosis

Health & Medicine, Disease

Imagine someone being born with a genetic disease that has no cure and they only have a certain amount of years to live before they die. Huntington’s Disease is defined as being an inherited disease in which causes the body nerve cells to continuously breakdown (Rarediseases. info. nih. gov, 2019). George Huntington, an American physician wrote the first description of this disease called Huntington’s Disease in the year of 1872. He proceeded to call it “ hereditary chorea” to understand its features (Ninds. nih. gov, 2019). The word “ Chorea” derives from the Greek word dance which describes uncontrollable dance-like movements with people who suffer from HD (Ninds. nih. gov, 2019). Currently, there are more than 30, 000 individuals who are diagnosed with HD. The rate of new infections has been reported due to ethnicity and geographical location, confirming that depending on the demographic differences influence the number of individuals with HD (Anon, 2019). In Europe, it was estimated that an average of 4-7 persons per 100, 000 were affected with the disease (Anon, 2019). Canadian studies suggested 2. 4-8. 4 persons per 100, 000 had HD. In the United States, it was estimated that 4. 1 – 5. 2 persons per 100, 000 were living with the disorder (Anon, 2019).

The age range for this disease where signs and symptoms start to develop is between 30-50 years. The life expectancy for someone with this condition ranges between 15-20 years (Rarediseases. info. nih. gov, 2019). Therefore, the likelihood of an individual inherited with Huntington’s Disease and exceeding to live more than twenty-years is not possible due to studies shown.

According to (Ninds. nih. gov, 2019), HD is passed from the parent to the child through a gene mutation. Genetic diseases happen when the DNA change messes up the instructions in a gene. People with HD most times have a parent who is affected. The history of that person’s family can sometimes appear negative for reasons even though that parent may carry or carries a mutated HTT gene (Rarediseases. info. nih. gov, 2019). This gene mutation is dominant, which means that any child who inherits the Huntington’s Disease will eventually develop the disease (Alzheimer’s Disease and Dementia, 2019). This is said to be a trickier question than people may think (Anon, 2019). The risk is pretty simple to find out for a person with HD. In the likelihood that a parent does not carry the disease, means none of the children will either. Parents cannot pass on the gene that causes the disease because they do not have it (Anon, 2019). For example, if an individual friend great aunt had HD, but their grandparents did not, then they can say they could not have the disease. This means, if the grandparents don’t have the disease, then they do not have it (Anon, 2019). However, an individual gets HD because they happen to get a broken copy of a specific gene whether coming from the mother or father.

Usually, if one parent has the disease there is a 50% chance of each child getting the disease (Anon, 2019). The percentage (50%) derives from one parent carrying the disease and others not carrying. Now, everyone is born with two copies of each gene. Unfortunately, the child does not get to choose which copy they want from either parent. Furthermore, if the mother does not have HD and the father has HD. The father has two good copies of his gene which results in the child getting a good copy. However, if the mother

has one good copy and one broken copy, the child will get one of them randomly, so their chances of getting the broken copy are one in two or 50% (Anon, 2019). The great news is that if the child does not get the broken copy then they cannot pass on the disease to their children. Now, if the child happens to get the broken gene, the scientist has found a way to make sure that children do not get it by performing a procedure called preimplantation genetic diagnosis (PGD) (Anon, 2019). This procedure is done by scientists fertilizing an egg outside of the mother such as in vitro fertilization (IVF). Scientists wait until the embryo grows a little, in which they can secure a small part of it to test for the broken gene (Anon, 2019). If the embryo has the broken gene, they would not implant it back into the mother, therefore not having a child with the disease.

Genetic diseases often happen when a DNA change messes up the instructions in a gene (Anon, 2019). Some diseases are not as dominant as HD. According to (Anon, 2019), many genetic diseases result from both copies of a gene that are lost and broken. However, HD is different because one broken gene is enough to cause the disease in a person. The body is made up of genes which has instructions for a specific protein (Anon, 2019). The proteins in the body each have a specific job in the cell. For example, if someone carries a cancer gene then that gene has instructions for making a cancer protein. Similarly, this is how it is when someone has the gene of HD. Sometimes the proteins in a person’s body stops working and may gain another function. With HD, a part in this gene continuously repeats itself causing a longer form of the huntingtin’s disease to be made (Anon, 2019). Once this HD protein continues to get longer it causes the other proteins to

stick together and stops their function. Eventually, the cells in a person’s body dies because so many things are not functioning. Therefore, gaining a longer protein of HD stops everything from working (Anon, 2019). In further, not everyone with HD has the same length of proteins which makes the HD complicated.

Huntington’s Disease has gray areas that make it possible for a child to get the HD even if their parents do not have it (Anon, 2019). This refers back to the different lengths of the huntingtin’s protein which are safe and others that cause the child to get HD. Each person has different lengths because each person is different (Anon, 2019). A mother may have an intermediate length of protein and has the right genetic background in which she can deal with her cells (Anon, 2019). However, her daughter is passed with those cells from her mother but cannot deal with the proteins being that long. In certain cases, a healthy parent with slightly longer proteins passes the gene to their child and may become longer resulting in the child to inherit HD (Anon, 2019). If the person undergoes a genetic test to know if they have the gene, there’s uncertainty whether they will get it (Anon, 2019).

A diagnosis of HD is based on different findings from psychological, genetic, and neurological testing (Ninds. nih. gov, 2019). In the neurologist testing, they begin by interviewing the patient’s medical history and look through all the conditions. During this test, the focus is on physical functions such as balance, movements, hearing, walking, mental status, and muscle tone (Ninds. nih. gov, 2019). The patient undergoes several laboratory tests in which individuals with HD can seek other health care professionals such as,

speech pathologists, genetic counselors, psychiatrists, and clinical neuropsychologists (Ninds. nih. gov, 2019). The tool used to diagnose HD in family history is called a pedigree or genealogy. The most effective and accurate method of testing for HD is genetic testing. This test is performed by using DNA taken from a blood sample from the patient (Ninds. nih. gov, 2019). Before genetic testing, clinical also use a method call linkage testing. This testing requires a DNA sample from a closely affected relative, a parent, to identify markers close to the HD gene (Ninds. nih. gov, 2019). In some cases, if the patient’s family genetic testing and family history are inconclusive, then diagnostic imaging (brain imaging, magnetic resonance imaging, and computed tomography) is recommended (Ninds. nih. gov, 2019).