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Review Article

An Overview Of Muscular Dystrophy

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ABSTRACT

Muscular dystrophies are a diverse category of diseases characterized by pathologic changes in the muscle tissue upon biopsy. In clinical presentation, these conditions are defined by a gradual weakening of skeletal muscles. The X-linked recessive condition known as Duchenne Muscular Dystrophy is the most prevalent kind of muscular dystrophy. Typically manifesting in males and females between the ages of 40 and 60, distal muscular dystrophy mostly impacts the lower limbs, including the hands, feet, arms, and legs. The common symptom of congenital muscular dystrophy is the onset of muscle weakening during infancy or early childhood, usually before the age of two. Respiratory problems are a common outcome of most forms of MD, which impact the diaphragm and other muscles involved in breathing. Several MD subtypes are associated with cardiomyopathy or cardiac arrhythmias. gene transfer and gene correction treatments are mostly focused on this group of diseases.

INTRODUCTION

Muscular dystrophies are a diverse category of diseases characterized by pathologic changes in the muscle tissue upon biopsy. When applied in its most pathogenic definition, the word "dystrophy" describes significant and persistent myopathic alterations in muscle. At the end of the illness progression, fibrosis and fatty replacement are pathologic hallmarks of the majority of muscular dystrophies.[1] In terms of clinical presentation, these conditions are defined by a gradual weakening of skeletal muscles; however, there is a great deal of variation in terms of genetic and biochemical characteristics, the location of the affected muscles, the severity of cardiac and respiratory compromise, and the involvement of other organ systems like the central nervous system and eyes.[1,2] Age of start, severity, progression, prognosis, and effective treatment might vary greatly, even among people with the

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same disease and genetic abnormalities. Different types of muscular dystrophy have quite different disease mechanisms. Mutations in genes that code for proteins that attach to cell membranes, enzymes that regulate transcription, and proteases have been identified as causing disease. In the annals of contemporary molecular genetics, muscular dystrophies have a unique place. [3] The dystrophin-encoding Duchenne muscular dystrophy (DMD) gene was the first to be cloned using positional cloning technology. Our knowledge of the genetic spectrum of these disorders has grown over the last three decades thanks to persistent progress in gene mapping and identification; moreover, new diagnostic and exploratory techniques (like genome-scale sequencing) hold the potential to uncover more genes responsible for less common syndromes. We can expect a deeper comprehension of molecular pathogenesis with the use of these same techniques. Despite the lack of an authorized treatment for muscular dystrophy, research into novel molecular therapy classes, such as gene transfer and gene correction treatments, is mostly focused on this group of diseases. [4]

EPIDEMIOLOGY

The X-linked recessive condition known as Duchenne Muscular Dystrophy is the most prevalent kind of muscular dystrophy. The disorder, which affects 1 in 3,500 live births, is brought on by a mutation in the DMD gene, which causes the dystrophin protein to lose its function. Although muscular dystrophies are uncommon on an individual basis, they make up a substantial portion of the neuromuscular illness patients seen in both outpatient and inpatient settings.[5] Nearly 8.3 per 100,000 boys have Duchenne muscular dystrophy (DMD), the most prevalent hereditary muscle illness in childhood; about 7.3 per 100,000 boys have Becker muscular dystrophy, a relative of DMD. The two most frequent types of dystrophy in adulthood are myotonic dystrophy (affecting 10.6 per 100,000 individuals) and facioscapulohumeral dystrophy (affecting 3 per 100,000 people). There is a substantial geographical disparity in the incidence of congenital muscular dystrophies. Although Ullrich congenital muscular dystrophy is more frequent worldwide, a recessive founder mutation in Japan makes Fukuyama muscular dystrophy the most common kind. [6]

TYPES OF MUSCULAR DYSTROPHY

Dystrophy Distal Muscular Dystrophy

Typically manifesting in males and females between the ages of 40 and 60, distal muscular dystrophy mostly impacts the lower limbs, including the hands, feet, arms, and legs. In comparison to other types of muscular dystrophy, it typically impacts fewer muscles and develops at a slower rate. [7]

Duchenne Muscular Dystrophy

When it comes to pediatric muscular dystrophy, Duchenne is by far the most frequent kind. It develops quickly after its first appearance in early life, which affects exclusively guys. By the age of 12, the majority of children require a respirator for their breathing, and they still can't walk.[6,7] Because the condition affects the body's muscles, it can cause a person to have trouble walking, fall often, and eventually need a wheelchair. Scoliosis, a debilitating spinal curvature that can worsen breathing problems, and cardiomyopathy, a degeneration of the heart muscle, are complications of Duchenne muscular dystrophy.[8]

Myotonic Muscular Dystrophy

Among adults, myotonic muscular dystrophy is by far the most prevalent diagnosis. It has the same effect on males and females. Relaxation issues, weakening of the distal limbs (wrists, hands, etc.), cataracts, and gastrointestinal issues (constipation, diarrhea, etc.) are all symptoms of this kind of muscular dystrophy. Endocrine disorders,

including diabetes and thyroid issues, might potentially develop as a result.[9]

Oculopharyngeal Muscular Dystrophy

Muscular dystrophy of this kind weakens the muscles that control swallowing and vision in the face, neck, and eyelids. Muscular dystrophy of the oculopharynx often manifests in adults between the ages of 40 and 50.[10]

Becker Muscular Dystrophy

Despite sharing many similarities with Duchenne, Becker's muscular dystrophy is far less prevalent and has a considerably slower progression. Typically, a diagnosis is made between the ages of 11 and 25, and it primarily affects boys. Those affected with Becker muscular dystrophy, whether they are boys or men, gradually lose strength in their hip, thigh, pelvic, and shoulder muscles. Although the progression of Becker muscular dystrophy varies from person to person, the ailment usually has little impact on a person's life expectancy. [11]

Congenital Muscular Dystrophy

Approximately 30 different kinds of congenital muscular dystrophies impact both sexes equally. Both congenital and precocious onsets of the illness are possible. Issues with the joints, scoliosis, breathing, swallowing, convulsions, or eyesight are all possible complications for children born with congenital muscular dystrophy. Possible effects on the central nervous system include difficulties with vision and communication.[12]

Facioscapulohumeral Muscular Dystrophy

The muscles of the upper arm, shoulder blades, and face are impacted by facioscapulohumeral muscular dystrophy. This disorder often manifests in males and females before the age of 20, while it can manifest as late as the age of 40. The weakening of the muscles surrounding the eyes and mouth, as well as the shoulders, upper arms, and lower legs, is the initial symptom experienced by individuals with facioscapulohumeral muscular dystrophy. In due time, the muscles in your hips and abdominals will also start to weaken.[13]

Emery–Dreifuss Muscular Dystrophy

The Emery-Dreifuss muscular dystrophy syndrome primarily affects males in their adolescent years, however it can also strike females. Those who suffer from this disorder may find that their shin, upper arm, and shoulder muscles are weaker than usual. Muscle stiffness around a joint, known as contractures, can also develop, producing deformity in the afflicted area and restricting movement. A person's range of motion is limited because their spine becomes less flexible.[14]

Limb-Girdle Muscular Dystrophy

The muscles of the hips, shoulders, upper limbs, and legs are impacted by limb-girdle muscular dystrophy. This specific kind of muscular dystrophy is found in over 20 different types. It can start in persons as early as two years old and as late as forty years old. It has the same effect on males and females. [15]

CLINICAL FEATURES

Duchene muscular dystrophy symptoms can manifest as early as infancy and often manifest by the time a child is six years old. The condition manifests itself in pseudohypertrophy, frequent falls, and difficulties with motor abilities (running, leaping, and hopping) in youngsters whose weakness starts in the pelvis and upper legs. Fatigue, learning disabilities (IQ < 75), and potential intellectual impairment are further symptoms. The key difference between Becker and Duchenne muscular dystrophy is that patients with Becker may still walk at the age of 16 (and some even into old age), as Becker is a milder form of the disease. [16] Muscle cramps, trouble getting up off the floor, and walking on tiptoes are all possible symptoms. Signs and symptoms of facioscapulohumeral dystrophy, a kind of muscular dystrophy affecting the shoulders and face, include a lack of strength in the muscles that

close the eyes, reduced reflexes in the biceps and triceps, problems with eating and speaking, and even temporary hearing loss.[7,11] Myotonic muscular dystrophy is characterized by a reluctance to relax muscles after a sharp contraction. Other clinical signs include drooping eyelids and visual issues, swallowing difficulty, a long and thin face, weight loss, baldness, heart illness, testicular atrophy, and bad responses to anesthesia. It most commonly affects both men and women between the ages of 20 and 30. The common symptom of congenital muscular dystrophy is the onset of muscle weakening during infancy or early childhood, usually before the age of two. This category of disorders is known as muscular dystrophy. Babies with these problems may initially appear "floppy," and they may have trouble sitting up, rolling over, and walking later on. Rarer types of congenital muscular dystrophy might be associated with learning impairments or mental retardation.[7] Emery-Dreifuss muscular dystrophy manifests early on with symptoms such as walking on toes, trouble bending the elbows, and an increased risk of fainting owing to cardiac irregularities and tight Achilles tendons in the heels.[17] Deterioration of the shoulders and a stiff back are additional common symptoms. It is also possible to feel a slight wasting away of facial muscles. A dysfunction of voluntary muscles is the primary manifestation of limb-girdle muscular dystrophy and associated muscular dystrophies. Weakness in the muscles of the legs and hips is a common symptom for people with this illness. When you have shoulder weakness, it's hard to lift large things above your head. The advancement of limb girdles can be rapid or gradual, but after 20 years of diagnosis, the majority of individuals will become severely crippled. Weakness and atrophy of the distal muscles are symptoms of rare types of distal myopathies, which often do not affect other muscle groups. In contrast, oculopharyngeal muscular dystrophy affects other muscles in the head and limbs when increasing ptosis and dysphagia begin in late adulthood.[18]

PATHOPHYSIOLOGY

Contraction of Muscles Learning the basic mechanics of how muscles work is a prerequisite. Muscle tension is depicted in the sliding filament model as an outcome of filament contraction, which is facilitated by calcium, which, upon delivery from the sarcoplasmic reticulum, causes depolarization of the muscle. Tropomyosin can detach from the G-actin site when intracellular calcium binds to troponin C's anionic charge. After being exposed, a myosin head binds to the given G-actin site, creating a pivot that runs on ATP (adenosine triphosphate). The process of muscle shortening is transferred to the glycoprotein-rich cytoskeleton of the muscle cell using this pivot, which allows actin filaments to glide past myosin filaments. Dystrophin can only be found on the inner surface of the plasma membrane of muscle fibers; it links the cytoskeleton inside to the extracellular matrix through glycoproteins that cross the plasma membrane. This cytoskeletal protein provides the structural stability of a protein complex in cell membranes. In particular, inside a membrane-glycoprotein complex, dystrophin is responsible for anchoring the actin cytoskeleton to the basement membrane.[17,19]The proteins that make up this cytoskeletal structure include dystrophin and laminin. In the extracellular matrix (ECM), dystrophin binds to F-actin and β-dystroglycan before attaching to α-dystroglycan and laminin. Integrating the cytoskeleton with the extracellular matrix is dystrophin's job. Therefore, dystrophin alters tension transfer in a contracting muscle when it is not functioning properly. Typically, myosin and contractile actin proteins shorten, leading to weakened muscles and cell membrane damage progressively.[20] Upon muscle injury, creatine kinase leaks out of every cell and into the plasma at unusually high concentrations. Pseudohypertrophy, a hallmark of

muscular dystrophy, is the result of scar tissue development prompted by this inflammatory reaction to CK release. Muscles appear hypertrophied, however they are feeble due to a lack of functional contractile filaments in the tissue. From the time of fetal development forward, the deficiency is prevalent.[21] Inflammatory cells phagocytose injured muscle cells, leading to scarring and increased dysfunction. A protein network known as the dystrophin-glycoprotein complex seems to fortify the sarcolemma. When a node in the network goes down, it could cause other nodes to shift. For example, dystrophin could be destroyed together with other sarcoglycans if dystrophin is lost first. Muscle cell death occurs as a result of the membrane becoming weaker. Fat and connective tissue eventually replace skeletal muscle nearly entirely. Over time, the skeleton deforms, leading to a progressive loss of mobility. Fibrosis is a common occurrence in the cardiac and smooth muscles of the gastrointestinal tract. Inconsistent structural abnormalities of the brain are seen.[19.22]

DIAGNOSIS

A medical professional's hands-on examination is the first point for a diagnosis of MD. A thorough evaluation of the patient's medical history, including any history of muscular difficulties, as well as their family medical history (including any members with MD) will be conducted by the doctor. A person with MD may have abnormally high amounts of certain chemicals measured in their blood.[23] Additional testing may be necessary if these compounds are found at elevated levels, as they can indicate muscular weakness, injury, or illness. Potential examples of these include Damage to muscle fibers caused by the release of the enzyme serum creatine kinase into the circulation. One enzyme that aids in converting carbohydrates into energy is serum aldolase.[24] The oxygen-carrying and --storing protein myoglobin. A needle or tiny incision is used to remove a little sample of muscle tissue for a muscle biopsy. Medical professionals look for telltale signs of MD by microscopically examining the tissue. Genetic testing is often necessary to confirm abnormalities in genes in patients who are detected by muscle biopsy. Genetic testing to identify genes that are known to cause or be related to hereditary muscular illness. MD is one of several neuromuscular illnesses that DNA and enzyme tests may confirm.[22,24] Neurological tests to rule out other nervous system problems, uncover patterns of muscular weakness and wasting, test reflexes and coordination, and detect contractions. Diagnostic procedures for the heart include electrocardiograms (ECGs) and echocardiograms (Echos), which examine the heart's structural integrity and the force of the heartbeats. Irregular heartbeats and other cardiac issues are symptoms of certain types of MD. Evaluations of the patient's breathing and muscular strength during exercise, as well as the detection of any increases in the rates of certain indicators following activity.[21] Tests that capture photos of the inside of the body using radio waves magnetic fields (MRI) or sound waves (ultrasound) allow doctors to see things like muscle quality and mass as well as the amount of fat that has replaced muscle.[23]

COMPLICATIONS

Respiratory problems are a common outcome of most forms of MD, which impact the diaphragm and other muscles involved in breathing. Frequent and sometimes fatal lung infections are another complication of respiratory insufficiency. Different forms of muscular dystrophy manifest at very different ages, and breathing difficulties are no exception. Several MD subtypes are associated with cardiomyopathy or cardiac arrhythmias.[25] These complications often manifest in the latter stages of the disease and pose a serious risk to the patient's life. Among muscular dystrophies,

Emery-Dreifuss, LGMD, and DMD are the most frequently linked to heart problems. The vast majority of individuals will develop dilated cardiomyopathy. Arrhythmias of the ventricles can be seen in both DMD and BMD. Loss of fatty tissue in the RV and LV as well as problems with conduction are prevalent in LGMD. As a result of muscular dystrophy, scoliosis occurs in nearly all individuals.[6] The inability to stand or walk is often the first symptom of this condition, which develops as the muscles that support the spine become weak. Scoliosis can be present at birth in some cases of congenital MD. [21,25]

TREATMENT

Treatments for MD that are now on the market can alleviate or at least lessen the impact of symptoms. Treating or at least alleviating the symptoms of some forms of MD is an area of active investigation, and there is hope in this area thanks to ongoing research, including some gene-based therapies. Any therapies or drugs mentioned herein may not apply to or recommended for individuals suffering from MD. Seek advice from your doctor if you're confused about how to treat MD.[26] National Institute of Neurological Disorders and Stroke (NINDS) is in charge of NIH research on MD. Additional information on therapies can be found on the NINDS webpage on MD.[27]

MD treatments may include the following.

Physical Therapy

The key to maintaining strong, flexible muscles is to start physical therapy early on. Patients with MD may benefit from a regimen that includes both structured exercise and more passive stretching techniques.[28]

Respiratory Therapy

Weakened muscles from MD may impact breathing since the body uses muscles like the diaphragm to breathe. Until they have trouble coughing or have pneumonia from an infection, many MD patients may not know that their respiratory strength has diminished. Promptly following a diagnosis of MD, experts might provide therapies to forestall or postpone respiratory complications. Some MD patients may require a ventilator at some point in their disease's progression.[29]

Speech Therapy

Patients with myofascial weakness may find relief with speech therapy exercises designed to strengthen the muscles of the face and throat. One approach is to speak more slowly, another is to pause more often between breaths, and still, another is to use specialized communication aids. [30]

Occupational Therapy

Occupational therapy can assist people with MD in relearning motor skills that have been lost or in finding alternative methods to work with muscles that have been weakened, as physical capacities change. People with MD can learn to utilize wheelchairs, eating utensils, and personal things like hairbrushes and combs via occupational therapy.[25,29]

Surgery

Many people with multiple sclerosis (MD) eventually require surgical intervention to alleviate symptoms caused by the disease. People with myotonic MD may require cardiac procedures such as pacemaker implantation or surgical procedures to correct vision issues such as cataracts, a clouding of the eye's lens that prevents light from reaching the retina. Spinal curvature, or scoliosis, may necessitate surgical correction for some MD patients.[26.31]

Drug Therapy

Muscle deterioration can be postponed or MD symptoms reduced with the use of certain drugs. Among them, you may find that the United States has authorized the use of glucocorticoids like prednisone and deflazacort. Food and Drug Administration (FDA) for treating DMD in 2017. Research shows that prednisone when used

regularly, can improve respiratory function, strengthen muscles, and delay the deterioration of strength in multiple sclerosis.[15,13] Experimental trials using vamorolone, a novel glucocorticoid medication, are now underway in the treatment of Duchenne muscular dystrophy in boys. Researchers supported by the NICHD discovered that nandrolone alleviated symptoms of limb-girdle MD in animal models, and preliminary data indicated that the medication was just as effective as prednisone without the negative side effects. Seizure medication. [28] Medications used to treat epilepsy may also assist MD patients manage seizures and some types of muscular spasms. Drugs that inhibit the immune system. Immunosuppressant medications, generally prescribed for autoimmune disorders including dermatitis and lupus, may postpone the death of certain muscle cells in MD. Some forms of MD are linked to the use of beta-blockers, ACE inhibitors, and other drugs used to treat cardiac conditions such as high blood pressure and heart failure.[29]

Gene-Based Therapy

Researchers are actively working to find a way to restore a gene's protein-producing capabilities as a cure for MD, but there are currently very few options available. While some strategies aim to fix a single gene's function, others scan the entire genome.[30] The medicine eteplirsen is one example of a gene-based approach; it employs a technique known as "exon skipping" to generate functional dystrophin protein by omitting the problematic region of the gene. Despite being shorter than normal protein, exon skipping increases the amount of useable muscle protein.[23] Treating Duchenne muscular dystrophy (DMD) was authorized by the FDA in 2016, Goodison in 2019, and Viltolarsen in 2020. These therapies do not cure Duchenne muscular dystrophy and need weekly injections into an IV.[31] All three of these medications are still in the research phase, but there is hope that they may show therapeutic benefits beyond just increasing dystrophin production. Less than 25% of patients may get a positive response to these therapies due to the huge size of the DMD-causing gene. Investigation into alternative medications that enhance fetal dystrophin production and address other issues with protein instructions and synthesis is also continuing.[32

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