

# Impacts of huntington's disease (hd)



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## **Abstract**

Huntington's disease has been in existence for many years. People used to mistake it for other conditions such as for Parkinson's disease or possession by the evil spirit. As a result, an incorrect measure of dealing with the disease was used. A scientific breakthrough, for instance, the discovery of the gene that is associated with the disease formed a milestone upon establishing the correct measures to take. It also allowed early diagnosis so that people can be able to receive better care. The prevalence of the disease is low in many areas, but a particular region in Venezuela has recorded a substantial number of people suffering from the disease. Advancement in technology has allowed for pretest in this area, and since it was identified to be a hereditary disease, then people are advised how they could be able to control the disease. One control measure that has been taken into account, includes using odd contraceptives to control the population in the area. Apart from the stated objectives, the paper will explore the impacts which Huntington's Disease has had and continues effecting on the human population.

Huntington's disease (HD) refers to an autosomal disorder which leads to the progressive damage of the brain in late adulthood and subsequent brain atrophy. The area of the brain affected is responsible for a person's body movements thus the disease plays a crucial role in the movement. The degeneration of the brain results to movement problems for instance chorea, dystonia, and incoordination's among others. Huntington's disease was first discovered as an inherited disorder by a twenty-two year old American doctor in the year 1872. George Huntington was the man responsible for the

discovery of the disease, and it was later named after him. In the first paper he wrote about the infamous disease, he referred to it as Chorea.

Huntington's paper was published in the *Medical and Surgical Reporter* in the same year, and the disease at that time was referred to as Huntington's Chorea. The word "Chorea" was derived from the Greek and Latin word which means a group of dances or the chorus. Chorea was given as a name to refer to a group of disorders without known cures at the time that were discovered during the middle ages. Twitching and involuntary muscle jerks are characteristics of the Huntington's Disorder, and these kinds of movements caused havoc on those who were present during prior centuries. For instance, during the pre-medically advanced days, people with Chorea were often believed that the evil spirits possessed them. During the time, one of the witches thought to be cursed with the disease was executed in the Salem, Massachusetts during the 17<sup>th</sup> century and she ended up having the disease. Nowadays, the name "Huntington's Disease" is used commonly as compared to the Huntington's Chorea. During those early days, people rarely understood the disease, and thus most of them died even before the significant signs of the presence of HD begun to show up. Thus, the discovery of the gene in the year 1993 became a major milestone in medical history for humans. The scientists, with perseverance and patience, worked long and tedious hours to figure out the root cause and try to find a cure so that the disease could be treated. Till this day, there has not been any sign for discoveries of a cure for the disease. In this context, we explore how Huntington's Disease has impacted the advancement of the human population and whether there is a possibility for a cure to be discovered.

Huntington's disease is a Neurodegenerative disease in which abnormalities in the brain can be detected when a person undergoes an MRI test even one or two decades before the clinical diagnosis. The detection of the genes allows for trials before the manifestation of the symptoms so that an intervention plan can be considered before the major symptoms begin to show up. The symptoms of HD usually begin to show up in between the ages of 30-50, but in some of the cases, they can manifest themselves in the first two years after birth and the late 80s. The symptoms are characterized by the involuntary body movements, for instance, the uncontrolled movement of the leg, arm, face, head or the upper body. HD, in most cases, leads to lower reasoning and thinking skills, and this leads to effects such as lower concentration, memory loss, poor judgment, and lack the ability of planning and organizing everything. Ultimately, the disease alters the brain in such a manner that the mood of the person is drastically affected in a negative measure. This usually results in anxiety, depression, and uncharacteristic irritability and anger. Another primary symptom of HD involves the person becomeong obsessed with one essential subject. Thus it is common to find someone with the disease constantly stating the same phrase or repeating the same activity over and over again. The discovery of the gene that makes one vulnerable to acquire the disease has been a breakthrough discovery, and those who qualify, can now get tested to confirm if they carry the gene for the Huntington protein. In this manner, if the tests confirm positive, they can begin to get professional help before the symptoms advance to worse conditions.

Huntington's disease is an inherited neurological disorder which results from the defect in a single gene in the human cell. Although the discovery of the gene led to a breakthrough that allowed early detection of the disease, the cure or the appropriate treatment has never been discovered since then. In most cases, the humans are affected because their cell's mutation process replicates the gene in the brain and the brain offers some of the favorable conditions that allow its survival (Lopes et al.,). Most animals do not have the gene that causes the disease in their cells, because of different mutations and evolutions. Thus, they cannot be affected by the disease. This is especially harmful because, unless special modifications are done on the bodies of the test animal subject, the scientist cannot use them to run the tests. Scientists have established that, as the genes become inherited from a line of offspring, those genes reproduce themselves and become defective with every subsequent generation. In this case, the ultimate sufferers of the disease usually experience severe symptoms of the disease when compared to the parents they inherited the disease from. In most of the cases, laboratory testing with animals usually fell into a dead end, unless the mice had been modified. The mice are not able to replicate most of the symptoms associated with the brains of humans. Use of modified primates has been the current approach since primates have a closer resemblance to the brain of a human, in fact being the most closely related. The fact that animals are not as impactfully affected by the disease compared to humans makes it hard to proceed with a successful test that could lead to the discovery of the cure.

The prevalence of Huntington's diseases amongst humans varies worldwide with the highest figures recorded in the European population. The African

population, overall, have had a however lower prevalence of possessing the disease. Being a genetic disease, those who inherit the disease tend to have an abnormal dominant allele which interferes with the functioning of their nerve cells. This slowly erodes the control they have over their minds and bodies and subsequently leads to death. Most people who encountered the disease have been known to consider committing suicide due to the severity of the symptoms. There is a meager prevalence rate in many parts of the world. However, a fishing village in Venezuela has a very high prevalence rate (Chao). This part of the world contains many individuals with HD compared to any other part of the world. More than half of the population contains the gene that leads to the development of the disease at a later stage. The study, however, shows that the population affected is from an extended nuclear family in the region and thus the high prevalence. In the history of human population, people who were believed to have this disease were considered to have been possessed by the evil spirits, and thus an act of exorcism was performed on them to remove the evil spirit. In most cases, the acts committed led many to death rather than helping to live longer. The fact that HD does not have a high prevalence rate amongst human beings means that there were no considerable decreases in population when people affected by the disease lost their lives to it. However, in a population where the disease prevalence is very high, such as in Venezuela, there is a severe impact on the population (Chao).

Although HD has been in existence from an immemorial time, it has been until the 20<sup>th</sup> century when a big breakthrough in the disease begun with the help of advanced technology. Understanding the disease changed the

course of history when George Huntington discovered the gene responsible for the disease. This allowed the scientists and other medics to be able to study the progression of the disease, the signs, and symptoms, how the disease was spread and how it could be continued. Another breakthrough was through observation when it was noted that there was a high prevalence of people who have dementia and other HD associated symptoms in a small region in Venezuela (Chao). The symptoms of the diseases were running amongst families, not just individuals. This discovery led to scientist linking HD to gene inheritance of the autosomal genes, thus attempting to trace family members with the disease taking priority. The study of the Venezuelan families who suffered from the diseases exposed the precise causal end using the genetic linkage analysis method. At the same time, various scientific discoveries regarding how the disorder even began in cells took place. Also, scientists developed a quench to come up with a treatment and a cure plan in which they began using animal testing — this involved use of the transgenic mouse that was developed in the year 1996 (Pandey, Mritunjay and Usha Rajamma). This test allowed for a large-scale experiment. These experiments allowed the scientists to have a test that would bring fast results since the test subjects had a shorter lifespan compared to humans and higher metabolic rates thus speeding up the process of obtaining results. Extensive research on the field allowed scientists to develop potential drug treatments, reproduce the gene to use it for testing purposes, and develop the care methods. Also, research allowed scientists to change the disease's name from Huntington's Chorea to Huntington's Disease, since not all patients developed chorea. Use of the current technological advancements has fueled research on these fields, and <https://assignbuster.com/impacts-of-huntingtons-disease-hd/>

various genomes have been discovered that further stir scientists toward developing the cure for the disease. One of the most significant breakthroughs over the past years has been the development of antisense oligonucleotides (ASOs) which are the DNA molecules used to bind the HD causing RNA and, in the process, degrades them. The molecules are injected in the patient via the lumbar puncture with the hope that they will penetrate the area of the brain and reach basal ganglia which is the most affected area of the brain by the HD (WEN-JUAN HUANG). The result has gained a tremendous achievement and was presented in the previous scientific conferences.

Currently, there haven't been any cures found for HD, and no medical treatment has been developed that has been able to stop the progression of the disease. However, understanding the genetic causes of the disease came to light, and how it is spread especially through the genetic makeup of people who were related, allowed people to make informed decisions when they wanted to make reproduction choices(Kumar). In the current technological world, diagnosis for the disease is becoming more affordable to many and thus giving them a chance to identify their causative genes before the onset of the disease. If a person discovers that they do in fact carry the dominant allele for the gene, then making reproductive choices with consideration of the imminent disease become essential. For instance, in Venezuela where the prevalence rate of the disease is very high, workers are given contraceptives to control the population in cases where people get diagnosed with the causative genes. However, this is not always the case as some may decide to go on to have their children, despite carrying the genes



for the disease. Despite the decision they make in reference to this knowledge, it has proven important that understanding the disease through its evolution has been a breakthrough advancement that still gives scientists hope of discovering the ultimate cure. In the meanwhile, scientists and medics have established a better care plan to reduce the severity of the suffering of an individual. This comes with help.

In conclusion, Huntington's disease has existed amongst the human population for a long time, however, its symptoms have been long misinterpreted for other things such as possession by the evil spirit or mental issues, until recent discovery. This has barred proper treatment of the individuals. However, recent breakthroughs in this field have propelled the enthusiast to find a solution to treat those people who suffer from the disease. Huntington's disease possesses severe effects not only on the people suffering, but to the entire population and offspring that emanate in that line. Therefore, discovering a cure for the disease will be crucial in helping thousands of those directly and indirectly suffering.

#### Work cited

Chao, Michael J., et al. "Population-Specific Genetic Modification of Huntington's Disease in Venezuela." *PLoS Genetics*, vol. 14, no. 5, May 2018, pp. 1-25. *EBSCOhost*, doi: 10. 1371/journal. pgen. 1007274. This academic journal discusses the specific form of Huntington's disease as studied in the country of Venezuela. This specific form of the disease, only present in Venezuela, happens to be the certain form of the virus researched. The history of the disease is mentioned, including when and

where it was discovered, also talking about what methods were used in the treating processes. How the disease had a negative impact on the Venezuelan population is discussed also, and studies were done on those impacted families because HD is genetic, running through families.

Kumar, Sumeet, et al. "Advances in the Discovery of Genetic Risk Factors for Complex Forms of Neurodegenerative Disorders: Contemporary Approaches, Success, Challenges and Prospects." *Journal of Genetics*, vol. 97, no. 3, July 2018, pp. 625-648. EBSCOhost, doi: 10.1007/s12041-018-0953-5.

This academic journal focuses more on the ways that humans have tried to decipher the disease and how they have tried to use the Huntington's Disease cure as a gateway cure to other diseases that still have no known cures. HD has had a positive influence on advancement in medicines and scientific studies of diseases. Studying how the disease affects the people it effects, and why it does, end up being a contributing factor in these medical advancements. Even though there are no current cures, there are many treatments that recently have been discovered to help families live more normal lives.

Lopes, Carla, et al. "Dominant-Negative Effects of Adult-Onset Huntingtin Mutations Alter the Division of Human Embryonic Stem Cells-Derived Neural Cells." *PLoS ONE*, vol. 11, no. 2, Feb. 2016, pp. 1-16. EBSCOhost, doi: 10.1371/journal.pone.0148680.

This academic journal really helped to inform me about how a simple mutation led to a disease, Huntington's Disease, which became so important to be cured in order for humanity to advance and survive. Back when cures  
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were rare for any disease, and anything as small as the common cold could mean death for humans, this disease was a major set-back in the human population evolving and getting more populous. Nowadays the human population is too large to get fully destroyed by a disease like Huntington's Disease, but it was a threat to humanity in the early stages.

Pandey, Mritunjay, and Usha Rajamma. "Huntington's Disease: The Coming of Age." *Journal of Genetics*, vol. 97, no. 3, July 2018, pp. 649-664.

*EBSCOhost*, doi: 10.1007/s12041-018-0957-1.

This academic journal discusses the beginning discoveries and the first sign of studying

Huntington's disease. A brief background is given describing how humans reacted and

treated the disease originally. Once medicines and technology advanced, treatment of HD

advanced along with it significantly. Reading this article made most sense to me and was

fairly easy to understand compared to the other academic sources.

WEN-JUAN HUANG, et al. "Huntington's Disease: Molecular Basis of Pathology and Status of Current Therapeutic Approaches." *Experimental & Therapeutic Medicine*, vol. 12, no. 4, Oct. 2016, pp. 1951-1956. *EBSCOhost*, doi: 10.3892/etm.2016.3566.

This academic journal discusses the chemistry of HD and why the disease occurs in technical

terms. Details about the whole beginning process of the disease is described, which helped

me understand exactly how and why the disease happens. The reason for it being present only

in humans is also stated, because of the complex components of the human brain having

negative downsides, such as being vulnerable to diseases such as HD.