The Ultimate Guide to Duchenne Muscular Dystrophy: Causes, Symptoms, Diagnosis, and Treatment

Duchenne muscular dystrophy (DMD) is a rare, progressive genetic disorder that affects primarily males. It is caused by mutations in the dystrophin gene, which is responsible for producing a protein called dystrophin. Dystrophin is essential for maintaining the structural integrity of muscle fibers. Without dystrophin, muscle fibers become weak and damaged, leading to progressive muscle weakness and wasting.

DMD typically affects boys around the age of 3 or 4, although symptoms can sometimes appear as early as infancy or as late as adolescence. The condition is characterized by progressive muscle weakness and wasting in the legs, thighs, pelvis, and trunk. As the disease progresses, muscle weakness spreads to the arms, hands, and eventually the respiratory and cardiac muscles.

A Guide to Duchenne Muscular Dystrophy: Information and Advice for Teachers and Parents by Vicki Iovine

4.8 out of 5

Language	: English
File size	: 2536 KB
Text-to-Speech	: Enabled
Screen Reader	: Supported
Enhanced typesetting	: Enabled
Word Wise	: Enabled
Print length	: 234 pages

There is currently no cure for DMD, but there are treatments available to slow the progression of the disease and improve the quality of life for those affected. These treatments include physical therapy, occupational therapy, speech therapy, and medications such as corticosteroids and anticonvulsants.

Causes of Duchenne Muscular Dystrophy

DMD is caused by mutations in the dystrophin gene, which is located on the X chromosome. The dystrophin gene provides instructions for making the protein dystrophin, which is essential for the structural integrity of muscle fibers.

Mutations in the dystrophin gene can range from small deletions or insertions to large-scale rearrangements of the gene. These mutations disrupt the normal function of the dystrophin gene, resulting in the production of either a non-functional or absent dystrophin protein.

Because the dystrophin gene is located on the X chromosome, DMD is an X-linked recessive disorder. This means that males, who only have one X chromosome, are more likely to be affected by DMD than females, who have two X chromosomes. Females who carry a mutated dystrophin gene are called carriers. Carriers do not typically develop symptoms of DMD, but they can pass the mutated gene on to their children.

Symptoms of Duchenne Muscular Dystrophy

The symptoms of DMD typically begin to appear around the age of 3 or 4, although they can sometimes appear as early as infancy or as late as adolescence. The most common early symptom is difficulty walking or running, which may be noticed by parents or caregivers.

Other early symptoms of DMD include:

- Frequent falls
- Difficulty getting up from a sitting or lying position
- Waddling gait
- Enlarged calf muscles (pseudohypertrophy)
- Muscle weakness in the arms and hands
- Learning difficulties
- Behavioral problems

As DMD progresses, muscle weakness spreads to the arms, hands, and eventually the respiratory and cardiac muscles. Individuals with DMD may experience difficulty breathing, swallowing, and speaking. They may also develop scoliosis and other orthopedic problems.

The life expectancy of individuals with DMD varies depending on the severity of the condition. With early diagnosis and treatment, most individuals with DMD can live into their 20s or 30s. However, some individuals with severe forms of the condition may not live past their teenage years.

Diagnosis of Duchenne Muscular Dystrophy

DMD is typically diagnosed based on a physical examination, family history, and genetic testing.

A physical examination can reveal muscle weakness, wasting, and enlarged calf muscles. A family history of DMD can also increase the

likelihood of a diagnosis.

Genetic testing can confirm the diagnosis of DMD by identifying mutations in the dystrophin

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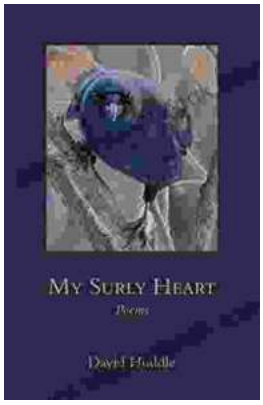

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