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A STUDY ON DUCHENNE MUSCULAR DYSTROPHY: CLINICAL FEATURES AND DISEASE PROGRESSION

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ABSTRACT

Duchenne Muscular Dystrophy (DMD) is a genetic disorder characterized by progressive muscle weakness and degeneration, primarily affecting boys. It is caused by mutations in the dystrophin gene. DMD leads to loss of muscle function, respiratory and cardiac complications, and reduced life expectancy. It primarily affects boys and usually appears in early childhood. Symptoms include difficulty walking, frequent falls, and heart and respiratory problems. Although there is no cure, treatments such as corticosteroids, gene therapy, and physiotherapy can slow disease progression and improve quality of life. Current management focuses on slowing disease progression, managing symptoms, and improving quality of life. On-going research explores innovative treatments and potential cures.

KEYWORDS: DMD, Pseudo hypertrophic progressive disease, Muscular Dystrophy, Duchenne's disease, X-linked recessive disease.

1. INTRODUCTION

Duchenne muscular dystrophy (DMD) is a severe, progressive, and incurable X-linked disease. Mutations in the dystrophin gene cause DMD. The lack of functioning dystrophin protein in DMD patients causes long-term injury to muscle fibers during contraction, which eventually degrades muscular quality and causes muscle mass loss.[1]

The dystrophin gene, one of the biggest human genes with 79 exons and over 2.4 million base pairs, has more than 7,000 mutations that result in the absence of functional dystrophin protein in patients with DMD.[2] Dystrophin is crucial for maintaining muscular function because it connects the trans membrane elements of the dystrophin-glycoprotein complex (DGC), such as dystroglycan, sarcoglycans, and sarcospan, to the intracellular cytoskeleton network of muscle fiber cells.[3]

The extracellular surface of sarcolemma, a specialized cell membrane that surrounds striated muscle fiber cells, contains α-dystroglycan, which is a receptor for laminin-2 and connects the DGC to the extracellular matrix (ECM); on the other hand, β-dystroglycan is a trans membrane protein that is closely linked to α-dystroglycan and binds to dystrophin.[3]

Lack of dystrophin damages muscle fibers during contraction by weakening the connection between the cytoskeleton and sarcolemma.[4] Both inflammation and the suppression of muscle fiber regeneration are the outcomes of this long-term injury. About 1 in 5,000 males have mutations in the dystrophin gene, which results in insufficient dystrophin production for appropriate muscle function because the gene is located on the X chromosome.[5]

Early childhood is typically when symptoms first appear, and they gradually worsen.

A female will be a carrier for the condition if she inherits a dystrophin mutation on one of her X chromosomes, but she will typically receive enough dystrophin from her healthy gene on the other X chromosome. Males are usually affected, and it is inherited.

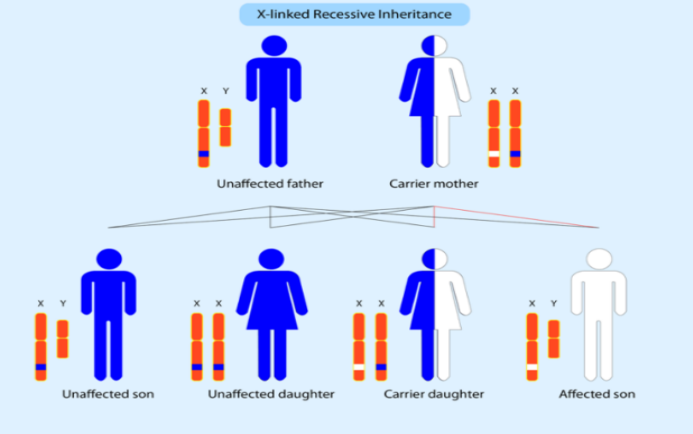

Figure 1: Functional Decline In Dmd.

Clinically, DMD symptoms first appear between the ages of two and three. These are followed by a progressive decline in multiple systems, such as respiratory insufficiency, musculoskeletal abnormalities, cardiomyopathy, and muscle weakness.[6]

Dystrophin depletion results in membrane instability with increased vulnerability to damage, fiber necrosis, and disruption of the dystrophin-associated glycoprotein complex (DGC). Gene therapy is a viable treatment for the majority of DMD patients. It involves delivering a therapeutic gene to the heart and skeletal muscle to replace the dystrophin protein.

The discovery of the genes and proteins they encode that cause human genetic illnesses has exploded as a result of current molecular genomic methods. The identification of the gene altered by mutations in Duchenne and Becker muscular dystrophy was one of the earliest examples. DMD has a genetic origin, and repairing the damaged reading frame is necessary. Genetic methods that target the impacted muscle cells may be able to treat the condition.[7]

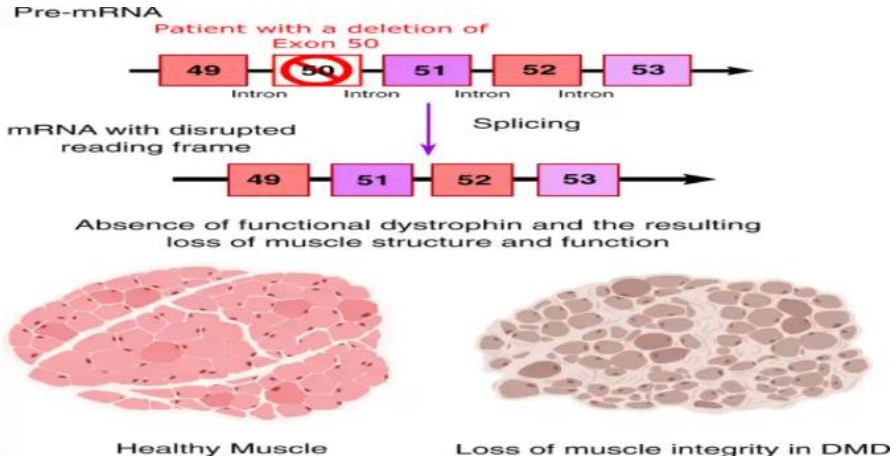

Figure 2: Variants in DMD gene.

DMD INHERITANCE

DMD is X-linked recessive, typically inherited from family members, and is caused by a gene on the X chromosome, one of the two sex chromosomes:

- Boys have one X and one Y chromosome, with the X coming from the mother and the Y from the father.
- Girls possess two X chromosomes, one from each parent.

Since boys only have one X chromosome, they will develop DMD if that X chromosome possesses the gene mutation that causes it. This is because they lack a backup copy on a different X chromosome to accommodate. However, girls usually won't be impacted because they have a backup copy on their other X chromosome.

The symptoms in girls are either very mild or none. Girls have the potential to become carriers. The gene mutation is inherited by carriers, although they show no symptoms. Their future offspring might inherit the gene.

HISTORY

- An essay describing a disease that caused gradual muscle weakness in boys was written in 1830 by Sir Charles Bell, a Scottish surgeon, anatomist, physiologist, and neurologist. Bell was the first to report on muscular dystrophy.[8]
- The cases of two brothers with growing muscle weakening were reported by Conte and Gioja in 1836, while a family consisting of four sons with notable muscle abnormalities was reported by Edward Meryon in 1852.The French neurologist Guillaume Benjamin Amand Duchenne was the first to describe in detail 13 boys who had a severe form of the disease that would later be known as Duchenne muscular dystrophy (DMD) in 1868.[8]
- In the 1950s, German physician Peter Emil Becker was the first to describe the milder form of DMD, and he also gave the illness its own name[9].The DMD gene was the first gene to be discovered by positional cloning.[10] In 1987, Dr. Michael Koenig cloned the complete gene. The gene for DMD was known to be found on the X chromosome since the disorder was inherited in an X-linked way.
- As the disease progresses, it affects the heart and respiratory muscles, significantly impacting quality of life and expectancy. The progression of Duchenne Muscular Dystrophy (DMD) follows four clinical stages.

Table 1: Stages of DMD.

Stages	Age range	Key features
Early Ambulatory	2-7 years	Delayed motor milestone, Difficulty in running/climbing, Gower's sign (using hands to push off thighs when standing up).
Late Ambulatory	7-12 years	Frequent falls, Trouble with stairs, Fatigue, Starts to use wheelchair occasionally.
Early Non-Ambulatory	10-14 years	Loss of walking ability, Wheelchair full-time, Arm/ trunk weakness, Risk of scoliosis.
Late Non-Ambulatory	Teens and other	Severe upper limb weakness, Breathing issues (requires ventilation support)

2. AIM OF DUCHENNE MUSCULAR DYSTROPHY

To study Duchenne Muscular Dystrophy (DMD) with emphasis on its genetic basis, clinical features, diagnostic methods, and current treatment strategies, while exploring recent advancements in therapy and patient care.

OBJECTIVE OF DMD

1. Symptom management: Controlling muscle weakness, respiratory issues, and cardiac complications.
2. Functional preservation: Maintaining mobility, strength, and independence.
3. Complication prevention: Preventing or minimizing respiratory, cardiac, and skeletal complications.
4. Support and care: Providing emotional, social, and psychological support for individuals and families.
5. Research and innovation: Advancing understanding, treatment, and potential cures through research and innovation.
6. To highlight the role of multidisciplinary care in managing DMD patients.

3. ETIOLOGY

A mutation in the dystrophin gene, which is found on chromosome Xp21, causes DMD, a hereditary disorder. Although it is inherited as an X-X-linked recessive condition, novel mutations account for about 30% of cases.[11,12] A crucial structural protein complex necessary for preserving muscle integrity and dynamism is dystrophin which is absent, unstable, or drastically decreased as a result of mutations in the DMD gene, most frequently frame shift or nonsense mutations.

The largest human gene is dystropin, it has 79 exons and 2.4 million base pairs. The term dystrophinopathies refers to disorders caused by mutations in the dystrophin gene, which include Becker muscular dystrophy, Duchenne muscular dystrophy, and an intermediate variant. The brain, retina, striated and heart muscle, and other tissues all express dystrophin. Compared to muscles, the brain has a smaller distribution.[13]

The mutation primarily affects males and is usually inherited in an X-linked recessive fashion. It may impact between 2.5% and 20% of female carriers. The Lyon hypothesis explains this, according to which the mutated X chromosome is expressed while the normal X chromosome is rendered inactive.

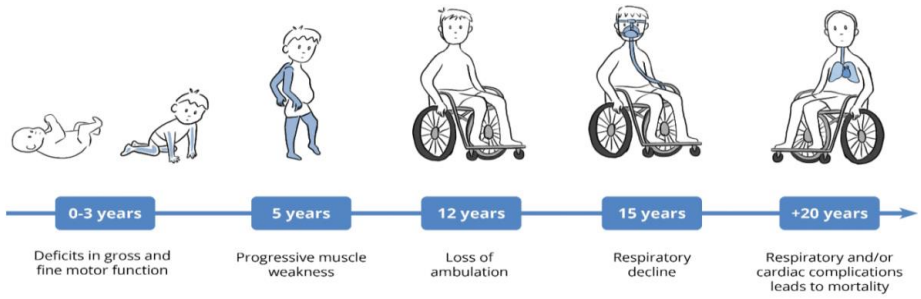

Figure 3: functional decline of dmd.

Table 2: Comparison Chart Decreased Vs Increased Dystropin.

Feature	**Decreased/Absent Dystrophin *(as in DMD)***	**Increased/Restored Dystrophin *(via therapy)***
Muscle Cell Stability	Fragile, easily damaged	More stable, better protected
Muscle Function	Progressive weakness, degeneration	Improved function, slower decline
Mobility	Loss of ambulation (typically by age 10–12)	Prolonged ability to walk
Cardiac Health	Cardiomyopathy, heart failure risk	Better cardiac function
Respiratory Function	Breathing difficulty, need for ventilation in later stages	Slower respiratory decline
Muscle Repair	Inefficient; leads to fibrosis and fat replacement	Improved regeneration, less fibrosis
Quality of Life	Reduced independence and activity	Improved mobility and function
Lifespan	Shortened(20s–30s average without intervention)	Potentially extended with therapy
Disease Progression	Rapid and severe	Slower and milder (especially in Becker-like phenotypes)

4. PATHOPYSIOLOGY

Mutations in the DMD gene result in the absence of dystrophin, a crucial structural protein in muscle cells, which causes Duchenne Muscular Dystrophy (DMD), an X-linked recessive illness.

1. **Genetic Basis:** The DMD gene is one of the largest human genes, located on X chromosome (Xp21.2).Mutations (e.g., deletions, duplications, point mutations) result in nonfunctional or absent dystrophin.[14]
2. **Lack of dystropin protein:** A component of the dystrophin-glycoprotein complex (DGC), which links the extracellular matrix and muscle cell actin cytoskeleton, is dystrophin. During contraction, it stabilizes the muscular membrane (sarcolemma).[15]
3. Both the dystrophin and DGC proteins are absent in Duchenne muscular dystrophy. This leads to oxidative damage, increased membrane permeability and fragility, and disruption of calcium homeostasis, all of which promote muscle cell necrosis.[16]
4. **Membrane Fragility and Muscle Damage:** Lack of dystrophin makes the sarcolemma fragile. Muscle contractions lead to microruptures, which causes calcium influx, activation of proteolytic enzymes (e.g., calpains) and muscle cell necrosis. Thus it results in muscle weakness and degeneration.[17]
5. **Inflammation and Fibrosis:** Damaged muscle fibers trigger chronic inflammation. Inflammatory cells release cytokines (e.g., TNF-α), worsening damage. Macrophages and cytokines exacerbate muscle destruction. Fat and fibrotic tissue gradually replaces muscle.[18]
6. **Progressive Muscle Degeneration:** Progressive muscle weakness starts with Proximal limb muscles (pelvic and shoulder girdles) then respiratory and cardiac muscles which results in cardiomyopathy and respiratory failure.[19]

FLOWCHART OF DMD MECHANISM

Genetic Mutation in DMD Gene (X-linked Recessive Disorder)

↓

Deficiency or Absence of dystrophin protein

↓

Without dystrophin muscle cell membrane becomes fragile and muscle fiber damage during contraction due to ca^{2+} influx and enzyme leakage.

↓

Cycles of muscle degeneration and regeneration

↓

Replacement of muscle tissue by fat and fibrosis which causes progressive muscle weakness and atrophy

↓

Loss of Ambulation → Respiratory & Cardiac Muscle Involvement

5. SIGNS AND SYMPTOMS

- **Early Signs and Symptoms (Usually between ages 2–5)**
  Delayed motor milestones, Frequent falls, Waddling gait, Difficulty running or climbing stairs, Gower's sign,[20] Enlarged calf muscles (pseudohypertrophy)
- **Progressive Symptoms (School age to adolescence)**
  Progressive muscle weakness, starting in the pelvic & shoulder girdle muscles, Contractures, Lordosis, Wheelchair dependency (usually by age 12), Scoliosis.[21]
- **Late Symptoms (Teenage years and beyond)**
  Cardiomyopathy – progressive heart muscle disease,[20] Arrhythmias, Respiratory difficulties due to weakening of diaphragm and intercostal muscles, Fatigue and poor endurance, Learning and behavioral difficulties (in some cases).

6. COMPLICATIONS

a) **Musculoskeletal complications:** Progressive muscle weakness, Loss of ambulation, Contractures, Scoliosis.[6]

b) **Respiratory complications:** Respiratory muscle weakness, Recurrent chest infections, Restrictive lung disease, Respiratory failure.[22]

c) **Cardiac complications:** Dilated cardiomyopathy, Arrhythmias, Heart failure.[23]

d) **Gastrointestinal & Nutritional complications:** Dysphagia, Delayed gastric emptying and constipation, Obesity, Malnutrition.[24]

e) **Other complications:** Cognitive / learning difficulties, Psychological issues.[25]

Foods Beneficial for DMD

The goal is to support muscle health, reduce inflammation, maintain healthy weight, and support the heart and bones.

- ✓ Anti-inflammatory foods such as fatty fish (salmon, sardines, mackerel), Nuts and seeds (chia, flax, walnuts).
- ✓ High-Fiber fruits and vegetables such as berries, leafy greens, broccoli, carrots, sweet potatoes.
- ✓ Whole grains such as oats, quinoa, brown rice, whole wheat.

- ✓ Lean proteins such as poultry, legumes, beans, tofu.
- ✓ Vitamin D and calcium-rich foods such as fortified plant milks, leafy greens, almonds.
- ✓ People with DMD may need: Vitamin D, Calcium, CoQ10, Creatine monohydrate (some evidence it may help muscle strength), Omega-3 fatty acids.

Foods to Avoid (or Limit) for DMD:

These are foods that may worsen inflammation, promote weight gain, or stress the cardiovascular and gastrointestinal systems:

- ✓ Highly processed foods such as chips, fast food, packaged snacks.
- ✓ Sugary foods and beverages such as candy, soda, sweetened cereals.
- ✓ Excessive sodium present in processed meats, canned soups, frozen meals.
- ✓ Saturated & Trans fats in fried foods, margarine, commercial baked goods.
- ✓ Red and Processed meats such as bacon, sausages, hot dogs.

7. DIAGNOSIS AND TEST OF DUCHENNE MUSCULAR DYSTROPY

A neuromuscular specialist who can do a clinical evaluation on the kid and quickly access and interpret relevant studies within the framework of the clinical presentation should make the diagnosis. Geneticists and genetic counselors will frequently provide further help to families following a diagnosis.

a. CLINICAL SINGS AND SYMPTOMS

- ✓ DMD is usually diagnosed around age five.
- ✓ Due to delay that will attain developmental milestones. (eg: Independent walking) So, the diagnosis may have been suspected much earlier.
- ✓ A progressive loss of muscular mass begins in the shoulder and pelvic girdles.
- ✓ Calf pseudo hypertrophy, which is an enlargement of the calves brought on by connective tissue and fat.

b. LABORATORY TESTING

Creatinine Kinase test

- ✓ One of the first tests done when DMD is suspected.
- ✓ The heart, brain, and skeletal muscle are the primary locations for the enzyme creatine kinase.
- ✓ The most common sign that raises the possibility of DMD may be an increase in serum levels of creatine kinase (CK), a biomarker of muscle degeneration and membrane fragility.

c. GENETIC TESTING

- ✓ Multiplex PCR is a genetic test frequently used to detect dystrophin mutations in DMD.
- ✓ Performed via blood sample.
- ✓ Mutations in the X chromosome's dystrophin gene cause DMD.
- ✓ Most common mutations: deletions, duplications, point mutations.

METHODS

MLPA (Multiplex Ligation-dependent Probe Amplification): Good for detecting large deletions/duplications.

Next-Generation Sequencing (NGS): Detects small mutations.

d. MUSCLE BIOPSY (used less frequently now)

- ✓ Muscle biopsy is used if genetic testing is inconclusive.
- ✓ In cases of dispute, muscle biopsy can give phenotypic diagnosis, lead cost-effective molecular studies, and establish the toxicity of novel gene variations.
- ✓ It can be carried out under local anesthetic using either an open or needle method. The diseased muscle should be biopsied, but it shouldn't be very weak or atrophic.
- ✓ It shows Muscle fiber degeneration and regeneration and Absence or deficiency of dystrophin protein via immunohistochemistry or Western blot.

8. TREATMENT AND MANAGEMENT

- ➢ DMD does not currently have a cure. Symptom management and slowing progression are the primary objectives of treatment.
- ➢ This congenital dystrophy has a terrible prognosis and no known medical treatment. Glucocorticoid therapy, contracture prevention, and medical management of cardiomyopathy and respiratory impairment are the mainstays of treatment.[26]
- ➢ Vamorolone, a novel glucocorticoid therapy, is being researched for boys with DMD. According to preliminary findings, the treatment provided advantages comparable to those of prednisone without the negative side effects.[27]
- ➢ Treatment research is still ongoing, and some gene-based strategies appear promising for delaying or even reversing some of the symptoms of specific MD types.[28] It is best to manage patients with DMD with an interprofessional team that consists of therapists and nurses with specialized training
- ➢ DMD is known to be caused by a number of distinct mutations, or genetic alterations. Not all DMD patients have the same genetic mutation.[29]
- ➢ There is no proof that taking vitamins or using any other alternative therapy can help people with DMD avoid or treat muscle weakness.[30]

1. CORTICOSTEROID THERAPY

The first-line pharmacological treatment for DMD is corticosteroids. They increase muscle strength and function, prolong walking, and postpone heart problems.

BENEFITS: Enhances motor function and muscle strength. Slows the deterioration of cardiac and pulmonary function. Prednisone and Deflazocort reduces the Inflammation and slows the muscle degeneration.

2. MEDICATIONS

- **Duvyzat (givinostat):** By preventing the synthesis of an inflammatory protein, this drug, which is taken twice a day with food, reduces muscle degeneration in DMD patients.
- **Corticosteroids:** By lowering inflammation, these drugs help DMD patients avoid damaging their muscles. Corticosteroids also aid in maximizing muscular function.
- If cardiomyopathy develops as a result of DMD, beta-blockers can assist stabilize and manage cardiac function by lowering heart rate and increasing the strength of the heart muscles as they pump.
- DMD is treated with medications called angiotensin converting enzyme (ACE) inhibitors, which also control heart function.

3. EXON SKIPPING THERAPY

Exon skipping therapy is a genetic treatment that skips over defective regions (exons) of a gene during mRNA processing by using tiny molecules known as antisense oligonucleotides (AONs).

BENEFITS: Slows Disease Progression, delays Loss of Ambulation, Improves Respiratory and cardiac function.

4. GENE THERAPY

Gene therapy addresses the underlying genetic issue rather than just treating its symptoms.
Fixes or replaces the damaged dystrophin gene in DMD.

BENEFITS: helps make dystrophin, improving muscle strength and stability, missing or nonfunctional.

5. CARDIAC MANAGEMENT

Annual cardiac MRI or echocardiogram (from age 6–10 forward)
Finds early cardiac abnormalities therefore symptoms show up.

BENEFITS: Delays onset of cardiomyopathy, Preserves heart function longer, Reduces risk of heart failure and arrhythmias.

6. PHYSICAL THERAPY AND SUPPORTIVE CARE

PHYSICAL THERAPY: Daily stretching: Prevents muscle contractures.
Range of motion exercises: Maintains joint flexibility.
Orthotic devices: Ankle-foot orthoses (AFOs) support walking and prevent deformities.
SUPPORTIVE CARE: Occupational therapy: Aids in daily activities and independence.
Nutritional support: Prevents obesity and supports overall health.

DRUGS USED TO TREAT MUSCULAR DYSTROPHY

For Duchenne Muscular Dystrophy, a number of medications are currently on the market, including nonsteroidal medicines like Duvyzat (givinostat), Exondys 51 (eteplirsen), and Exondys 53 (golodirsen), which are Exon Skipping treatments. There are other coticosteriods like prednisone (prelone) and deflazacort (descort).

1) PREDNISONE (Prelone)

Prednisone is used to treat problems like allergies, infections, blood disorders.

USES: Reduces Inflamamtion, Suppresses the Immune system and slows muscle degeneration in muscular dystrophy. Controls Asthma and also COPD

FIGURE 5: PREDNISONE.

2) EFLAZOCORT(descort)

Deflazocort is commonly used to treat Duchenne Muscular Dystrophy.

USES: Slows the muscle weakness, manages the autoimmune diseases like Rheumatoid arthritis.

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