

Introduction

Muscular dystrophy is a group of inherited diseases. They cause muscles of the body to become gradually weaker. Duchenne muscular dystrophy is the most common type of muscular dystrophy. It mostly affects boys. There is no cure for it but there are ways to treat it.

This reference summary explains Duchenne muscular dystrophy. First is an overview of muscular dystrophy in general. Then the signs, causes, diagnosis and treatments for Duchenne Muscular Dystrophy are reviewed. At the end is a section about how to cope with muscular dystrophy.



What is Muscular Dystrophy?

Muscular dystrophy is a group of inherited diseases. They cause muscles of the body to become gradually weaker. There are more than 30 types of muscular dystrophy. Nine of them are more common than the rest. Duchenne muscular dystrophy is the most widespread of them all. It affects 1 out of every 3,500 baby boys.

Muscular dystrophy can start in childhood or adulthood. It can affect the muscles very slowly or very quickly. It is always hereditary. This summary is about Duchenne muscular dystrophy. Becker muscular dystrophy is similar to Duchenne.

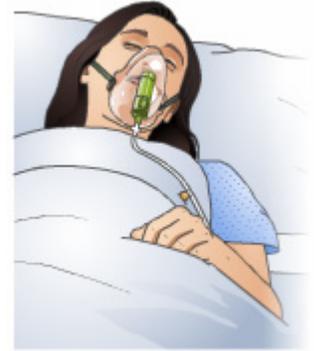
There is not just one way to treat muscular dystrophy. Treatments include:

- Physical therapy
- Assistive devices
- Medication
- Surgery



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Treatment does not cure muscular dystrophy. However, it can make the disease progress more slowly. This improves the person's quality of life. The various types of muscular dystrophy progress at different speeds. In the slowest type, the person could have a normal lifespan.



In a rapidly progressing type of muscular dystrophy, such as Duchenne, the lung muscles become weak early in life. This causes respiratory problems that usually lead to death in the late teens to early twenties.

Signs of DMD

Since Duchenne muscular dystrophy is genetic, babies are born with it. However, the symptoms start showing when a child is around 2 years old. The symptoms of Duchenne get worse fairly fast compared to other types of muscular dystrophy.

Around the age of 2, a boy with Duchenne muscular dystrophy will have some trouble keeping up with other kids when running. He will sort of waddle when he walks and runs. After a while, the boy will struggle going up stairs. For example, he will have to get both feet on a step before moving on to the next one. He may start pulling himself up using the railing.

Over the next few months, the boy will start walking on tiptoes and his spine will start to curve forward. At this stage, his muscles look healthy or may even be larger than normal, especially his calves.

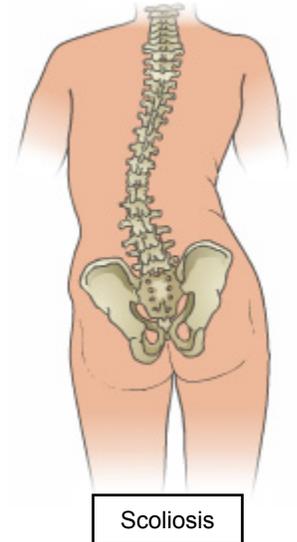
The child will start falling down a lot. He will need to pull himself up by hanging on to things. He might put his hands on the floor while standing up and then lift his upper body by walking his hands up his legs. He will soon have to get out of chairs the same way. Between the ages of 8 and 12, the muscles eventually become so weak that the child cannot walk. He will need a wheelchair.



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Even though the arms are weak in the early stages of the disease, they do not get weak as fast as the legs do. After getting a wheelchair, the boy's upper arms and shoulders become so weak that he cannot lift things. In time, he will not be able to lift his hand to his mouth. The boy will gradually lose strength in his fingers and hands, making intricate movements difficult.

As the muscles of the back weaken, the spine begins to curve. This is a condition called scoliosis. Over time, the spine may curve so much that the ribs rest on the hip bones. The tendons shrink at the hips, knees, ankles, shoulders and elbows. The legs will bend up against the stomach. The feet will point down and the arms will lock into the sides with bent elbows. These are called contractures.



As the muscles of the chest become weak, breathing is affected. The boy will not be able to breathe deeply or cough very well. At this stage, minor colds can cause infections and pneumonia.

As the chest muscles get weaker, the boy may need a respirator. Infections could make the lungs unable to provide oxygen to the body. This would cause death. In some cases, death occurs because the heart muscle becomes so weak that it just stops.

Duchenne muscular dystrophy does not affect the bladder, bowels or sexual functions. There is no unbearable pain associated with Duchenne muscular dystrophy. Contractures can cause painful sensations but they can be managed with medicine.

Causes

Duchenne muscular dystrophy mostly affects muscles that are attached to bones. A person can control these muscles. They are called skeletal, or voluntary, muscles.

Tendons attach muscles to bones. The Achilles tendon is one of the biggest tendons in the body. Muscles are made of long, specialized cells that form muscle fibers. When a muscle contracts, it becomes shorter. This pulls the tendon and moves the bone it is attached to.

Like all other cells in the body, muscle cells have walls, or membranes. To stay strong, cell membranes need a protein called dystrophin. A healthy person's body makes dystrophin but in people with Duchenne muscular dystrophy, the body does not make dystrophin. Without dystrophin, cell membranes become weak. This allows unwanted

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substances to leak in, which increases pressure inside cells. With too much pressure inside, muscle cells die. When muscle cells die, the muscles become swollen and damaged.

The body makes thousands of proteins that cause us to look and feel the way we do. These proteins are made according to genes on our chromosomes. In Duchenne muscular dystrophy the gene for making dystrophin is defective. Parents with defective genes may give them to their children. This is why Duchenne is hereditary, or genetic. So why is it mostly a male disease? We have to understand how genes are organized in the body to answer this question.

Genes, the hereditary part of the body, are found in chromosomes. The human body has 23 pairs of chromosomes. In fact, each cell of the body has these 23 pairs, or 46 chromosomes, in it. The only cells in the body that do not have the 23 pairs of chromosomes are sperm and eggs. Male sperm and female eggs only have 23 single chromosomes - 1 from each pair.

When a sperm fertilizes an egg, the 23 chromosomes from the man and the 23 chromosomes from the woman come together to make 23 pairs. The new 23 pairs of chromosomes is a whole new person.

On the chromosomes are genes. A person has 2 of each kind of gene - 1 from their mother and 1 from their father. For example, a baby has 2 eye color genes - 1 from its dad and 1 from its mom. If the dad gives a gene for brown eyes ("B") and the mom gives a gene for blue eyes ("b"), the baby will be Bb. Since a brown eye gene (B) is stronger than a blue eye gene (b), we say the B is dominant and the b is recessive. This means that the baby will have brown eyes since it has Bb.



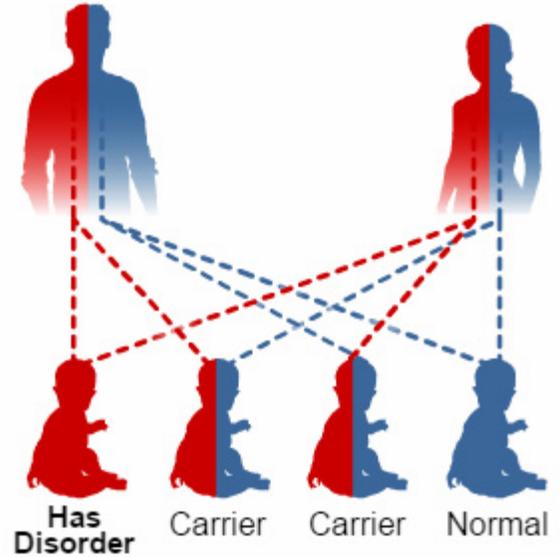
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In Duchenne muscular dystrophy, the defective gene is recessive. This means the good gene is dominant.

Let's say M is a normal dystrophin gene and m is a defective dystrophin gene. If at least one of them is an M, the person will not have Duchenne. M, the good gene, is stronger and dominant. It can make enough dystrophin for the muscles. A person who is Mm is a carrier. This means they carry the gene for muscular dystrophy, but do not have it.

Now going back to why muscular dystrophy mostly affects boys. Most pairs of chromosomes have 2 similarly shaped genes. The only pair that is NOT shaped the same is the sex chromosomes

- the ones that make us a boy or a girl. The 2 shapes of sex chromosomes are X and Y. They are called X and Y because that is what they look like under a microscope. Girls have 2 X chromosomes (XX) and boys have 1 X chromosome + 1 Y chromosome (XY).



When a male and a female create a baby, they each give 1 sex chromosome. If the man gives a Y, the child is a boy. The woman can only give an X. So, the man's chromosome determines the sex of the baby.

The dystrophin gene is only on X chromosomes. The following are all the combinations of sex chromosomes with dystrophin genes.

- $Y X^M$ = a male that does not have DMD
- $Y X^m$ = a male that has DMD
- $X^M X^m$ = a female that does not have DMD but is a carrier
- $X^m X^m$ = a female that has DMD

If a man with muscular dystrophy has a baby with a woman who is a carrier, the baby could be a girl with muscular dystrophy. In reality, this almost never happens since men with the disease have very poor health by the time they could have a child. That is why it is very rare for a girl to have Duchenne.

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DMD is inherited from women carriers. If a woman who is a carrier marries a healthy male, the chance that their baby boy will have DMD is 1 in 2, or 50%. The chance that their baby girl will be a carrier is also 1 in 2, or 50%. Since the chance of having a boy is always 50%, combined with the 50% chance that the boy will have DMD, the chance of this couple having a baby with DMD is 1 in 4, or 25%.

Genes rearrange themselves. These rearrangements are called mutations. Thousands of years ago, human ancestors had different mutations of the dystrophin gene, some of them were not harmful and others resulted in the inability of the body to produce a functioning dystrophin protein.

Since then the disease has been inherited and passed from generation to generation. However, it is theoretically possible for a regular dystrophin gene in a healthy person to mutate and cause muscular dystrophy in his or her descendants. Women thinking of having a baby, who have Duchenne muscular dystrophy in their family, may want to consider genetic counseling. Genetic tests can determine if a female is a carrier or not.

Diagnosis

When a person has symptoms of muscular dystrophy, a doctor needs to diagnose it. He or she must rule out other muscle and nerve diseases.

The doctor will take a medical history and do a physical examination. Extra tests may be needed to be sure about the diagnosis. These might include blood tests, muscle biopsy or genetic testing.

Blood tests look for special substances in the body, such as the enzyme creatine kinase or CK. High levels of this enzyme are associated with muscular dystrophy. In electromyography, the doctor measures the electrical activity generated by the muscles and nerves. Changes in electrical activity could suggest a muscle disease.

A thin-needle electrode is inserted through the skin into the muscle to be tested. The patient is asked to contract and relax the muscle as the machine records the electrical activity. Several muscles are tested to find the ones that are affected.



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Ultrasound takes pictures of structures inside the body. Echo waves are applied on the skin. As structures inside the body reflect the waves, the computer creates a picture of these structures. Ultrasound is painless. A muscle biopsy takes a small piece of muscle from the body to be examined under a microscope. Doctors called pathologists study it in a lab and look for muscular dystrophy.

Genetic testing is the analysis of a person's chromosomes. Medical technology can now zoom in on extremely small structures to find out if the dystrophin gene is defective. Genetic testing can also show whether a female is a carrier.

Treatment

There is no cure for Duchenne muscular dystrophy. However, there are ways to treat it that make it take longer to progress. Treatments also limit the complications, help the person stay independent and improve quality of life. Treatment options include physical therapy, occupational therapy, medications and surgery.

The aim of physical therapy is to keep the body as flexible, straight and mobile as possible. Stretching exercises can help postpone or even prevent contractures in men with Duchenne muscular dystrophy. Physical therapists teach patients how to move the joints and help keep tendons from shortening.

The shortening and stiffening of tendons is called contracture. Contractures are most common in the:

- Achilles tendons or the ankle
- Knee flexors or the hamstrings
- The hip flexors

Braces can prevent or delay contractures. These braces help keep the ankle bent upward and the knee straight so it is not bent backward. The names of these braces are ankle-foot orthosis (or AFO) and knee-ankle-foot orthosis (or KAFO). Hot baths, also called hydrotherapy, can also help maintain range of motion in joints.

Occupational therapy helps patients do things on their own. For example, patients learn how to dress, walk, use the bathroom, use a computer and other daily tasks. New skills have to be learned just to do basic things, as the disease gets worse.



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Several devices are available to help patients perform daily tasks. Such devices include:

- Raised toilet seats
- Shower chairs
- Desk tops
- Ramps
- Handrails
- Wheelchairs

Surgical options are available to relieve pain caused by contractures. For instance, the Achilles tendon in the heel can be lengthened to allow the foot to go back to its normal position. After this surgery, the child has to wear a cast and leg braces to keep the contracture from forming again.

After the boy is in a wheelchair, surgery may be recommended to prevent scoliosis. Steel rods can be implanted to strengthen the spine and prevent it from bending. A medication called eteplirsen has been approved by the FDA for treating some cases of Duchenne muscular dystrophy. This medication may slow down the progression of DMD. Ask your health care provider for more information about eteplirsen. Doctors may prescribe corticosteroid medications such as prednisone. These help preserve muscle strength and allow the person to walk a while longer.



Corticosteroids do have serious side effects, though. These include:

- Weight gain
- Acne
- Bones weakening
- Weak immune system

In its advanced stages, Duchenne muscular dystrophy affects the muscles of the lungs and chest, making it difficult to breathe and fight respiratory infections. That is why the patients should keep up with flu and pneumonia shots and check with the doctor if they notice any signs of respiratory problems. Using a face mask attached to a ventilation machine can help the person breathe. This treatment is called “non-invasive ventilation”.

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As DMD progresses it weakens the lung muscles. This causes the person to get less oxygen at night. Using a ventilator at night improves the quality of sleep and oxygenation. This in turn improves the quality of life during the day.

Coping

Parents and family members of people with Duchenne muscular dystrophy are faced with major physical, emotional and financial challenges.

Learning about the disease and what to expect is one aspect of being prepared for the challenges of the disease.

Joining a support group can also help patients, family members and friends cope. They can talk with other families who are facing similar experiences.

Ask your doctor about support groups in your community. Several online support groups exist. You can search for them online.



It is best to answer questions your child asks as honestly as possible. You should use simple terms that are easy to understand. Encourage your child to maintain a level of independence and do not pamper him. With the love and support you provide, also provide discipline and responsibility. Do not blame yourself for your son's disease. We all inherit thousands of diseases from our parents. Break up the responsibilities of caring for your son when possible, so you can have some time for yourself.

Summary

Duchenne muscular dystrophy is a genetic disease that results from a defective gene on the X chromosome. It affects mostly young boys and is the most common type of muscular dystrophy. The child is born with the disease. At about age 2, the signs start appearing which include difficulty getting up from a lying position and a waddling walk. As the child's muscles become weaker, he will be ultimately unable to walk on his own and will need a wheelchair.

Several therapies are available that will postpone the contractures of the joints and prolong the ability of the child to move on his own. These include physical therapy, occupational therapy, assistive devices, medications and surgery.

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A cure is not currently available for Duchenne muscular dystrophy. Over the past few years, research in muscular dystrophies has been encouraging and suggests the possibility of treating the disease in the near future by helping the body create dystrophin or a substance similar to it, which is needed to keep the muscles healthy.

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