Class:

## Information Gathering Worksheet Answer Key Huntington's Disease

Which genetic disorder are you researching? <u>Huntington's Disease</u> Be sure to cite your sources and use reliable sources throughout.

## What are the symptoms of this disorder?

The symptoms of HD can vary a lot from person to person, but they usually include:

- Personality changes, mood swings & depression
- Forgetfulness & impaired judgment
- Unsteady gait & involuntary movements (chorea)
- Slurred speech, difficulty in swallowing & significant weight loss

Most people with HD experience problems with thinking, behavior, and movements. Symptoms usually worsen over the course of 10 to 25 years and affect the ability to reason, walk, and talk. Early on, a person with HD or their friends and family may notice difficulties with planning, remembering, and staying on task. They may develop mood changes like depression, anxiety, irritability, and anger. Most people with HD become "fidgety" and develop movements of the face and limbs known as chorea, which they are not able to control.

Because of the uncontrolled movements (chorea), a person with HD may lose a lot of weight without intending to, and may have trouble walking, balancing, and moving around safely. They will eventually lose the ability to work, drive, and manage tasks at home, and may qualify for disability benefits. Over time, the individual will develop difficulty with speaking and swallowing, and their movements will become slow and stiff. People with advanced HD need full-time care to help with their day-to-day activities, and they ultimately succumb to pneumonia, heart failure or other complications. The symptoms of HD are sometimes described as having ALS, Parkinson's and Alzheimer's – simultaneously.

Source: https://hdsa.org/what-is-hd/overview-of-huntingtons-disease/

## How common is this disorder?

Today, there are approximately 41,000 symptomatic Americans and more than 200,000 at risk of inheriting the disease.

Source: https://hdsa.org/what-is-hd/overview-of-huntingtons-disease/





Become a Genome Engineer and Explore CRISPR-Cas9's Potential to Cure Human Genetic Disorders! Activity – Information Gathering Worksheet Answer Key Huntington's Disease Name:

Which specific cells, tissues, and organs are affected by this disorder?

Huntington's disease (HD) is an inherited disorder that causes nerve cells (neurons) in parts of the brain to gradually break down and die. The disease attacks areas of the brain that help to control voluntary (intentional) movement, as well as other areas.

Source: https://www.ninds.nih.gov/health-information/disorders/huntingtons-disease

If applicable, what are the current treatments? How successful are they?

There is currently no cure or treatment that can halt, slow, or reverse the progression of the disease. However, there are many treatments and interventions that can help to manage HD symptoms.

Source: https://hdsa.org/what-is-hd/overview-of-huntingtons-disease

What specific gene is mutated in people with this disorder?

Huntingtin

Source: https://hdsa.org/what-is-hd/overview-of-huntingtons-disease

Is this mutation dominant or recessive?

Dominant

Source: https://hdsa.org/what-is-hd/overview-of-huntingtons-disease

What specific mutation causes this disorder? List the healthy and mutated DNA sequence below. Include at least 15 nucleotides before and after the mutation.

HD is caused by a stretch of the letters C-A-G in the huntingtin gene that repeat over and over, too many times...CAGCAGCAGCAGCAG. This is known as a CAG repeat expansion. In the huntingtin gene, most people have around 20 CAG repeats, but people with HD have around 40 or more.

Source: https://hdsa.org/what-is-hd/overview-of-huntingtons-disease





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