

# DMD FAQ

## What is Duchenne muscular dystrophy?

**Duchenne muscular dystrophy (DMD)** is the most common form of muscular dystrophy. It is a genetic disorder, meaning it is caused by a change, or mutation, in an individual's DNA sequence. This change causes the muscles that control movement to get weaker over time.

Because the DMD gene is located on the X-chromosome, it primarily affects males, while females typically are carriers. DMD affects approximately 1 in 5,000 live male births.



## My child was diagnosed with DMD. What do I need to know?

A diagnosis of DMD can be overwhelming. Families are suddenly faced with the realities of life with a progressive disease. If your child is diagnosed with DMD, there are at least three things to know:

**1 You are not alone in this.** Extensive, multifaceted support is available for children and families. A good place to start is the **MDA Care Center** network and MDA's website, **mda.org**. It may be helpful to connect with other parents of children with DMD to find support and to share resources. With advancement in social media and technology, parents and/or their child may find comfort in making personal connections with other families of a child with DMD.

**2 Be proactive in seeking resources.** Ask your MDA Care Center providers what

financial or community resources your child may be eligible for, as there may be waitlists for certain services. Being proactive also includes informing your child's school of their diagnosis before their first school year. This allows you to collaborate with the school and your child's medical team to create an appropriate education plan.

**3 Ask questions.** Don't be afraid to tell your child's healthcare provider(s) if you do not understand something. It is important for parents and caregivers to communicate openly and honestly.

## How can I support my child's social development and mental health?

Support your child's social development by encouraging them to participate in social activities, social gatherings, summer camps, school clubs or organizations, and other activities. It is important for a child with DMD to have the opportunity to feel a sense of belonging.

Encouraging autonomy and independence is also crucial for social development. For example, allow your child to ask questions and answer them honestly in an age- and developmentally appropriate way. Also, include them in decision-making and give them the option to meet with their healthcare provider(s) alone at an appropriate age. Sometimes, a social worker at the MDA Care Center will ask to meet with your child alone, as this gives them a safe space to confide emotions, thoughts, and experiences that they may feel apprehensive about sharing in front of their parents or caregivers.

It is important for parents and caregivers to recognize symptoms of anxiety, depression, and other mental health disorders that may develop in their child (or in themselves). Encourage your child to share their emotions, worries, and stressors.

This allows them to express themselves and lets the parent in on what may be going on in their mind and body. It also is important for parents to model appropriate coping for their child, as children are like sponges. If marital or committed relationships between caregivers become stressed, consider engaging in couples counseling. Parents and caregivers may experience anticipatory anxiety and/or grief about their child's future, which is important to acknowledge and address.



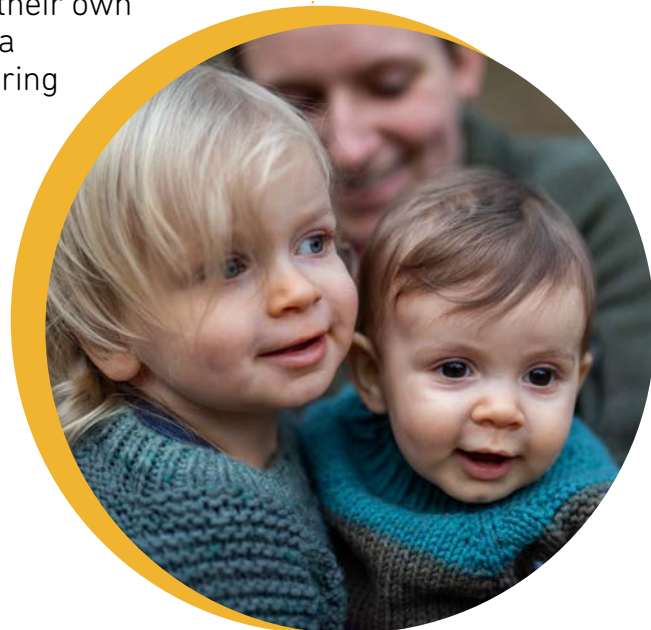
## What considerations should I make for siblings of a child living with DMD?

It is imperative for parents to give one-on-one attention to siblings of a child with DMD. Inevitably, the child with DMD will receive an immense amount of attention, and siblings may experience feelings of jealousy or loneliness. Siblings also may experience feelings of guilt for being a “healthy child,” anxiety about their sibling’s future, or helplessness in their sibling’s care. Having open conversations with each child will help parents to be aware and mindful of their children’s emotional well-being.

Some siblings become involved in caregiving for the

child with DMD, especially older siblings. However, it is important to encourage these children to develop their own identity and to find a balance between caring for their sibling and developing their own independence.

It also is important to talk with your child’s healthcare provider about whether siblings should be tested for DMD or for being a carrier.



## What are the current treatments for DMD?

Corticosteroids (such as prednisone) are commonly used in DMD to help preserve muscle strength and function, prevent scoliosis, and prolong the time that people with DMD can walk. It’s thought that steroids work, at least in part, by reducing inflammation. However, corticosteroids also cause unwanted side effects such as increased appetite, weight gain, loss of bone mass, and cataracts. **Emflaza** is the only corticosteroid that has been approved by the US Food and Drug Administration (FDA) for the treatment of DMD in individuals ages 2 and older.

Since 2016, the FDA has approved three exon-skipping drugs to treat DMD:

- ▶ **Exondys 51** targets a section of DNA called exon 51. It is approved by the FDA for treatment of individuals who have a confirmed mutation of the *DMD* gene that is amenable to exon 51 skipping and may help up to 13% of individuals with DMD.
- ▶ **Vyondys 53** and **Viltepso** both target a section of DNA called exon 53. They are approved by the FDA for treatment of individuals who have a confirmed mutation of the *DMD* gene that is amenable to exon 53 skipping and may help up to 8% of individuals with DMD.



## What's upcoming in clinical research for DMD?

Research in DMD is thriving. There is great hope in the neuromuscular disease community that the combination of advances in biology and genetics will result in better comprehensive care and a cure for DMD.

There are many active clinical trials for DMD in the following areas:

### ► **New steroid regimens:**

Because of new drugs and treatments, life expectancy for DMD has increased. Researchers are now looking into better steroid drugs, as well as regimens with existing steroids that will work better and reduce side effects in lifelong management of DMD.

### ► **Gene-targeted therapy:**

This is the most promising area of research for finding disease-modifying treatments for DMD, and potentially a

cure. Since DMD is caused by a gene mutation, this therapy aims to fix or alter the mutated gene. There are three main ideas being explored:

#### ■ **Shortened dystrophin:**

Viral vectors are used to reintroduce dystrophin to cells as a shortened version, called micro-dystrophin or mini-dystrophin. There are a number of clinical trials exploring this idea.

#### ■ **CRISPR/Cas 9 gene editing:** A gene editing

tool is used to alter the dystrophin in the body. This therapeutic idea still has to be researched in clinical trials, but animal trials have shown promise.

#### ■ **Exon skipping:** A mutated section of DNA is bypassed, or skipped, leaving shorter versions of the DNA block that can help retain and possibly improve muscle function. The FDA has approved three exon-skipping drugs to treat subsets of DMD.

## Understanding exon skipping

DMD is caused by mutations in the dystrophin gene that result in little or no production of a protein also called dystrophin, which is essential to muscle cells. Exon skipping is a strategy that coaxes cells to skip over a targeted exon (section of genetic code), allowing cells to create shortened but partially functional dystrophin protein.

To understand this better, think of the genetic code for a protein as a sentence. Cells have to read the genetic "sentence" in units of three "letters" each. For example:

**The mad cat ate the fat rat and the big bat.**

Errors can occur when a deletion mutation disrupts the three-letter reading pattern, creating "words" that don't make sense. When cells can't read the genetic sentence, they can't make functioning dystrophin protein.

**The mad cat ate the fat rat and the big bat.**

*deletion*

**The mat cat ate the tra tan dth ebi gba t.**

**Exon skipping** causes cells to skip a targeted section, creating a readable sentence.

**The mat cat ate the tra tan dth ebi gba t.**

*skipped exon*

**The mad cat ate the big bat.**

## How can I find the latest information about DMD clinical trials?

There are many avenues to find out about DMD clinical trials. Be sure to contact your healthcare provider to discuss trial options. The following is a list of resources where information on clinical trials can be found.

- MDA Care Centers
- MDA's Clinical Trial Finder
- Clinicaltrials.gov
- MDA's Quest Newsletter
- Patient advocacy groups
- Other DMD families

## Is healthy eating important for people with DMD?

Healthy eating is important to maintain a healthy weight, have good bone density, and prevent GI issues like constipation and acid reflux.

People with DMD are at risk for being both overweight and underweight across the spectrum of the disorder. Maintaining a healthy weight helps reduce other conditions associated with being overweight (diabetes, cardiovascular disease,

increased dependence on care providers) or underweight (increased weakness, skin breakdown).

Respiratory complications occur in both groups and can be life threatening. Individuals with DMD should keep their BMI (the measure of body fat based on height and weight) below 85% to avoid issues with obesity, but over 10% to avoid issues with undernutrition.

## What are some tips to prevent excessive weight gain for people with DMD?

Replace sugary beverages and juice with water and nonfat milk.



Eat more fiber-rich foods. Try these strategies:

- ▶ Eat three fruits per day as snacks.
- ▶ Fill half your plate with vegetables at lunch and dinner.
- ▶ Eat ½ cup legumes or beans at least four times per week.
- ▶ Make sure all your grains are whole, like whole-grain bread and whole-grain pasta.
- ▶ Eat a high-fiber cereal for breakfast — oatmeal or any cereal that has the word bran in it.

Eat breakfast every morning, and don't skip meals.

Use smaller plates and cups to decrease portion sizes.

Eat less of these salty foods:

- ▶ Pizza
- ▶ Burritos and tacos
- ▶ Sandwiches
- ▶ Breads and rolls
- ▶ Cold cuts and cured meats
- ▶ Canned soups
- ▶ Condiments like ketchup, soy sauce, and bouillon

Use your hand to determine one serving of different types of food:

- ▶ Two cupped hands: fresh vegetables, salads
- ▶ One cupped hand: nuts, seeds, dried fruit
- ▶ Fist: cooked rice, pasta, oatmeal, or vegetables; fresh or canned fruits, milk, yogurt
- ▶ Palm: fish, chicken, turkey, pork, or beef; slice of bread
- ▶ Thumb: oils, salad dressing, peanut butter, cheese



Choose healthier snacks:

- ▶ Unsalted pretzels
- ▶ Light buttered popcorn
- ▶ Oven-baked chips
- ▶ Fresh fruits and vegetables
- ▶ Nonfat yogurt

Eat all meals and snacks at the kitchen or dining room table with screens turned off.

Get a good night's sleep as often as possible.



Use the plate method to eat a well-balanced meal. [Choosemyplate.gov](https://www.choosemyplate.gov) has tons of healthy recipes.



Move your body in any way possible several times during the day.

## What are some tips to help people with DMD gain weight?

Eat three meals and two to three snacks daily.

Drink high-calorie oral supplements if food is not enough. You can purchase them at a store or make them at home as shakes and smoothies.

In some cases, a feeding tube may be needed to supplement intake of foods by mouth.

Add one of these calorie boosters to all foods:

- ▶ Butter
- ▶ Oil
- ▶ Peanut butter
- ▶ Eggs
- ▶ Heavy cream
- ▶ Avocado
- ▶ Cheese
- ▶ Sour cream
- ▶ Mayonnaise
- ▶ Yogurt



Eat large portion sizes.

## What can help someone with DMD in each ambulatory stage?

Early ambulatory phase (walking easily):

- ▶ Active movement, such as sports, martial arts, cycling, swimming, coloring/drawing
- ▶ Stretching four to six times a week
- ▶ Aquatic therapy



Late ambulatory phase (walking with difficulty):

- ▶ Nighttime splinting
- ▶ Adaptive clothing
- ▶ Adaptive shoes
- ▶ Clothing fasteners
- ▶ Aquatic therapy



Nonambulatory phase (unable to walk):

- ▶ Feeding and grooming tools
- ▶ Mobile arm support
- ▶ Smart home technology
- ▶ Video games
- ▶ Aquatic therapy

## What are some mobility options for people with DMD?

Low-tech mobility options:

- ▶ Adaptive stroller
- ▶ Manual wheelchair
- ▶ Transport chair



Lighter weight power options:

- ▶ Power scooter
- ▶ Power-assisted wheelchair
- ▶ Folding power wheelchair



Power wheelchair options:

- ▶ Drive type: front-, mid-, or rear-wheel drive
- ▶ Joystick operating system: standard, custom, mini
- ▶ Seating: linear, custom, hybrid

## What other types of durable medical equipment (DME) are available for people with DMD?

Bathroom equipment:

- ▶ Raised toilet seat
- ▶ Handheld urinal
- ▶ Shower chair/rolling commode
- ▶ Tub transfer bench
- ▶ Tub transfer system
- ▶ Tub lift



Lifts and lift systems:

- ▶ Hydraulic or electric
- ▶ Sit-to-stand
- ▶ Sit-to-stand with options
- ▶ Overhead lift
- ▶ Stair lift
- ▶ Elevator
- ▶ Wheelchair van lift



Standing systems:

- ▶ Prone standers
- ▶ Supine standers
- ▶ Sit-to-stand

## At what age does an individual with DMD transition from a pediatric to an adult clinic?

The age can vary depending on the MDA Care Center you visit. Generally, it is between ages 18 and 26.

## At what age does an individual with DMD become responsible for their own medical visits?

If the individual does not have a cognitive disability, at age 18, doctors will ask the individual for their permission to speak to and in front of their parents or caregivers. We know that parents and caregivers have been part of their child's life, and that won't change on their 18th

birthday. But, in the United States, 18 is the age of legal independence, and at this age a person becomes responsible for their medical care. For individuals who are not cognitively typical, the responsibility for medical decisions shifts to parents or a conservator.

## What is a conservatorship?

A conservatorship is a legal concept in which someone manages financial affairs and/or daily life decisions for another person due to their intellectual, mental, or physical limitations. The process to attain conservatorship varies from state to state. Physicians often can provide comments about an individual's cognitive capacity, ability to make informed decisions, and ability to perform activities of daily living during

a conservatorship hearing. Usually, the physician will need to fill out a form.

To get conservatorship, the person's parents or guardian must file paperwork with the court and then have a hearing. There is generally a cost to go through the conservatorship process. If you believe conservatorship is necessary, it is best to start the process several months to a year before the person with DMD turns 18.





## Is a person with DMD able to apply for Social Security Income (SSI)?

SSI is available for children and adults with DMD. The person with DMD or their parents must apply with the local SSI office. Your healthcare provider can provide records regarding the DMD diagnosis. Fill out a medical release form before your appointment with the Social Security office to allow your provider to send the medical information. Generally, you'll need a bank account to receive SSI deposits.

## How can an individual with DMD find a medical team once they age out of pediatric care?

It's important to find a primary neuromuscular care provider who is comfortable with complex care. Here are some tips:

- ▶ Look for an MDA Care Center that treats adults with neuromuscular diseases.
- ▶ Ask if you can stay at the same MDA Care Center. At some Care Centers, the pediatric and adult clinics are managed by the same team.
- ▶ Ask your pediatric team for recommendations. Most pediatric providers are familiar with the local adult care providers.
- ▶ Consider finding a provider at an academic center.



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To learn more about DMD and to view a DMD fact sheet for families, visit [mda.org/disease/duchenne-muscular-dystrophy](https://mda.org/disease/duchenne-muscular-dystrophy).



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