

Name:

Date:

Class:

Information Gathering Worksheet **Answer Key Duchenne MD**

Which genetic disorder are you researching? Duchenne Muscular Dystrophy (DMD)

Be sure to cite your sources and use reliable sources throughout.

What are the symptoms of this disorder?

Muscle weakness is the principal symptom of DMD. It can begin as early as age 2 or 3, first affecting the proximal muscles (those close to the core of the body) and later affecting the distal limb muscles (those close to the extremities). Usually, the lower external muscles are affected before the upper external muscles. The affected child might have difficulty jumping, running, and walking. Other symptoms include enlargement of the calves, a waddling gait, and lumbar lordosis (an inward curve of the spine). Later, the heart and respiratory muscles are affected as well. Progressive weakness and scoliosis result in impaired pulmonary function, which can eventually cause acute respiratory failure.

Source: <https://www.mda.org/disease/duchenne-muscular-dystrophy>

How common is this disorder?

In Europe and North America, the prevalence of DMD is approximately 6 per 100,000 individuals.

Source: <https://www.mda.org/disease/duchenne-muscular-dystrophy>

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

The dystrophin protein keeps muscle cells intact. Due to a mutation of the dystrophin gene, muscle cells become damaged, causing weakness and degeneration of muscles.

Source: <https://www.mda.org/disease/duchenne-muscular-dystrophy>

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

There is no known cure for Duchenne muscular dystrophy. Treatment aims to control symptoms to improve quality of life. Steroid drugs can slow the loss of muscle strength. They may be started when the child is diagnosed or when muscle strength begins to decline.

Source: <https://medlineplus.gov/ency/article/000705.htm>

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What specific gene is mutated in people with this disorder?

Dystrophin

Source: <https://www.mda.org/disease/duchenne-muscular-dystrophy>

Is this mutation dominant or recessive?

DMD has an X-linked recessive inheritance pattern and is passed on by the mother.

Source: <https://www.mda.org/disease/duchenne-muscular-dystrophy>

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

60-70% of cases are caused by large deletions of exons. Another 10% of cases are caused by large duplications. 15-30% of cases are caused by point mutations, including nonsense mutations. Large deletions or duplications are not a good target for CRISPR therapy, so students should be guided to look for examples of point mutations that cause this disorder. One example is the rs77354421 variant, where a mutation from G to A causes a nonsense mutation (early stop) in the protein.

Healthy sequence:

5' CCCTTTCAGTAGATGCCTTTTGAACATTCAAACCTTTT 3'

Mutated sequence:

5' CCCTTTCAGTAGATGCCTTTTAAACATTCAAACCTTTT 3'

Source: <https://www.parentprojectmd.org/about-duchenne/what-is-duchenne/types-of-genetic-variants>
https://www.ncbi.nlm.nih.gov/snp/rs77354421#variant_details