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# Muscular Dystrophy

## Patient Education

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## ABSTRACT

Muscular dystrophy is a group of mostly inherited diseases that lead to muscle weakness and a gradual loss of muscle mass. This condition can make everyday activities like walking, climbing stairs, or holding objects challenging, as it can affect different muscles throughout the body. The main issue in muscular dystrophy is the insufficient production of dystrophin, a protein crucial for maintaining muscle tissue integrity and function. Without enough dystrophin, muscles progressively weaken over time. Muscular dystrophy can manifest at any age and varies widely in severity and progression, depending on the specific type. Although there is no cure for muscular dystrophy, various treatments and therapies are available to help manage symptoms and improve quality of life. These options include physical therapy, medications, nutritional

support, and in some cases, surgery. This patient education book provides an extensive overview of muscular dystrophy, covering its types, genetics, symptoms, diagnosis, and management in simpler terms.

**Keywords:** Becker muscular dystrophy; Collagen type VI-related disorders; Congenital muscular dystrophy; Diagnosis of muscular dystrophy; Distal muscular dystrophy; Duchenne muscular dystrophy; Emery-Dreifuss muscular dystrophy; Facioscapulohumeral muscular dystrophy; Genetics of muscular dystrophy; Limb-Girdle muscular dystrophy; Living with muscular dystrophy; Myotonic muscular dystrophy; Oculopharyngeal muscular dystrophy; Symptoms of muscular dystrophy; Treatment and management of muscular dystrophy; Types of muscular dystrophy; X-linked muscular dystrophies

## INTRODUCTION

Muscular dystrophy comprises a group of diseases characterized by progressive weakness and loss of muscle mass. Various types of muscular dystrophy affect specific muscle groups, and symptoms can range from mild to severe. Individuals with muscular dystrophy may experience difficulty walking, sitting up, or moving their limbs, depending on the affected muscles. This condition is typically inherited, though it can also result from new gene mutations. The underlying issue in muscular dystrophy is the insufficient production of a protein called dystrophin, essential for maintaining muscle tissue integrity and function. Without adequate dystrophin, muscles progressively weaken over time. Diagnosing muscular dystrophy involves several steps, beginning with the observation of muscle weakness. The diagnostic process may include genetic testing to identify specific mutations, blood tests to measure muscle enzymes, and imaging

techniques such as muscle biopsies or MRI scans to assess muscle condition. Although no cure exists for muscular dystrophy, current treatments aim to manage symptoms and improve quality of life. These treatments encompass physical therapy to maintain muscle strength and flexibility, medications to slow muscle degeneration, and sometimes surgery to correct deformities or relieve tight muscles (1-7). This patient education book offers a comprehensive overview of muscular dystrophy, covering its types, genetic factors, symptoms, diagnostic processes, and management and treatment options, all explained in simpler terms.

## TYPES OF MUSCULAR DYSTROPHY

Muscular dystrophy is not a single disease, but a group of inherited diseases that damage muscle fibers, which are essential for movement. Each type affects different muscle groups in various ways and progresses at different rates. Some of the common types of muscular dystrophy are presented below in alphabetical order.

### Becker Muscular Dystrophy (BMD)

Becker muscular dystrophy (BMD) is a less severe but still serious form of muscular dystrophy, primarily affecting males. It is characterized by a slower progression of muscle weakness due to mutations in the gene that produces dystrophin, a vital muscle protein. Unlike more severe forms, the body produces partially functional dystrophin in BMD, leading to milder and more varied symptoms. Symptoms typically begin in adolescence or young adulthood and may include muscle weakness in the legs and pelvis, difficulty walking, and a tendency to tire easily. Muscle cramps after exercise are also common. Over time, heart and respiratory issues can develop, requiring ongoing monitoring and management. Although progression is

slower, many individuals with BMD eventually require mobility aids, such as canes or wheelchairs.

## Collagen Type VI-related Disorders

These disorders are a spectrum of genetic conditions that primarily affect the body's ability to produce collagen VI, a protein crucial for the structural integrity and function of muscles and connective tissues. Symptoms typically include muscle weakness, joint laxity (excessive flexibility), and skin abnormalities. The most common forms are Ullrich congenital muscular dystrophy, presenting from birth with severe muscle weakness and joint issues, and Bethlem myopathy, a milder form that may appear in early childhood or adulthood, characterized by slowly progressive muscle weakness, joint stiffness, and contractures. Symptoms can vary widely but often include difficulties walking, a tendency to fall, and respiratory function issues. These disorders can be inherited in an autosomal dominant or recessive pattern.

## Congenital Muscular Dystrophy (CMD)

Congenital muscular dystrophy refers to muscular dystrophies that appear at birth or within the first few months, marked by early onset muscle weakness. These disorders are characterized by significantly reduced muscle tone and may affect various proteins necessary for muscle function. Symptoms commonly include floppy limbs, weak muscle tone, and difficulties with motor functions such as sitting, crawling, and walking. Some types of CMD may also impact brain development, potentially leading to learning difficulties or intellectual disabilities. Complications can include breathing and swallowing difficulties.

## Distal Muscular Dystrophy

This group of rare muscular dystrophies primarily affects the muscles farthest from the center of the body, such as those in the hands, feet, lower arms, and lower legs. Symptoms typically begin in adulthood, from the 20s to the 50s, and include weakness and wasting of the distal muscles, leading to difficulties in fine motor skills like writing, walking, or gripping. Distal muscular dystrophy progresses slowly and is less likely to affect life expectancy, with the severity and progression varying widely depending on the specific type and the individual.

## Duchenne Muscular Dystrophy (DMD)

Duchenne muscular dystrophy is a severe type of inherited muscular dystrophy that primarily affects boys and typically becomes apparent between the ages of three and five. Characterized by rapid progression of muscle weakness that begins in the legs and pelvis, it eventually affects the entire body. DMD results from mutations in the gene that produces dystrophin, a protein essential for maintaining muscle fiber integrity. Insufficient dystrophin causes muscles to weaken over time, leading to difficulties in walking, standing, and eventually impairing breathing and heart function. As the disease advances, most individuals require wheelchairs by their early teens and need comprehensive medical care to manage complications such as cardiac and respiratory conditions.

## Emery-Dreifuss Muscular Dystrophy

Emery-Dreifuss muscular dystrophy is a rare form that primarily affects muscles used for movement, such as those in the upper arms, lower legs, and heart. Symptoms typically begin in childhood or early adolescence and include muscle weakness, stiffness, and joint deformities (contractures), especially in the elbows, ankles, and neck.

A distinctive feature of this condition is the early onset of contractures, which precede muscle weakness and can significantly limit range of motion. As the disease progresses, it often leads to cardiac complications, including arrhythmias and heart block, which can be life-threatening if not properly managed. This type of muscular dystrophy can be inherited in various patterns, including X-linked (most common), autosomal dominant, or autosomal recessive.

## Facioscapulohumeral Muscular Dystrophy (FSHD)

Facioscapulohumeral muscular dystrophy primarily affects the muscles of the face, shoulders, and upper arms. Symptoms usually begin in adolescence or early adulthood and can vary widely from person to person. Some may experience mild symptoms like difficulty smiling, whistling, or lifting objects overhead, while others may encounter more severe muscle weakness that can extend to the lower body. Unlike some other muscular dystrophies, FSHD does not typically affect lifespan, but the muscle weakness can progressively worsen, significantly impacting quality of life. The condition is caused by a genetic mutation that affects the regulation of a specific piece of DNA, leading to muscle tissue degeneration.

## Limb-Girdle Muscular Dystrophy

Limb-girdle muscular dystrophy encompasses a group of genetic disorders characterized by progressive weakness and wasting of the muscles around the hips and shoulders, known as the limb-girdle muscles. Symptoms typically begin in childhood or young adulthood and can vary significantly in severity and progression, depending on the specific genetic mutation involved. As the disease progresses, individuals may find it difficult to walk, climb

stairs, and lift objects. Over time, muscle weakness may spread to other parts of the body, including the arms and legs. There are several subtypes of limb-girdle muscular dystrophy, each associated with defects in different genes that affect muscle cell function.

## Myotonic Muscular Dystrophy

Myotonic muscular dystrophy is a type of muscular dystrophy characterized not only by progressive muscle weakness but also by myotonia, where muscles have difficulty relaxing after contraction. It is the most common form of adult muscular dystrophy and uniquely affects multiple body systems, including muscles, the heart, brain, and endocrine system, leading to a wide range of symptoms. Common signs include prolonged muscle tensing, difficulty releasing a grip, facial muscle weakness, drooping eyelids, and problems with walking and balance. It may also cause cataracts, heart conduction defects, sleep apnea, and hormonal imbalances. Myotonic dystrophy can manifest at any age, from birth to adulthood, and symptoms often worsen with each generation—a phenomenon known as anticipation. The progression is slow, with life expectancy varying based on symptom severity and complications.

There are two main types of myotonic muscular dystrophy: Type 1 (DM1) and Type 2 (DM2), both involving muscle weakness, myotonia, and multi-organ issues. However, they differ in genetic causes, age of onset, severity, and symptom distribution.

Type 1 Myotonic Dystrophy (DM1), also known as Steinert's disease, is more common and severe. Symptoms usually appear in early adulthood, caused by a mutation where a CTG nucleotide sequence is excessively repeated in the

DMPK gene. The number of repeats tends to increase with each generation, leading to earlier onset and more severe symptoms over time.

Type 2 Myotonic Dystrophy (DM2) typically has a milder presentation and later onset, usually affecting individuals in middle age. It involves a mutation where a CCTG nucleotide sequence is excessively repeated in the CNBP gene. Unlike DM1, symptoms in DM2 do not usually worsen significantly in subsequent generations.

## Oculopharyngeal Muscular Dystrophy (OPMD)

Oculopharyngeal muscular dystrophy begins typically in middle age, primarily affecting the muscles around the eyes and throat. Key symptoms include drooping eyelids (ptosis) and difficulties swallowing (dysphagia), potentially leading to frequent choking or recurrent respiratory infections. As the disease progresses, it may also affect the muscles of the face, neck, and limbs, although this progression is generally slower than other types of muscular dystrophy. OPMD is inherited in an autosomal dominant pattern, meaning the disorder can be caused by just one copy of the altered gene.

## Other Forms of Muscular Dystrophy

There are additional, rarer types of muscular dystrophy that affect specific muscle groups, each progressing at its own rate. Despite their differences, all types share the common feature of progressively worsening muscle weakness.



# GENETICS OF MUSCULAR DYSTROPHY

Muscular dystrophy is primarily a genetic condition caused by mutations in genes essential for maintaining healthy muscle fibers. These mutations impair the proteins needed to build and sustain strong muscles, leading to muscle weakness and degradation. The genetic mutations can be inherited from one or both parents, or they may occur spontaneously in an individual's genes. The genetics of muscular dystrophy can be complex due to the variety of forms, each linked to different mutations.

## X-linked Muscular Dystrophies: Duchenne and Becker

In Duchenne and Becker muscular dystrophies, the faulty gene is located on the X chromosome. Since males have only one X chromosome, a single altered copy of the gene can cause the disease. Females, having two X chromosomes, are typically carriers and usually do not show symptoms because they have a second, normal copy of the gene.

*Duchenne Muscular Dystrophy (DMD):* Caused by mutations in the DMD gene responsible for producing dystrophin, a protein that helps stabilize and protect muscle fibers. In Duchenne, these mutations lead to the absence or near absence of dystrophin, resulting in muscle cell damage and progressive muscle weakness.

*Becker Muscular Dystrophy (BMD):* Also caused by mutations in the DMD gene. However, in Becker muscular dystrophy, the mutations allow some production of dystrophin, but the protein is either insufficient or partially

functional. This results in milder and more slowly progressing muscle weakness compared to Duchenne.

## Other Types of Muscular Dystrophy

*Myotonic Muscular Dystrophy:* Includes two types, each associated with a different genetic mutation. Type 1 (DM1) involves an expanded CTG repeat in the DMPK gene, while Type 2 (DM2) has an expanded CCTG repeat in the CNBP gene. These expansions disrupt normal muscle cell function, causing myotonia and muscle weakness.

*Limb-Girdle Muscular Dystrophy:* Involves various genes such as CAPN3, SGCA, SGCB, SGCD, and SGCG, responsible for proteins that support muscle cell structure and function. Mutations in these genes lead to weakness primarily in the shoulders and hips.

*Congenital Muscular Dystrophy (CMD):* Linked to mutations in several genes, including LAMA2 for laminin and COL6A1, COL6A2, and COL6A3 for collagen VI, both crucial for muscle structure and stability.

*Facioscapulohumeral Muscular Dystrophy (FSHD):* Associated with genetic changes near the end of chromosome 4, particularly a deletion in the D4Z4 region affecting the regulation of the DUX4 gene, leading to muscle cell damage.

*Emery-Dreifuss Muscular Dystrophy:* Typically linked to mutations in the EMD and LMNA genes, which affect proteins essential for the structural health of the cell nucleus, compromising muscle cell stability and function.

*Distal Muscular Dystrophy:* Involves mutations in genes like DYSF, GNE, and MYOT, essential for normal muscle function, affecting the distal parts of the limbs.

*Oculopharyngeal Muscular Dystrophy (OPMD):* Caused by an abnormal expansion in the PABPN1 gene, leading to the production of an abnormal protein that disrupts muscle cell function, particularly in the eyelids and throat.

*Collagen Type VI-Related Disorders:* Such as Ullrich congenital muscular dystrophy and Bethlem myopathy, are caused by mutations in the COL6A1, COL6A2, and COL6A3 genes, critical for producing collagen VI, a key component of connective tissues.

The broad variation in genetic causes among the different types of muscular dystrophy explains the wide variability in symptoms, severity, and age of onset. Although the genetic basis of muscular dystrophy is well-understood, many families may not be aware of the disorder until a child is diagnosed, which can occur due to new mutations or mild symptoms in carriers.

## SYMPTOMS OF MUSCULAR DYSTROPHY

The symptoms of muscular dystrophy vary widely depending on the type and severity of the disease. However, certain common signs can help indicate its presence. The most noticeable symptom is muscle weakness, which typically begins in the lower limbs and pelvic area but may also start in the arms and other parts of the body, depending on the type of muscular dystrophy. For instance, children with Duchenne muscular dystrophy often experience difficulty running, jumping, and climbing stairs. They may also have a tendency to fall frequently and struggle to rise

from a sitting or lying position. This struggle often involves a characteristic maneuver known as the Gowers' sign, where the child has to "walk" their hands up their legs to stand up. As the disease progresses, muscle weakness spreads to other parts of the body, and the affected muscles decrease in size, a condition known as muscle wasting. In some cases, the muscles may appear larger due to an increase in fat and connective tissue, a phenomenon called pseudohypertrophy, which is particularly common in the calves.

Muscular dystrophy can also lead to stiff or tight joints (contractures) due to the weakening of muscles around the joints. This weakening can severely limit mobility and flexibility over time. Additionally, some types of muscular dystrophy may cause difficulties with swallowing or breathing as the muscles responsible for these functions weaken.

In more advanced stages, the disease can affect the heart and respiratory muscles, leading to serious complications such as cardiomyopathy—a disease of the heart muscle that makes it harder for the heart to pump blood to the rest of the body—and respiratory failure. These complications can significantly affect life expectancy and quality of life.

## DIAGNOSIS OF MUSCULAR DYSTROPHY

Diagnosing muscular dystrophy is a comprehensive process that often involves several healthcare professionals, including neurologists, geneticists, and physical therapists. The diagnostic procedure begins with the recognition of symptoms such as muscle weakness, difficulty walking, or frequent falls, which may suggest a muscle disorder. A detailed family history is essential, as it

can reveal whether there is a pattern of similar symptoms, suggesting a genetic condition. During the physical examination, the doctor assesses muscle strength and checks for signs typical in some forms of muscular dystrophy, such as enlarged calves or abnormal postures.

If muscular dystrophy is suspected, the next steps generally include blood tests. One common test measures levels of enzymes such as creatine kinase (CK) that leak out of damaged muscle cells into the bloodstream. Elevated CK levels can indicate muscle damage associated with muscular dystrophy or other muscle diseases. Genetic testing, which searches for specific mutations in genes known to cause the disease, is increasingly used to identify the exact type of muscular dystrophy. This testing can confirm the diagnosis, help predict the disease's progression, and inform family planning decisions.

While muscle biopsy was once a staple in diagnosing muscular dystrophy, it is less frequently used today due to advances in genetic testing. However, it remains valuable when genetic tests are inconclusive, showing abnormalities in muscle fibers or the absence of certain proteins typically found in muscular dystrophy.

Imaging tests like MRI (Magnetic Resonance Imaging) are also employed to visualize the condition of the muscles. MRI can reveal areas where muscle has been replaced by fat and connective tissue, aiding in assessing muscle damage and the progression of the disease. Electromyography (EMG), which measures the electrical activity of muscle fibers, may be performed to differentiate muscular dystrophy from other conditions affecting muscle function and nerves. Additionally, Nerve Conduction Velocity (NCV) tests measure the speed at which electrical impulses travel through the nerves, helping to distinguish

between muscle diseases and nerve disorders that could cause similar symptoms. In cases of muscular dystrophy, the primary issue is with the muscles themselves, not the nerves. Confirming normal nerve function is crucial for ruling out neuropathic conditions that can mimic or complicate muscle diseases.

## TREATMENT AND MANAGEMENT OF MUSCULAR DYSTROPHY

Treating and managing muscular dystrophy focuses on slowing disease progression, alleviating symptoms, and enhancing quality of life, as there is currently no cure. The approach varies depending on the type of muscular dystrophy, symptom severity, and the age at which the disease presents.

### Physical Therapy

Physical therapy is crucial for managing muscular dystrophy, aimed at maximizing mobility and function while minimizing muscle weakness and degeneration. Physical therapists design specific exercises to strengthen remaining muscle function, enhance flexibility, and reduce stiffness. They work to prevent or manage contractures—stiff joints caused by tight muscles—and regularly incorporate stretching exercises to maintain joint flexibility and ease daily activities. Physical therapists also recommend assistive devices such as braces, walkers, or wheelchairs to prolong independence and tailor exercise programs to individual needs, ensuring safe and effective activities that improve quality of life. They also educate patients and families about the disease's progression and necessary care adjustments.

## Medications

No medications currently cure muscular dystrophy, but corticosteroids like prednisone are commonly prescribed to slow muscle degeneration and improve strength and function. These steroids can have side effects such as weight gain, bone thinning, and increased infection risk, requiring careful management. Muscle relaxants help alleviate muscle stiffness and spasms, common as the disease progresses, easing discomfort and improving mobility. However, their use must be closely monitored due to potential side effects like drowsiness and dependency.

## Cardiac Care

Cardiac care is vital since many muscular dystrophy types can affect the heart, potentially leading to life-threatening complications. Routine cardiac evaluations, including electrocardiograms (ECG) and echocardiograms, are essential to monitor heart function and detect cardiomyopathy or arrhythmias early. Treatment may include medications like beta-blockers and ACE inhibitors to manage blood pressure and reduce heart strain. In severe cases, devices such as pacemakers or implantable cardioverter defibrillators (ICDs) might be necessary to maintain normal heart rhythm.

## Respiratory Care

As muscular dystrophy progresses and affects respiratory muscles, maintaining adequate breathing becomes crucial. Regular assessments by pulmonologists, including spirometry tests, are essential to monitor lung function and manage breathing difficulties, particularly during sleep or infections. Treatment strategies may involve non-invasive ventilation devices like BiPAP machines at night or mechanical ventilators for advanced respiratory muscle weakness. Respiratory therapists also provide breathing

exercises to maintain lung capacity and teach techniques to clear airways and prevent infections.

## Surgery

Surgery in muscular dystrophy is considered for specific complications that can be surgically addressed to improve mobility, function, or quality of life. Common procedures include releasing contractures to increase joint motion and flexibility and spinal surgeries like spinal fusion to correct or stabilize scoliosis, which improves pain management, prevents further curvature, and supports better lung function. In severe cases, procedures such as tracheostomy may be performed to assist breathing, and cardiac surgeries might be needed to manage heart complications related to the disease.

## Nutritional Support

Nutritional support is crucial in managing muscular dystrophy, helping to optimize overall health and address specific symptoms of the disease. As muscular dystrophy progresses, muscle weakness can complicate the simple acts of eating and digesting food, potentially leading to nutritional deficiencies and weight management issues. Dietitians or nutritionists work closely with patients to customize diets that meet their individual needs. A well-balanced diet rich in proteins, vitamins, and minerals is essential to maintain muscle health and energy levels. Additionally, caloric intake may need careful adjustment as some individuals with muscular dystrophy may struggle with being overweight due to reduced mobility, while others may experience weight loss due to muscle wasting. When swallowing difficulties arise, dietary modifications such as softer foods or thickened liquids become necessary to prevent choking and ensure adequate nutrition. Nutritional supplements might also be prescribed to address specific



deficiencies. Maintaining proper nutrition is crucial for supporting immune function, promoting muscle and bone health, and enhancing the quality of life.

## Psychological Support

Psychological support is an integral part of managing muscular dystrophy. Living with a chronic, progressive condition can be challenging not only for the patient but also for their family. Mental health professionals, support groups, and community resources provide necessary support to manage the emotional aspects of the disease.

A multidisciplinary approach involving neurologists, cardiologists, pulmonologists, and physical therapists ensures comprehensive care tailored to each individual's needs, helping to manage symptoms effectively and improve overall well-being.

## LIVING WITH MUSCULAR DYSTROPHY

Living with muscular dystrophy is a journey that requires adapting to physical changes and challenges over time. The impact of the disease extends beyond physical symptoms; daily activities such as walking, dressing, or eating can become increasingly difficult. As the disease progresses, assistive devices such as wheelchairs, braces, and specialized vehicles become essential for maintaining mobility and independence, though adapting to these tools can take time and effort.

Home modifications like ramps, wider doorways, and adapted bathrooms with grab bars can significantly ease daily life. Families often need to consider these modifications early to keep ahead of changing needs.

Education and employment may also be affected; children may require school adaptations, while adults might need flexible working arrangements or career changes if physical limitations impact job performance.

Regular visits to various specialists are necessary to monitor the progression of the disease and manage symptoms. This requires organization and can be time-consuming but is crucial for effective disease management.

The emotional and psychological impacts of muscular dystrophy are profound. Individuals and their family members may experience feelings of grief, frustration, and isolation. Support groups and counseling provide emotional support and practical advice, helping connect with others facing similar challenges. These networks offer comfort and companionship.

Financial planning is another important consideration, as medical costs and home modifications can be significant. Exploring insurance options, government assistance, and community support programs can help alleviate financial burdens associated with long-term medical care.

## CONCLUSION

Muscular dystrophy is a challenging condition affecting both the physical and emotional lives of those diagnosed and their families. While there is no cure, advances in medical research and treatment options continue to improve quality of life and extend capabilities. Early diagnosis is crucial for timely intervention through therapies and support that can significantly delay symptom progression. The journey involves adapting to changes in mobility and daily activities, navigating healthcare needs, and addressing the psychological impacts of the disease.

The support of a strong healthcare team, alongside love and understanding from family and friends, is essential. Community resources, support groups, and counseling offer additional layers of support, helping individuals and families lead fulfilling lives despite the limitations imposed by the disease.

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