

New Diagnosis and Early Care Guide



Duchenne Muscular Dystrophy: A New Diagnosis and Early Care Guide

No one is prepared to receive the diagnosis of Duchenne. Most often, parents and caregivers have no prior knowledge of the disease, and information online can be very overwhelming. PPMD worked to create this guide to provide accurate information that is easier-to-digest, and will help you to navigate caring for your child while maintaining a happy, rewarding life. PPMD is here to help guide you and provide you with whatever help you need. As you read this, we suggest you go at your own pace and comfort level.

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DISCLAIMER: The information and advice published or made available in this booklet is not intended to replace the services of a medical provider (physician, nurse, nurse practitioner, etc.), nor does it constitute a doctor-patient relationship. This advice should be taken in conjunction with that of your medical provider, whom you should consult in all matters relating to your health. This is particularly true with respect to symptoms that may require diagnosis or medical attention. Any action on your part in response to the information provided in this booklet is at your own discretion. Although every effort has been taken to ensure the accuracy and completeness of the information contained in this booklet, accuracy cannot be guaranteed, and care in each situation must be individualized.

A Letter from our Founding President

Friends,

It is not easy to enter the world of Duchenne. You probably feel overwhelmed, lost, and alone. It's been many years since my sons were diagnosed, and there are still days when it is difficult for me to see through the tears, and days when the anger is overwhelming. Why my sons? What did they do to deserve this diagnosis? So many questions without answers.

None of us arrived here by choice. So how do we navigate this unknown territory when we don't have a map? How will we manage this journey, this new world?

In your new world, you will learn a foreign language — the language of Duchenne. It will include words you have never heard before, and you may stumble. But you will repeat them and repeat them until your new vocabulary becomes familiar.

You will become a partner in your child's care. Your knowledge about your child's medical care, needs, and possibilities will expand. And you will find power in this new world. Perhaps it's power and strength you never thought you had. The good news is we are in a new era of Duchenne — one where dozens of companies are invested in finding treatments to stop or slow the progression of Duchenne. There is reason for real hope, hope that I never dreamed of when my boys were diagnosed.

Now that you've entered this new world, you might begin to notice that everything looks different. And it's not just the world that's changed. Your priorities have shifted. The word Duchenne accompanies your every waking moment, enters every conversation, changes every relationship.

You are indeed on a journey, and this journey will lead you to places you never considered going. It will lead you to people you never expected meeting. Some of those places are great, and many of those people are good, caring people. They will offer their hands to keep you steady and help guide your decisions as you explore the world of Duchenne.

At first, it may feel like the sun will never shine again. But slowly, as you wake up each morning in this new world, you will put one foot in front of the other. However shakily, however uncertainly, you will regain some balance, and your strength will return.

Some people in your life may not understand your Duchenne journey. They may slowly drift away. Others will welcome your journey as their own. They will stay close and offer to help. Family, friends, doctors, nurses, and physical therapists will become partners and advocates — a vital support system. And, as in any new place, you will connect with these previously unknown people. They will come into your life and help in unexpected ways. These new friends will feel like family.

The world of Duchenne is a challenging place — full of frustration and surprise, obstacles and wonder, sadness and joy. In this new world, yes, you will find tears. But I can promise, too, that you will once again find your smile.

Remember, PPMD is here for you every step of the way.



Pat Furlong

Founding President, Parent Project Muscular Dystrophy



"Look for my thoughts in bubbles like this one throughout the book."

NOTES

SECTION 1

What is Duchenne
Muscular Dystrophy?



SECTION 1

What is Duchenne Muscular Dystrophy?

This section provides an in-depth overview of Duchenne muscular dystrophy. Because Duchenne is a genetic disorder, it is very complex and can be difficult to comprehend. PPMD worked to supply you with the most accurate and up-to-date information possible, but we recommend reading at your own pace.

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What is Duchenne muscular dystrophy?

Duchenne (“doo-shen”) **muscular dystrophy is the most common type of muscular dystrophy in children. It is a progressive disorder that causes muscles to become weaker over time.**

Duchenne muscular dystrophy is one of a spectrum of muscle diseases known as *dystrophinopathies* that can range from mild to severe in presentation. Dystrophinopathies occur when the *dystrophin* protein (found in the body’s muscle cells) has an altered structure or function, or is entirely absent from the body’s muscle cells. Becker muscular dystrophy is also a dystrophinopathy with many similarities to Duchenne. And while Becker can be variable, depending on when symptoms first present, typically the symptoms are milder in progression compared to Duchenne. PPMD’s work extends to both Duchenne and Becker, but for simplicity we primarily refer to Duchenne. You can think of Duchenne and Becker as different ends of the spectrum of dystrophinopathies.

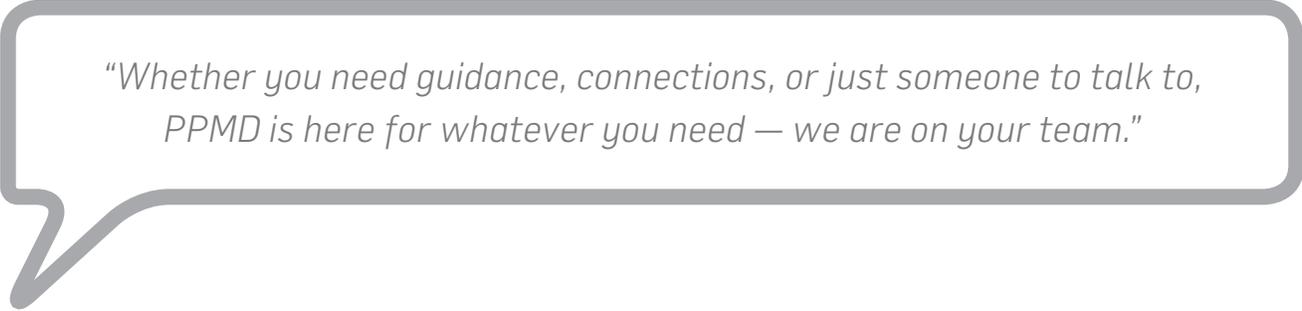
Duchenne is a genetic disorder caused by a change in the DNA (or gene) that codes for dystrophin (this is explained more in-depth later). Since the dystrophin gene is found on the X-chromosome, it primarily affects males. Females are typically known as *carriers* because they have one copy of the changed gene and one copy of the functioning gene. Carrier females most often do not have outward symptoms of dystrophinopathy. However, some carrier females can present with varying ranges of physical symptoms and are considered to have a dystrophinopathy as well, but are usually termed *manifesting carriers*. Affected adult women and young girls are discussed further in Section 3 of this guide.

Although no cure is available at this time, there are medical treatments that can help slow the progression of Duchenne. In addition, there are over 40 companies currently working towards new and novel therapies targeting this disease. People living with Duchenne can lead autonomous, happy, and fulfilling lives.

How common is Duchenne?

About one out of every 5,000 boys is born with Duchenne, which totals to about 20,000 babies per year worldwide, across all races and ethnicities. PPMD estimates that there are about 12,000–15,000 young men, as well as a number of affected young women, living with Duchenne in the United States.

In the US, a *rare disease* is defined as a condition that affects fewer than 200,000 people. Although Duchenne is considered rare, there is a huge active community at your fingertips working to end Duchenne.



“Whether you need guidance, connections, or just someone to talk to, PPMD is here for whatever you need — we are on your team.”

What causes the symptoms of Duchenne?

Duchenne is caused by changes in the piece of DNA, or gene, that code or instruct the body to make muscle protein called dystrophin. Dystrophin is in every single muscle cell in our bodies. Dystrophin acts as the glue that holds muscles together and the “shock absorber” that allows muscles to contract and relax without being damaged.

Without dystrophin, muscles are not able to function or repair themselves properly. As muscles are used for normal day-to-day activity, tiny tears are created in the muscle cells, which cause creatine kinase (CK) to leak out of the cells and results in high levels in the blood. The muscles cannot properly repair themselves, so the damaged muscle is replaced by fat and scar tissue. This causes the person with Duchenne to lose muscle function and strength.

Duchenne is a condition that progresses very slowly over time. The first symptoms are often subtle, such as delays in crawling, walking, and/or speech. Parents may or may not notice these delays, and primary care providers often attribute these delays to something else.

Clear symptoms typically develop between the ages of 2 and 5 years, which is typically when the diagnosis is made. Muscle weakness follows a *proximal to distal* pattern, meaning muscles near the trunk, such as the hips and shoulders, will be affected before those of the arms and legs. This is why you may notice your child having difficulty walking, running, or climbing stairs. When asked to get off the floor, your child will often use the *Gower's Maneuver*, or using their arms to “walk” up their legs to a standing position. They may also appear clumsy and fall a lot, and their calves may appear enlarged or overdeveloped (called pseudohypertrophy, which is replacement of the calf muscle with fat and scar tissue). This is when a primary care provider will likely order a creatine kinase (CK) blood test to check for muscular dystrophy.

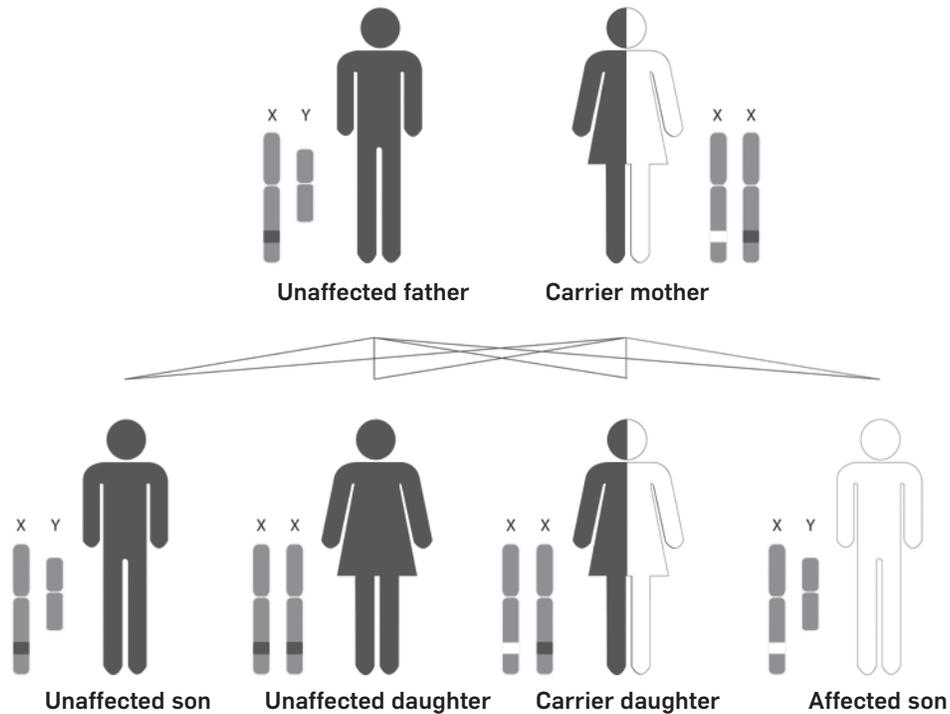
Why does my child have Duchenne?

Duchenne is a genetic disorder, which means it is caused by a change in the DNA sequence of a gene. Genes house the instructions for how to make proteins, and proteins are the building blocks of the body. In Duchenne, there is a change (often called a *mutation* or *variant*), in the gene that codes for the dystrophin protein, so the dystrophin protein is not made properly.

Duchenne is an *X-linked disorder*, which means the dystrophin gene is on the X-chromosome. It may be helpful to review some basic genetics. Every person has 23 pairs of chromosomes, which contain all of their genes and determine their sex. One of each pair of chromosomes are inherited from their mother, and the other from their father. Females always have two X-chromosomes, so mothers always pass down an X-chromosome to their child. Males have one X-chromosome and one Y-chromosome, so fathers can pass down either an X or Y-chromosome, which will ultimately determine the sex of the baby. Boys always inherit their X-chromosome from their mother and their Y-chromosome from their father, whereas girls inherit two X-chromosomes, one from each parent.

Since a boy only has one X-chromosome, if that chromosome contains the Duchenne gene mutation, he will have Duchenne. However, since girls have two X-chromosomes, if one of them has the Duchenne gene mutation, they are usually not affected because they have a second back-up copy of the gene on their other X-chromosome. Females with one X-chromosome with the Duchenne gene mutation are known as carriers of Duchenne. Carriers have the ability to pass on the mutated Duchenne gene to their children.

X-linked Recessive Inheritance



Since Duchenne is genetic, was it inherited from me?

You may have someone in your family who has or had Duchenne. As a genetic disorder, Duchenne can be passed down from a parent to a child, so it could already be in your family's genes. Many mothers who carry the Duchenne gene often do not know that they are carriers. Approximately two thirds ($\frac{2}{3}$) of newly diagnosed boys with Duchenne cases have inherited the gene from their carrier mother.

Duchenne can also occur even if no one in your family has had it before. The Duchenne gene is very large, which increases the incidence of mutation during pregnancy. This is why approximately one third ($\frac{1}{3}$) of Duchenne cases are caused by a *spontaneous mutation*, or a new change in the DNA that is not inherited from the mother. The only way to determine if a child with Duchenne has inherited the mutation or has a new mutation is for the mother to have carrier testing. Read more about carrier testing below.

Genetic testing

Genetic testing (also known as DNA testing) is the process of examining a person's DNA. In Duchenne, the dystrophin gene is analyzed for any changes, or mutations, that would prevent the gene from working properly. Genetic testing is typically performed on a blood or saliva sample.

Genetic testing is always necessary and should be offered to every family when a diagnosis of Duchenne is suspected. Different types of genetic tests are able to provide specific information about the change or mutation in the DNA. It is important to know the specific genetic change or mutation in the DNA for several reasons.

Why is genetic testing important?

In today's world where diagnoses are made earlier, care and management are better, and new therapies are on the horizon, genetic testing is more important than ever before. Some of the most important reasons for genetic testing include:

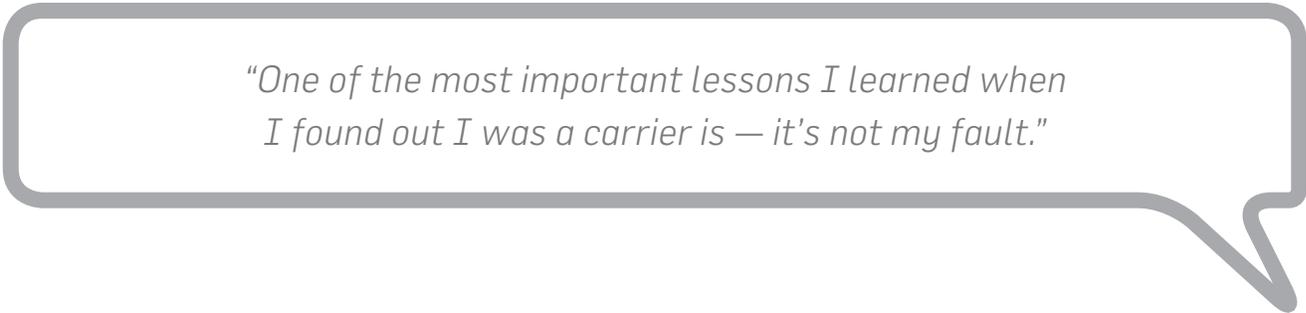
1. **To confirm your diagnosis:** Genetic testing will confirm if you have Duchenne, or if you may have another type of muscular dystrophy that shares some of the same features as Duchenne.
2. **To enable testing of family members:** Once the specific mutation in a family is known, then other family members can be tested to determine if they are carriers of the gene mutation. Genetic testing is the best method for performing accurate carrier testing.
3. **To determine what mutation-specific therapies may benefit you:** Many therapies in development and/or approved for Duchenne are "mutation-specific," meaning they will only benefit individuals with certain genetic mutations. You must know your particular mutation in order to participate in a clinical trial and to access any current or future mutation-specific therapies. If you have never had genetic testing or if you need repeated genetic testing, remember that PPMD's Decode Duchenne program provides free genetic testing to eligible individuals.

Decode Duchenne: PPMD's free genetic testing program

PPMD supports a genetic testing program called Decode Duchenne that offers free genetic testing to eligible individuals. We believe everyone affected by Duchenne needs and deserves genetic testing. This includes children who are suspected to have Duchenne as well as females who are potential carriers of Duchenne. The testing process is fast and easy, and includes interpretation of results and genetic counseling. Please visit parentprojectmd.org/decode for more information regarding this program.

Does the rest of my family need to be tested?

Mothers who have a child with Duchenne have a two thirds chance ($\frac{2}{3}$ or 66%) of being a carrier of Duchenne. Mothers of children with Duchenne should have genetic testing as well, and this is called carrier testing. This is important because carriers of a mutated Duchenne gene have an increased chance of having children with Duchenne and their own health may also be at risk. By being aware of their own carrier status, mothers can make informed decisions regarding future pregnancies and be aware of potential health implications of being a carrier.



"One of the most important lessons I learned when I found out I was a carrier is — it's not my fault."

Most female carriers of Duchenne are unaware that they carry a mutated Duchenne gene until they have a child with Duchenne. The genetic mutation may have existed in the family for generations without anyone knowing, especially if there were no previous male children. If a woman is found to be a carrier, it is important to inform any female relatives on the maternal side (sisters, cousins, aunts, etc.) as they too may be carriers. To learn more about which relatives in your family should have genetic testing, it is helpful to speak with a genetic counselor.

Additionally, we are continuously learning more about the effects of Duchenne genetic mutations on female carriers. It is important that female carriers inform their care providers about their carrier status or possible carrier status. 10–50% of carriers of Duchenne will develop cardiac changes, and should have regular heart evaluations. Additionally, a small percentage (10–20%) of female carriers of Duchenne are at risk for developing mild muscle weakness. Care considerations for female carriers is discussed in Section 3.

The decision to have siblings tested varies from family to family. If the mother of the child with Duchenne is found to be a carrier, this may lead the family to elect to have their other children tested, depending on factors such as age (especially if unable to legally consent), potential symptoms, and the family's personal beliefs. This is a topic that should be discussed with a genetic counselor.

Genetic counseling

For families, genetic counseling can offer several benefits. A genetic counselor can further explain the causes of muscular dystrophy and discuss and facilitate diagnostic and genetic testing options. In addition to focusing on the child with Duchenne, the genetic counselor can also explore and address family concerns.

In considering genetic testing, genetic counselors work with families and their healthcare providers to determine the best testing strategy. This will vary from family to family, depending on their specific testing goals and what testing has already been done, as well as any insurance or financial barriers. After testing has been completed, genetic counselors can help families and healthcare professionals understand the meaning of the results.

If you have a child with Duchenne and would like to receive genetic counseling, you can ask your child's primary care doctor or neurologist for a referral. If you have already found a pediatric neuromuscular care team, genetic counseling is often available within that clinic.

PPMD has genetic counselors who oversee The Duchenne Registry and the Decode Duchenne program, and they are available to help you via phone or email. They can discuss your genetic test results, give general guidance about testing other family members, and speak with you about Duchenne clinical trials. They can be reached at 888-520-8675 or by email at coordinator@duchenneregistry.org

The Duchenne Registry

The Duchenne Registry is PPMD's patient-reported registry for Duchenne and Becker muscular dystrophy. This international registry began in 2007 and has over 5,000 members. New in 2019 is The Duchenne Registry mobile app, which will make the collection of data even easier and more convenient for families. The goals of the registry are to collect data that will be used by researchers to improve care and treatments for Duchenne, and to connect patients and families with actively recruiting clinical trials and research studies. By joining The Duchenne Registry, you are strengthening the power of this incredible resource and helping in our fight to end Duchenne. Learn more at duchenneregistry.org

SECTION 2

Adjusting to the Diagnosis & Finding Support



SECTION 2

Adjusting to the Diagnosis

After receiving the diagnosis of Duchenne, parents often struggle with how to share this information with their children, other family members, and friends. While keeping this information to yourself can be incredibly burdensome, sharing the news of the diagnosis may help to relieve some of this stress. It will also begin to build a support system around you and your family. In addition, starting to talk with young children about their diagnosis will give them the information and tools they will need in the future to build a community of support that will enable them to thrive and achieve their goals, despite the challenges with their diagnosis.

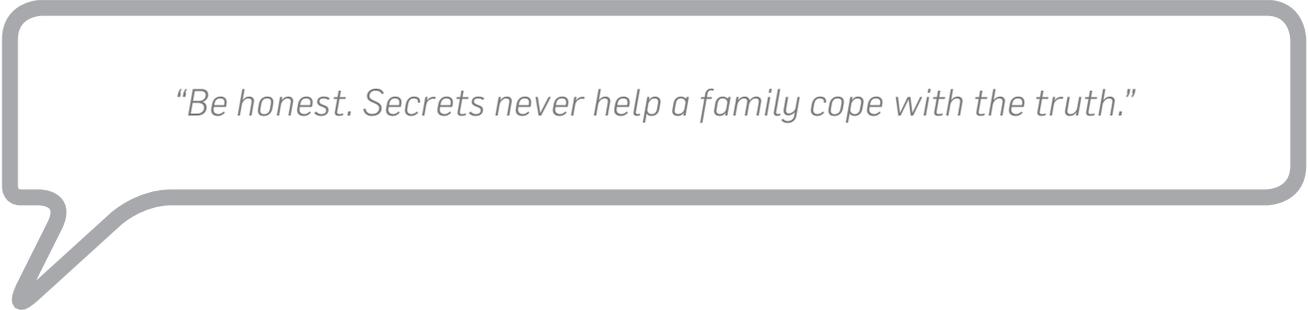
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Talking to your child with Duchenne about their diagnosis

Naturally, parents want to protect their children AND shield them from anything scary or sad. This can lead to parents delaying or avoiding talking about Duchenne altogether. They may be afraid of becoming emotional, saying something that causes distress or “makes things worse,” or of not having all the answers. However, even young children sense when something is not right. Silence is rarely the best approach. It implies the situation or illness is too horrible to think or talk about. It tells the child that parents don't trust them to be able to deal with the situation. It communicates the message that “we don't talk about difficult things”.

If children sense that whatever is happening to them is causing their parents pain, then they may be reluctant to ask questions. They may attempt to protect their parents by not asking questions or saying things about what is happening. The reality is often that talking about Duchenne is usually more distressing for parents than it is for the child with Duchenne.

The first step is to realize that children can usually cope with questions and difficulties better than we think. Rather than setting up a situation where they are afraid to ask questions, we need to create a space where they feel comfortable and safe to ask both the easy and hard questions about their diagnosis.



“Be honest. Secrets never help a family cope with the truth.”

Children need accurate information to help them cope and thrive with their diagnosis of Duchenne. Starting the discussion with them ensures that what they learn is accurate and up-to-date, and will also prevent them from jumping to the wrong conclusions or learning faulty information from other sources.

Talking about Duchenne is also the first step in helping them develop independence in the future. It gives them the words and know-how to advocate for themselves, take charge of their medical care, and educate others about Duchenne in the future. Remember, our job as parents is not to protect our children from all of life's difficulties, but to equip them with the skills they need to succeed **DESPITE** life's difficulties. Understand when you go into the conversation that your anxiety may be higher than theirs and that is okay.

When do I talk to my child about their diagnosis?

There is never a “good” time to talk about Duchenne, but typically earlier is better. Most children can start to understand a basic explanation by the time their mental development is at a 3 or 4 year old level. Starting early makes conversations more “normal” over time, because your child won't remember a time when you didn't talk about Duchenne. The longer you wait, the more awkward or difficult things become, and the more misunderstanding or misinformation you may need to correct. What you say to your children about their diagnosis depends on their ages and levels of understanding. It will be an ongoing conversation that **EVOLVES** over time; the first time you talk about Duchenne is only starting the process.

How do I talk about Duchenne with my child?

Parents often worry about what to say when it comes to Duchenne. You may be surprised about how much your young child already knows. You can first start by asking questions such as “Do you know why we go see Dr. ____?” or “Have you heard anyone talk about your muscles, and what did they say about them?” This way, you can understand how they perceive Duchenne, and if there is any information you need to correct.

With younger children, it is important to offer simple explanations that they will understand. Connecting explanations to their day-to-day experience will make the most sense to them. This can be discussed “in the moment” as well, rather than in a planned conversation. An example would be while at the park explaining “your running is not as fast because when you have Duchenne your muscles are different and get tired more quickly” or at home explaining, “We do this stretching because your muscles are different.”

Use words that are at their level of understanding, and avoid medical terms or scientific jargon (which can be gradually introduced over time). Use the term “Duchenne” or “DMD” from the beginning, even though your child does not know what that means. By using the correct term during these conversations, you will help them learn what Duchenne or DMD is.

For children of all ages, ask them to explain in their own words what you have told them. This allows you to give reminders about information you discussed, and make sure there is no confusion. It is also important to try to be mindful of and avoid using any negative descriptions such as “bad” or “terrible,” and words like “disease,” which children associate with being contagious.

Don't be afraid of saying the “wrong” thing. At some point as parents, all of us say something that is “wrong,” and discussions about Duchenne are no different. By talking about things often, you can always fix any mistakes. The risks of NOT talking about Duchenne far outweigh this.

The following are key points that may be helpful to cover during these conversations:

- Your child did not get Duchenne because he is bad or did anything wrong.
- Sometimes you are sad or emotional about your child having Duchenne, but it doesn't mean you are upset with the child. They did nothing wrong.
- Your child can't spread it to someone (like spreading a cold at school).
- Your child's Duchenne will not go away (like a cold does).
- Your family will work hard to keep your child as healthy as possible by going to doctor's appointments, physical therapy, etc. Scientists are working to find medicines and other therapies to help people who have Duchenne. You want to help them understand that there are hopeful things happening and that doctors are helping them to feel the best they can right now.
- Sometimes when you have Duchenne, you may need to do things in a different way.
- Don't avoid or discourage difficult questions your child may ask about Duchenne (“Am I going to die from Duchenne?”), because even young children are good at picking up on deception and “non-answers.” At the same time, you don't have to try to predict the future and every possible thing that might happen. It is okay to acknowledge the challenges of Duchenne, but balance this with hope and a positive message, and remind them that you will work together to solve problems.

Some children may become sad or upset when you talk to them about Duchenne. Although this is difficult for parents, it is a normal and temporary reaction. During these difficult times, it is important to validate your child's feelings and provide emotional support.

Balance times of hard conversations with fun times of bonding. Go for ice cream, play a family board game — you want them to associate the conversations with times of connection with you so that they feel safe talking to you again.

Talking to siblings about Duchenne

A diagnosis of Duchenne may alter the family dynamic due to the attention and focus on the child with Duchenne. However, parents, relatives, and friends must not forget about the other siblings who are struggling with these changes as well. Remember that each of your children deserves, as much as possible, a happy, and normal childhood. Some siblings are afraid to have "problems" or share them with parents because they are afraid it will add to the parent's stress. Make sure you check in with them and let them know they can also share their struggles with you.

Similar to the child with Duchenne, it is a good idea to talk to siblings as early as possible. Allow siblings to ask questions, even when you are at doctor's appointments for your child with Duchenne. Siblings can also help you take care of your child with Duchenne, but be intentional about it not becoming a job. We suggest thanking them for their help and avoid phrases such as "I couldn't do it without you." Make it a priority to spend individual time with siblings to promote their own interests and development.

Remember that for siblings, it is not all negative. The dynamic of Duchenne in the family often makes the siblings more compassionate toward others, better problem solvers, and loving caregivers. Siblings often thrive in a Duchenne family when they know that they are heard, cared for, and are an integral part of family life.



How do I talk to my friends and family?

One of your first thoughts may be: “how will I tell my friends and family my child has Duchenne?” These conversations may be very difficult and upsetting for you and the people close to you. Although this conversation may be daunting, it is important to inform your friends and family of your son's diagnosis so they have the opportunity to act as a support system for your family.

Keep in mind that everyone's reaction will vary, and that ways of coping with such serious news differ from person to person. For many, anger, shock, and sadness are the prevailing feelings. It will take time for families to come to terms with the diagnosis. Knowing that they have a solid support system in place to help them face fears with courage and optimism can often reduce the anxiety and stress associated with these feelings. Also remember that not everyone will “get it.” Those people who do “get it” will be your greatest lifelines in this journey.

If you find it hard to communicate all of your wishes regarding your Duchenne child to family members, it may be easier to put it in writing. For example, if you want them to follow a certain type of diet when the Duchenne child is in their care, write it down and be specific. Giving them a written plan for various aspects of your child's care may help in certain family situations.

Supporting yourself & relationships

All mothers and fathers want to protect their children, but a Duchenne diagnosis can leave you feeling helpless and isolated. The most important thing to remember when facing the challenges of Duchenne is that you are not alone. Keep in mind:

- Try not to neglect your relationships — especially your partner — as you begin to adjust to this diagnosis. Spending time with each other is critical to helping you to adapt to this new normal as a team.
- There are many professional counselors and other therapy options available to help families cope with the demands and challenges that accompany the diagnosis of Duchenne.
- Anger, guilt, fear, and sadness are common feelings associated with the diagnosis of Duchenne. Do not feel guilty for having these feelings.
- The course of coping is an ongoing process, filled with both good days and bad. Give each other permission and space to have good and bad days.
- Asking for help or advice should not be considered a weakness or failure; it's a positive and constructive action that can help everyone in the family deal with the issues at hand.

Remember that you and your partner (and/or your child's other parent) may adjust differently to the diagnosis. Many times, acute grief reactions occur that may last from several months to years. The experience typically includes overwhelming emotions of shock, denial or suspicion, despair, anger, sadness/depression, anxiety, and guilt. Family members may experience this at different rates, resulting in conflict. Keep communication honest and open and understand that adjusting to the diagnosis can be different for the both of you.

How do I address this with my child's school?

PPMD wants to make sure that families within our Duchenne community are equipped with as many tools as possible to ensure a smooth start to the school year. It's hard enough just to figure out the secret code to get through the front entrance, let alone to have to navigate considerations around 504 Plans and IEPs (individualized education programs), accessibility, and what information to provide to whom about your child's medical care.

For these reasons, PPMD has developed a resource called Education Matters, which is a guide for both families and teachers to help children living with Duchenne be set up for success at school. We also have many classroom resources on our website (parentprojectmd.org/classroom).

Talking to classmates and peers

When your child's peers do not understand another child's medical condition, they are more likely to tease, say hurtful things without meaning to, or ask repetitive questions in an intrusive manner. Research shows that when peers are given information about a child's condition, they are less likely to engage in teasing, are more likely to be accepting/inclusive, and also often are protective of and "stick-up" for the child. It is also a great opportunity to help your child's peers think of ways they can help your child such as help them to carry school materials, or being careful when playing.

Before talking to your child's peers, notify your child that you will be doing so, and ask if they want to be involved. Sometimes, children like to talk about themselves and answer questions their friends may have. If your child is shy, they may prefer to be absent from class when the discussion happens.

Similar to talking to your child, it is important to give basic information only with simple explanations. Explain that Duchenne is something they were born with, it is not their fault, and it is not contagious to other kids. It is helpful to give examples of how Duchenne impacts daily activities, for example, needing to take rests if they get too tired. Visual aids or demonstrations can also help explain what happens to the muscles in Duchenne. We have a collection of classroom resources for teaching peers about Duchenne on our website (parentprojectmd.org/classroom).

"Keep the lines of communication open."

Continue to live your life

Remember, in every family communication is important. In a family dealing with a diagnosis of Duchenne, communication is CRITICAL. The goal is not to just survive, but to thrive and have a full and happy life even while you are facing a devastating diagnosis. There is hope, fun and many days of laughter and love to be had, and one of your main goals as a parent is to keep the lines of communication open with your family so that those days happen often.

The above information was provided by James Poysky, PhD, a clinical psychologist, and also a parent of a son with Duchenne, with reference to resources created by David Shoenfeld, MD, a developmental-behavior pediatrician specializing in trauma and adjustment.

Get support

After talking to your family and friends about Duchenne, you may be interested in connecting with others who are affected by Duchenne as well. PPMD would love for you to become a part of our community! Below are some ways to meet other families, and to become involved in one of our many programs.

Find a family in your state

PPMD has an extensive grassroots network of families who are actively advocating for better care, funding important research, and creating global awareness of Duchenne. PPMD families refuse to accept "there is nothing you can do" as an answer to the diagnosis. Many states are part of our PPMD Connect program, which are volunteer-lead outreach groups that connect families to resources and each other. Visit our PPMD Connect webpage (parentprojectmd.org/connect) to learn if your state has an active group. To be connected to a coordinator directly via email please contact connect@parentprojectmd.org.

Join an online community

- Register with PPMD (parentprojectmd.org/newlydiagnosed) as a newly diagnosed family. This allows us to keep you updated with current efforts, new updates in care or research, as well as invite you to events. You will receive regular newsletters informing you of all of these topics and more.
- PPMD has a private Community Forum (community.parentprojectmd.org) that allows families all over the world to connect with each other to chat, organize, and share stories, news, and information.
- Facebook has a large number of active groups in the Duchenne community. A simple search under "Duchenne" and you will find an extensive list. Be advised that most of these groups are independently monitored, and depending on your level of comfort you may want to avoid over-saturation. The PPMD Facebook page (facebook.com/parentprojectmd) is a great place to receive daily Duchenne updates on research, care, and the community.



Attend PPMD's Annual Conference

PPMD's Annual Conference is a unique convergence of industry partners, scientific leaders, medical providers, people living with Duchenne, and their families. This exceptional meeting has grown to be recognized worldwide as the foremost Duchenne muscular dystrophy meeting. More than that, it is a way for families affected by Duchenne to connect with each other to build support networks and to realize no one is on their own in the fight to end Duchenne.

Each year, PPMD invites newly diagnosed families to attend the conference for free. For some families, the diagnosis is still too raw for them to attend a meeting such as this — and that's okay. However, when families are ready to take this step, they appreciate the opportunity to connect with the Duchenne community and learn all they can about care, advocacy and research, and most are glad they decided to attend. The community will be there for you, to hold you up when you are ready. Visit our conference application webpage (parentprojectmd.org/conference) to apply today.

Attend a PPMD End Duchenne Tour Stop

In an effort to reach every single family facing a Duchenne diagnosis in the U.S., PPMD launched a multi-year experience called the End Duchenne Tour. Combining each of the pillars that make up PPMD's mission, the End Duchenne Tour brings updates on research, advocacy, and care to cities across the country, featuring a roster of leading experts in the Duchenne space. Similar to PPMD's Annual Conference, End Duchenne Tour stops are a great opportunity to meet other families affected by Duchenne in your area.

To see if the End Duchenne Tour is stopping at a location near you, please visit parentprojectmd.org/tour.

Get involved

PPMD offers many opportunities to become involved in the Duchenne community. We understand not all families choose to become involved in our efforts, but many find them empowering and a way to cope. Getting involved is also a great way to meet families in your area.

Annual Conference

As mentioned above in the “Get Support” section, PPMD hosts an Annual Conference. Every summer, the conference is rotated to a different region of the country hoping to attract attendance nationwide. This is a unique opportunity to learn more about Duchenne care, research, and advocacy as well as network with families, industry partners, and clinicians from all over the country.

Advocacy Conference

Much of the progress that has been made in Duchenne care, research, and therapy development is a direct result of the federal advocacy of Duchenne families. PPMD hosts an annual Advocacy Conference in Washington D.C. At this conference, you will be trained to meet with congress staff and advocate for Duchenne. As a result of past advocacy efforts, there has been over \$500 million in funding for Duchenne related programs, a full drug development pipeline, and a standard of care established for those diagnosed with Duchenne markedly improving the quality and length of life. To learn more, please visit parentprojectmd.org/advocacyconference.

Race to End Duchenne

Race to End Duchenne has become our largest community fundraising program. PPMD leads teams in races all over the country. Whether you are a novice or experienced runner, we have a race for you to participate in. To learn more about our Race to End Duchenne program please visit racetoendduchenne.org.

“Do It Yourself” events

If you would prefer to host an event in your community, we have many opportunities to assist you. Visit parentprojectmd.org/diy to browse fundraising and event opportunities. Some of these include:

- DIY Race Series: host a 5k, Fun Run, Etc.
- Special Occasions
- Create Your Own Event: have an idea? PPMD will guide you and give you all the tools!

Attend a Coach to Cure MD Game

PPMD has partnered with the American Football Coaches Association (AFCA), a professional organization for over 10,000 college football coaches and staff, to produce the largest national charity devoted exclusively to Duchenne. The goals of Coach to Cure MD are to raise awareness and money for Duchenne and finding a cure.

On one football Saturday of each season, AFCA coaches nationwide wear armbands during a game, and mention Coach to Cure MD during on and off-field interviews. We encourage families to attend games in their area, as well as host tailgating parties. Sign up to represent PPMD at coachtocuremd.org and help us #TackleDuchenne.

NOTES

SECTION 3

Care Considerations



SECTION 3

Care Considerations

PPMD assembled a group of experts in the field of Duchenne to write the Duchenne Care Considerations, or guidelines for the care and management of Duchenne across the lifespan. Unanimously, it is agreed upon that Duchenne care requires the teamwork of doctors with different specialties (neurology, cardiology, etc.) to provide the best and complete care. Your neuromuscular care team aims to keep your child safe, healthy, and able to enjoy activities. The following section provides an overview of care you should anticipate as well as tips to avoid illness or injury to keep your child as healthy as possible.

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A short list of things to know & do now

The following information can also be found at parentprojectmd.org/newlydiagnosed.

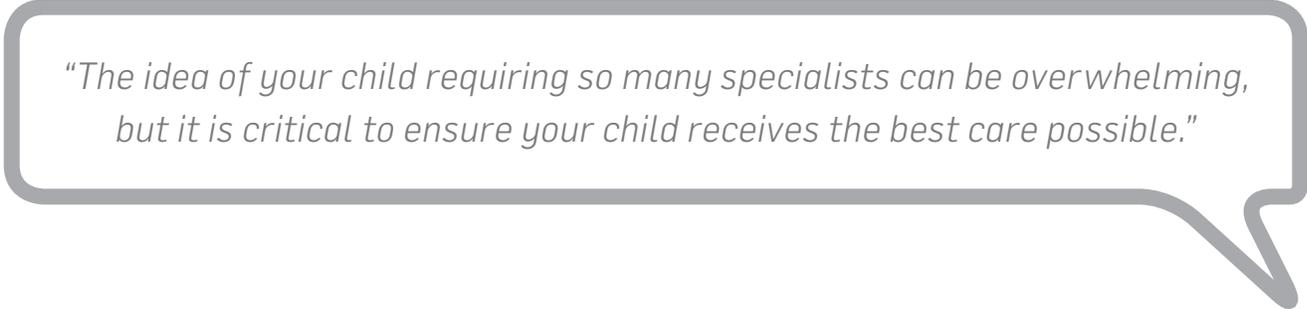
1. Register with PPMD as a newly diagnosed family.
2. Find a **neuromuscular provider** who knows how to care for patients living with Duchenne.
3. Talk to your child's healthcare provider about **physical activity**. Too much of some types of strenuous activity are bad for your child's muscles.
4. Ask your neuromuscular provider or **physical therapist about stretching**, and learn how to stretch your child's muscles appropriately.
5. If your child is getting **physical therapy (PT)**, ask the therapist to contact PPMD for **specific PT recommendations**.
6. Ask about the risks and benefits of **starting corticosteroids** (most children start before age 5).
7. Duchenne is a progressive disease (meaning that it gets worse over time), but it gets worse very, very SLOWLY and **patients progress at different rates**.
8. Most parents and families are able to **adapt to living with Duchenne**. It's not easy and it takes time, but we can help you get there.



Finding a care team

The diagnosis of Duchenne can happen when your child is a newborn, infant, or young child. At any of these stages, your child's primary care provider (PCP) will remain as your child's "medical home" and can offer a sense of stability and support during this new transition. This is typically a pediatrician, family practice physician, nurse practitioner, or internist. You should continue to visit your child's PCP regularly for wellness visits, scheduled vaccinations, and for acute medical issues such as mild illnesses. However, your PCP will need to refer you to a neuromuscular specialist (NMS) — a pediatric neurologist or rehabilitation physician who specializes in neuromuscular disorders — to manage your child's Duchenne.

This NMS will coordinate the care of your child and will serve as your child's lead clinician, taking overall responsibility for your child's Duchenne care, including in-between visits. This will be a very important person moving forward as they will guide you through your child's care plan. PPMD recommends you see a NMS who is a part of a coordinated multidisciplinary care team, meaning a group of many specialists who work together to provide the best and most appropriate care for your child.



"The idea of your child requiring so many specialists can be overwhelming, but it is critical to ensure your child receives the best care possible."

Other clinicians that may be involved during this time include:

- Genetic Counselor
- Physical Therapist
- Cardiologist (doctor who specializes in the heart)
- Pulmonologist (doctor who specializes in breathing)
- Registered Dietician Nutritionist (RDN)
- Psychologist, Psychiatrist, or Counselor
- Social Worker
- Speech-Language Pathologist (SLP)

PPMD's Certified Duchenne Care Center Program

PPMD has developed a Certified Duchenne Care Center (CDCC) Program. This program aims to ensure medical centers maintain the highest standards in clinical and sub-specialty services, rapidly apply new evidence-based knowledge, comply with standards in clinical care, and consists of a growing network of CDCC's across the country. To view a list of our current CDCCs, please visit parentprojectmd.org/carecenters.

Primary care

Everyone needs a primary care provider (PCP). This is typically a pediatrician, family practice physician, nurse practitioner, or internist who will serve as your child's "medical home." After the diagnosis of Duchenne is confirmed by a neuromuscular specialist (NMS), information and resources should be sent to your PCP about Duchenne. An ongoing medical relationship with the PCP will provide a continued, much-needed source of stability and support.

Your child's NMS will lead the management of your child's Duchenne. Your child's PCP will remain your first-line care provider for routine childhood well and sick care. During childhood, the PCP will evaluate developmental milestones, conduct annual hearing and vision screening, annual physical and mental health exams, and ensure vaccinations are given on schedule. Your PCP will be able to communicate and collaborate with your child's neuromuscular team to ensure your child receives comprehensive care.

Follow recommended vaccination schedules

Following recommended vaccination schedules is important for staying healthy and avoiding potentially harmful illnesses. It is important to speak with both your primary care provider and your neuromuscular care team about vaccinations — some changes may be necessary for people living with Duchenne. This is especially important for those planning to start steroid (corticosteroid) therapy. Learn more about vaccination recommendations by visiting parentproject.org/vaccinations.

Consider steroids

Your NMS will use standardized assessments at each visit to evaluate your child's physical symptoms and track changes closely. Your NMS is very educated on the natural history of Duchenne, which is how Duchenne progresses over time. Your NMS will use this assessment information to select therapies that are custom to your child's current needs, as well directed towards what they anticipate happening in the future.

One of the first therapies your NMS may mention is corticosteroids ("steroids"). Corticosteroids are different from the anabolic steroids that are sometimes misused by athletes who want to become stronger. Corticosteroids are the only medication known to help slow down the muscle damage and weakness caused by Duchenne. **Prednisone** (Prednisolone) and **deflazacort** (Emflaza) are both steroids used for the treatment of Duchenne. Studies have shown that steroids may extend ambulation by 2–3 years, as well as, preserve pulmonary function.

Experts in the field agree it is best to start steroids before the plateau phase, meaning when your child still physically acts similar to their peers. In this phase, your child will have learned all of their motor skills (crawling, walking, climbing stairs, etc.) and will have minimal trouble managing these skills independently. This is typically around age 4–5 years.

The decision to initiate steroids should not be made without extensive education by your NMS. While the benefits of steroids have been extensively documented, there are many side effects that you should be made aware of before starting steroids. In addition, there are multiple dosing regimens that your NMS may discuss. The two most common regimens in the US are doses given at the same time daily, or higher doses given only two days/week, also referred to as "weekend dosing." Be sure to discuss these options with your NMS.

Your NMS will monitor your child closely at each visit after starting steroids, and while working to reach a therapeutic dose. Some side effects due to steroids include:

- **Weight gain:** some children gain a great deal of weight when taking steroids; some do not.
- **Impaired growth (growth hormone deficiency):** daily steroids usually delay vertical growth.
- **Behavior issues:** some children exhibit difficulty controlling their anger and impulses; some do not.
- **Thinner, weaker bones (osteoporosis):** steroids decrease the amount of minerals deposited in the bones, particularly with daily steroids.
- **Stomach problems (gastroesophageal reflux or “heart burn”):** steroids can irritate the lining of the esophagus and stomach and should always be taken with food.
- **Eye issues:** some children develop cataracts and changes in the retina, which is more common with steroids. These are usually not dangerous, but should be checked every year.

It is important not to be discouraged if your child begins to show side effects of steroids. Your NMS can offer many remedies for these side effects, and can also try adjusting the dose or dosing schedule to offset these effects while still providing the therapy. Typically, families decide the benefits of steroid therapy outweigh the risks of side effects.

If your child experiences undesirable side effects, it is imperative that you **never stop taking steroids abruptly**. Steroids provide the body with the hormone cortisol, known as the *stress hormone*. When your child is on steroids, their body stops producing cortisol naturally. If your child abruptly stops taking steroids, it takes the body several months to begin to produce cortisol again. The lower-than-normal level or absence of cortisol can cause what is known as *adrenal insufficiency*. This can lead to an adrenal crisis, which is a life-threatening emergency. Your NMS will counsel you about what to do in this situation as well as provide education about *stress dosing* of steroids when indicated. Additional information about preventing adrenal insufficiency can be found in the PJ Nicholoff Steroid Protocol (parentprojectmd.org/pj).

Exercise & stretching

Your rehabilitation team, which will include a physical therapist (PT) and sometimes a Physical Medicine and Rehabilitation (PM&R) doctor, will be important moving forward. Even if your child is showing mild or no physical symptoms, it is important to be proactive with care.

Gradually introducing stretching regimens can keep muscles flexible and can decrease tightness at the joints. This can help maintain movement in the joint and decrease pain or cramping of the muscles. Your rehabilitation team will advise on stretching regimens that are specific to your child. Eventually, stretching should be a part of your daily routine. To view our series of stretching videos, please visit parentprojectmd.org/stretching.

Your rehabilitation team may recommend night splints (also known as AFOs) at an early age. Night splints are great for maintaining the flexibility of the Achilles tendon as well as hold the foot in a comfortable position. These are made of light plastic, and are made specifically to fit your child's foot. Some parents find it difficult to get their toddler or young child to wear these overnight. Your child may be able to choose patterns, designs, and colors of these splints, which may be helpful in acceptance. Starting early and regularly wearing night splints will help your child get used to them over time.

Busy children love to play and move! It is important to discuss safe play and exercise with your rehabilitation team. Exercise should have a focus on stretching and maintaining range of motion without being too hard on the muscles. Some exercise (such as jumping in a bouncy house or trampoline) may be harmful to muscles and should be avoided. We realize that it is sometimes difficult to prevent your child from participating in activities that may not be good for them. It is best you try to avoid harmful activities when possible, but not to overstress. When in doubt, it is best to try to choose activity and exercise that has been recommended by your physical therapist or PM&R doctor.

Activities that are low intensity and do not cause stress on the muscles, such as swimming, are recommended by physical therapists. If aquatic therapy is available in your area, it may be a great option for your child. Parents often report their child enjoys the opportunity for regular pool time because it allows them to be independent and enjoy the physical activity.

Growth & nutrition

Your child's standing height and weight should be measured at each neuromuscular care team visit. This information will be tracked on a growth chart. This allows your child's care team to detect any problems with weight gain or loss, as well as track growth.

It is important for your child to have a nutritional assessment at the time of diagnosis and/or when starting steroids. It may be suggested your child see a Registered Dietician Nutritionist (RDN) if they are found to be nutritionally deficient in any way. Their job is to assess current dietary habits as well as make recommendations to optimize health and growth as well as to avoid undesirable side effects of steroids.

Maintaining good nutrition is a family event, and is essential from diagnosis throughout life. Providing a healthy, well-balanced diet with a full range of food types is necessary for everyone. Extra attention to diet may be required at diagnosis and/or when steroids are started. Steroids may cause your child to feel hungrier, potentially causing them to over eat. This can cause too much weight gain, which is not healthy for any child and may need to be addressed by an RDN. The RDN can give you helpful tips on what to do if you experience this and how to prevent your child becoming overweight.

Steroids also put children at risk for weak or frail bones. Your RDN can review your child's diet to make sure they are getting enough vitamin D and calcium in their diet. Your neuromuscular team should check your child's vitamin D blood level yearly. If the vitamin D level is found to be low, or your child's diet is felt to be deficient in calcium, your RDN may advise supplements or increased levels in your child's diet. Vitamin D and calcium are both important to maintain strong bones and lessen your child's risk for fractures.

Learning, behavior, & speech

Children who have Duchenne have a higher risk for having learning and behavior difficulties as well as speech delays. It is unclear as to why these problems are more prevalent in people living with Duchenne, but they can be a result of several factors including stress, physical limitations, and steroid therapy. Often, these issues are the most difficult for parents and families to deal with rather than for the children. At least once a year, someone from your neuromuscular clinic should check with you and your child about behavior, learning, emotional adjustment, and social functioning. If you have any concerns or need a more comprehensive assessment, resources, or medication, ask for a referral to a professional who specializes in evaluating and treating behavioral health problems.

We recommend you think about seeking help if you notice any of the following:

- Delays in language development or problems with communication
- Has difficulty interacting with others and/or making friends
- Is often angry, argues a lot, or is very inflexible about what they want to happen
- Is forgetful or absent-minded, or has difficulty paying attention
- Is very quiet and withdrawn, and prefers to be by themselves
- Is very fearful or anxious
- Often has a negative/pessimistic attitude or says bad things about themselves
- Has difficulty learning new academic skills or earns poor grades
- Has rituals or routines that have to be followed in a particular way
- Has difficulty sitting still, is impulsive, or very impatient

If you notice one or more of these issues, take your child to a professional who specializes in neuropsychological or psychological testing. This person can do tests to identify learning, behavioral, or emotional issues (or risk factors for developing them) and can help you come up with a management plan. Ideally, this testing should be done around the time your child is diagnosed, as they start school (ages 5–6), or any time new concerns arise.

Depending on your child's needs, there are a number of interventions that may help. Examples include psychotherapy (individual or group counseling, parent training, family therapy, and applied behavior analysis), academic therapies and Special Education, social skills training, and medication. PPMD also offers resources to help navigate your child's Individualized Education Program (IEP) and 504 plans to ensure your child is receiving the resources they need. For more information about IEP, 504 plans, and more resources please visit parentprojectmd.org/classroom.

If your child has a speech or language delay, your PCP or NMS can refer you to a speech language pathologist (SLP). A SLP can work with your child to improve any issues they have with speech.

For more information about problems concerning psychosocial health, please visit our website parentprojectmd.org/learning.

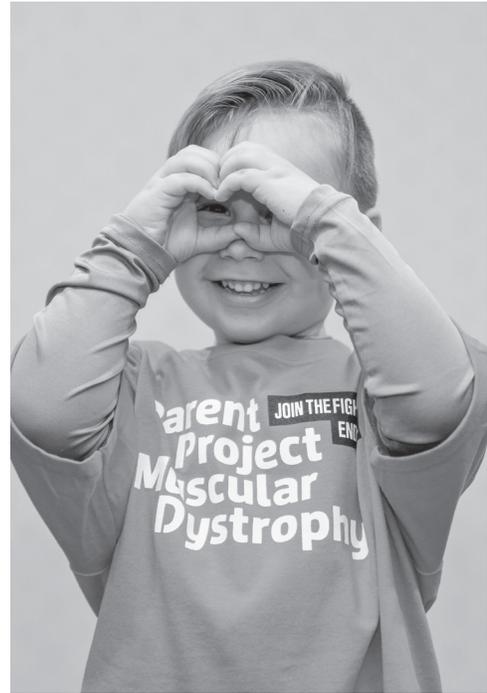
Heart & breathing muscles

Typically, problems with the heart and breathing muscles are not likely to be present at this stage, but both a cardiologist (heart doctor) and a pulmonologist (breathing doctor) should start seeing your child at diagnosis and be built into regular follow-up clinic visits to establish a baseline (what is “normal” for your child). If your child goes to a multidisciplinary center, a cardiologist and pulmonologist will be available to you.

Heart health

In Duchenne, the heart muscle, just like the skeletal muscle, is affected by the lack of dystrophin. Cardiomyopathy, or disease of the heart muscle caused by a lack of dystrophin, leads to lower-than-normal function and heart failure over time. Although “heart failure” sounds incredibly scary, it really just means that the heart is having difficulty meeting the energy needs of the body on its own. Heart failure can be managed with medication and monitoring for a very long time.

Cardiac monitoring, including an electrocardiogram (ECG) and cardiac MRI or echocardiogram, is recommended at diagnosis and then annually up to age 10. After age 10, your cardiologist will reevaluate whether your child needs to be seen at more frequent intervals. An ECG looks at the electrical conduction of your child’s heart. Both echocardiograms and cardiac MRI imaging look at the structure and function of your child’s heart. Cardiac MRIs are considered the “gold standard” for cardiac imaging in Duchenne because your cardiologist is able to see any fibrosis (scarring) of the heart muscle. However, echocardiograms are more widely available, and also allow your child’s cardiologist to monitor the heart function. It is a good idea for your child to have both an ECG and echocardiogram/cardiac MRI yearly, starting at a young age, so your child’s cardiologist can detect small changes over time.



Breathing muscle health

In Duchenne, the muscles that assist in breathing and coughing weaken over time. Typically, young children with Duchenne do not show any signs of impaired breathing or coughing. However, it is a good idea for them to visit a pulmonologist annually from the time of diagnosis. At their pulmonology appointment, a respiratory therapist will conduct a pulmonary function test. This test requires your child to breathe into a mouthpiece connected to equipment that measures various pressures associated with their breathing strength. Although it may be difficult to get your child to cooperate for this test, it is valuable for the pulmonologist to collect baseline measurements. Additionally, with each subsequent visit to the pulmonologist your child will improve their ability to do the testing.

PPMD has developed a series of videos to raise awareness about respiratory care in Duchenne. The first video in the series explains what to expect at your first pulmonologist visit. This may be helpful for you to watch as well as your child before your appointment. To view these videos, please visit parentprojectmd.org/pulmonaryvideos.

What to do in an emergency

It is important to know what to do in the event your child becomes sick or injured, requiring a visit to the emergency room or hospitalization. More often than not, the emergency room staff may not have extensive experience treating people with Duchenne. If your child is in the emergency room, it is best to personally call your neuromuscular team to let them know and NOT to rely on the emergency room staff to do so. Make sure you have all necessary information with you to show the emergency room clinicians in case of a medical emergency. There are two easy ways you can keep this information with you at all times:

- **PPMD Smartphone App:** If you have an Apple or Android smartphone, you can download the Parent Project Muscular Dystrophy App and access this emergency care information right from your phone.
- **PPMD Emergency Cards:** Visit parentprojectmd.org/emergency to obtain a copy of our wallet-size emergency information card or large emergency information card to attach to backpacks and scooters.



Safety & prevention

The best advice to stay out of the emergency room is prevention. Below are some tips to prevent emergencies from happening:

- Keep scheduled immunizations up to date and have the flu shot every year, lowering the risk for more severe forms of illnesses that may result in a hospital stay
- Prevent falls by keeping your child's path clear (remove throw rugs, cords, etc.) and take care on even surfaces (i.e. wear non-slip shoes)
- Ensure that your child always wears their seatbelt, both in the car and while riding any type of scooter or assistive device
- Children with Duchenne tire more quickly than peers, and while most children are good at self-limiting their activity so that they don't overtire, it is important to allow more rest periods so they do not injure themselves trying to "keep up"

Anesthetic precautions

If your child needs to undergo a medical procedure requiring anesthesia for any reason it is important to discuss the anesthesia plan beforehand. IV anesthesia is considered the safest option for people with Duchenne. Local anesthetics (topical creams, etc.) and nitrous oxide gas ("laughing gas") are safe as well. Inhaled anesthesia should be avoided when possible, or if absolutely necessary should be monitored extremely closely. **The drug succinylcholine should never be given to a person with Duchenne** due to the dangerous side effects.

Dental dilemmas

Dentistry generally can, and should, be performed with the minimal amount of anesthesia possible while providing the maximal physical and emotional comfort. As discussed above, local anesthetics and nitrous oxide are safe to use in people with Duchenne.

Falls & fractures

Young children with Duchenne may have an unstable gait, as well as tire quicker than their peers, putting them at risk for falls and other injuries. In addition, if they are on steroids, their bones may become less dense and prone to fractures. If your child has a leg fracture, ask for a referral to an orthopedic physician with experience in Duchenne. They can work with you to develop a treatment plan that will allow your child to get back on their feet as quickly as possible.

If your child has a fall or leg injury and has a rapid onset of shortness of breath/difficulty breathing, changes in alertness (confusion, agitation, etc.) this is an emergency. Go to the emergency room immediately and let the staff know these symptoms may be due to a **Fat Embolism Syndrome**. This is a **medical emergency** in which fat particles from the inside of the bone enter the blood circulation and can impair blood flow to major organs.

Missing steroid dose(s) and stress dosing

As discussed earlier in the steroid section, if your child is unable to take their oral steroid doses for more than 48 hours for any reason (vomiting, etc.) go to the emergency room. It is best to bring the PJ Nicholoff Steroid Protocol and show to the staff. They can administer IV steroids until oral medication can be tolerated again.

As mentioned previously, children taking steroids are unable to produce cortisol, the hormone that allows the body to handle stress. If your child is experiencing a traumatic or stressful event (severe illness, surgery, fracture, etc.), a stress dose of steroids may be needed. Recommendations for stress dosing can also be found in the PJ Nicholoff Steroid Protocol (parentprojectmd.org/pj).

“Duchenne is not your fault. You must take care of yourself so that you can be the best parent for your children.”

Females with dystrophinopathy or “manifesting carriers”

As mentioned above, females who carry the mutated Duchenne gene have a small risk for health problems. Potential symptoms may include changes in heart function, physical muscle symptoms, and psychosocial issues such as anxiety or learning disabilities. When young girls or women manifest these symptoms, they are considered to be *females with dystrophinopathy*, more commonly termed *manifesting carriers*. All female carriers of Duchenne should be evaluated by a healthcare provider familiar with Duchenne.

Skeletal muscle symptoms

Most carrier females (80–90%) have no problems with their skeletal muscles. Some may have mild muscle weakness, fatigue (a tired feeling), pain, or cramping in their muscles. However, some women, and even young girls, may have problems with balance causing falls and moderate to severe muscle weakness.

These issues should be addressed by a neuromuscular specialist and managed by a physical medicine and rehabilitation (PM&R) physician or a physical therapist (PT). Females with skeletal muscle symptoms should be followed regularly.

Cardiac function

Carrier females have an increased chance of changes to heart function. It is not yet known how common heart changes are, but some studies have estimated that 10-50% of carriers have heart changes. For this reason, it is important for all women who are carriers and women who are at high risk to be carriers (for example, have a son or brother with Duchenne) to have regular heart evaluations, beginning in their late teens to early adult years.

Women who know that they are carriers or women who suspect they might be carriers should discuss heart screening with their doctors. An appropriate heart evaluation may include an electrocardiogram (EKG) and echocardiogram. However, we do encourage women who are carriers to have a cardiac MRI if possible to look for fibrosis of the heart muscle.

Psychosocial care

Learning that one is a carrier can cause strong emotions. It is not uncommon for women to feel sad, worried, or guilty after learning that they are carriers. In addition, carrier females may have additional feelings related to the uncertainty around their own health. It can be especially challenging for females with dystrophinopathy who are trying to take care of an affected son while dealing with their own symptoms. Anxiety and depression can be serious conditions and should be treated by mental health professionals.

In addition, cognitive or behavioral issues such as ADHD or learning problems may also occur. Although these issues may or may not be related to being a carrier of Duchenne, it is important to be evaluated by a professional and treated early.

Young females with dystrophinopathy

Although rare, there are families with affected young girls in our Duchenne community. While there is limited information available about this population, we are working to raise awareness and improve care for all females with dystrophinopathy. Young girls with symptoms occur on a spectrum of severity ranging from mild balance impairment or muscle fatigue to more severe weakness and learning disabilities. In this case, girls and young women should be evaluated and followed by a comprehensive neuromuscular center, just like boys with Duchenne.

As discussed in Section 1, the decision to test young sisters of boys living with Duchenne remains a family decision, especially if the sister is under the age of consent. However, we do recommend bringing any concerns about delayed developmental milestones, skeletal muscle weakness, cognitive or behavioral issues to your son's neuromuscular provider. If symptoms of dystrophinopathy are identified, then follow-up with CK testing and genetic testing is recommended.

NOTES

SECTION 4

Research & Clinical Trials



SECTION 4

Research & Clinical Trials

With over 35 companies working on new therapies for Duchenne, you may be approached about participating in a clinical trial. Often, these conversations arise with one of your child's neuromuscular care team clinicians, a pharmaceutical company representative, or through another family with a child with Duchenne. Navigating clinical trial participation is a very complex system, requiring extensive education and understanding of the process. PPMD aims to provide you with the tools to make the best informed decision for your family. This section is meant to provide an overview of clinical trials and what it means to participate in a trial.

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What are clinical trials?

Clinical trials are research studies in humans designed to determine if an experimental therapy or treatment works. Clinical trials are one of the final steps in a long process that begins with research in a lab. Much of what we know about disease progression, including effective therapies and treatments, is the result of past clinical trials. Clinical trials can be used to determine whether a treatment is safe and how well the new treatment works in particular people.

Who can participate in a clinical trial?

Each trial or study has rules about who can or cannot be in the study. These rules are called “eligibility criteria” and they describe features that must be shared by all the people in the study. The eligibility criteria, also called the inclusion and exclusion criteria, differ from study to study.

It is important to understand that eligibility criteria are not used to reject people personally. Instead, the criteria are used to identify appropriate participants, keep them safe, and design trials that can determine whether an experimental product works in a specific population as quickly as possible. The criteria help ensure that researchers will be able to answer questions that they plan to study. Before joining a clinical trial, a participant must go through a screening process with the study team to determine if they meet the eligibility criteria.

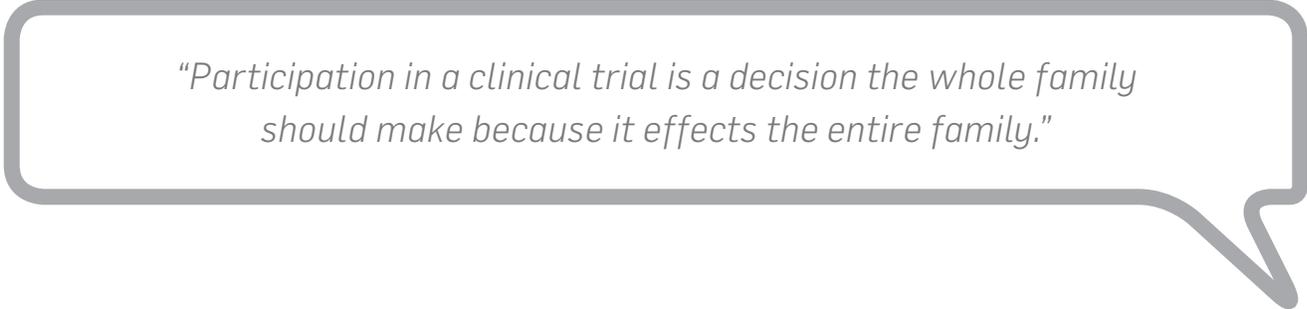
What happens during a clinical trial?

The clinical trial process depends on the kind of trial being conducted. The clinical trial team (or study team) includes doctors and nurses, as well as social workers and other health care professionals. They examine the health of the participant at the beginning of the trial, give specific instructions for participating in the trial, monitor the participant carefully during the trial, and stay in touch after the trial is completed.

Some clinical trials involve more lab tests, procedures, and doctor visits than the participant would normally have for an illness or condition. For all types of trials, the participant will always work closely with the study team at their trial site. Clinical trial participation is most successful when the protocol (the required treatments/lab/procedures) is carefully followed and there is frequent contact with the study team.

The decision to participate in a clinical trial

Clinical trials may lead to new advances in the treatment of Duchenne. However, the decision to join a clinical trial is yours, and no one should pressure you.



“Participation in a clinical trial is a decision the whole family should make because it effects the entire family.”

Understand the goals

A clinical trial may add important scientific knowledge and lead to new advances. However, it is not meant to help any one person. Some people may benefit, and others may not. It is important to fully understand the purpose of the clinical trial before signing up. parentprojectmd.org/consideringtrials includes a long list of questions that may help you prepare for your meeting with the clinical trial team.

Weigh the benefits and risks

Clinical trials measure benefits and risks over time. However, you have to choose at the beginning when benefits and risks are still uncertain, which is difficult. No trial can promise a benefit; in most trials, most people do not get better. However, this does not mean you can't benefit. Your health could improve and you may have other positive experiences such as feeling good that you are helping others or being more hopeful.

Being in a clinical trial can allow a person to:

- Play an active role in their own health care
- Gain access to new research treatments before they are widely available
- Obtain expert medical care at leading health care facilities during the trial
- Help others by contributing to medical research

Clinical trials are experiments and each trial has its own risks. Some potential risks include:

- Side effects which may be unpleasant, serious, or even life-threatening
- May require procedures (i.e. muscle biopsies)
- The experimental treatment may not be effective for everyone
- Your time and attention may be strained due to study visits, treatments, hospital stays, etc.
- Participating in a trial may be emotionally difficult
- Participating in a trial may exclude you from participating in future trials

Understand the commitment

Though joining a trial can be rewarding, it is a big commitment. It is important to ask many questions regarding the time commitment of participating in the trial. Ask questions about how long the trial lasts, how long the visits to the trial sites are, what happens at each visit, and if there are any costs associated with the trial.

Additionally, you may not live near the trial site, so travel may be required. Visiting trial sites may also mean that you may need to take time off of work and your child may need to take time away from school. Travel also may mean less free time for other activities important to you, your child, and other members of your family.

Use of placebo medications

Some trials use a placebo. Placebos help researchers understand if benefits and risks experienced during the trial happen because of the potential treatment, or if they happen by chance. Placebos are non-active alternatives, like a sugar pill. In most trials using a placebo, participants are randomized by chance to the placebo or the treatment group. You would not know whether your child is receiving the placebo or active drug. However, most people who are in the placebo group get the active drug later on during the trial. You can decide if a placebo trial is acceptable to you.

Tips to help you to decide

- Learn all you can from doctors, researchers, and other trusted sources
- Talk to other people about their experiences in trials
- Weigh day-to-day requirements, potential risks, and potential benefits
- Take your time deciding to make the best decision for you and your family
- Explain the trial to your child in an age-appropriate way and ask for their input

Clinical trial decision guide

PPMD offers worksheets that will help you decide whether or not clinical trial participation is right for you and your family. The goal of these worksheets is to help you think through your decision so that you have the most impactful experience possible. To view these worksheets, please visit parentprojectmd.org/consideringtrials.

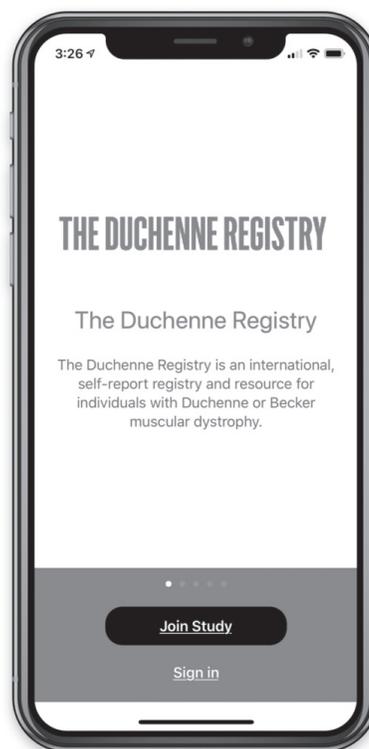
Exploring clinical trials

There are many different ways you can learn more about clinical trials. Ask your neuromuscular care team about clinical trials that your child may be eligible for. They can connect you with people who are informed about the various trials recruiting. PPMD keeps an updated list of all current Duchenne clinical trials that can be found at parentprojectmd.org/exploretrials. You may also visit clinicaltrials.gov, a database that hosts information about clinical trials.

PPMD has many valuable resources that aim to connect people with Duchenne with clinical trial opportunities if they choose. There is a Clinical Trial Video Library available that includes several videos that explain the various aspects of exploring the participation of clinical trials. To visit this series, please visit parentprojectmd.org/ctvideos.

Join the Duchenne Registry

The Duchenne Registry connects Duchenne and Becker patients with actively recruiting clinical trials and research studies, and educates patients and families about Duchenne and Becker research. At the same time, The Duchenne Registry is a valuable resource for researchers, allowing access to data provided by patients and their families — information that is vital to advances in the care and treatment of Duchenne. To learn more and to register, please visit duchenneregistry.org.



Use PPMD's interactive drug development pipeline

PPMD provides a variety of resources for learning about potential treatments and clinical trials for people with Duchenne and Becker, as well as female carriers. PPMD developed an interactive drug development pipeline to further help the community stay up-to-date on all therapies that are currently in development. To explore investigational drugs by therapeutic approach, please visit parentprojectmd.org/pipeline.

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Conclusion

We hope that you find this guide to be a useful resource. In my own journey with Duchenne, I realized quickly that knowledge is power. But I also know that everyone needs to absorb the overwhelming amount of information that exists, at their own pace.

There is a lot of information here and a lot of it is tough to absorb. But the most important take away from this entire guide is to remember you are not alone. You are now part of an incredibly strong, caring community that will help you and your family navigate this new diagnosis. It won't be easy and you will have many, many hard days. But you will also have wonderful days and you and your child will still create an amazing life together — it's just going to be different than you imagined.

"You are not alone. PPMD is here for you."

Let PPMD be your community. Let PPMD fight for you on days when you can't. And when you are ready, join us. We are all in this together and none of us can do it alone.

Together, we will end Duchenne.



NOTES

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