

Parent Project Muscular Dystrophy JOIN THE FIGHT. END DUCHENNE.

New Diagnosis and Early Care Guide



Duchenne & Becker Muscular Dystrophy: New Diagnosis and Early Care Guide

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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.



No one is prepared to receive the diagnosis of Duchenne or Becker. Most often, parents and caregivers have no prior knowledge of the condition, 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.

One-to-One Meetings for Personalized Support

The Duchenne and Becker journey is complex, but PPMD is here for you. Schedule a one-to-one meeting with an expert member of the PPMD team for personalized support. We're here to provide you with resources to help you navigate the concerns and decisions that are part of Duchenne and Becker. Remember, you are not alone.



parentprojectmd.org/foryou
careteam@parentprojectmd.org



A Letter from our Founding President



Friends,

It is not easy to enter the world of Duchenne and Becker muscular dystrophy. You may feel overwhelmed, lost, and very 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. Why my sons? What did they do to deserve this diagnosis?

Duchenne and Becker often arrive unexpectedly, without any family history.

In your new world, you will learn a new language—the language of Duchenne and Becker. It will include words you have never heard before, and you may stumble. But you will repeat them until you become familiar and understand what they mean.

You will become a partner in your child's care. Your knowledge about your child's medical care, needs, and possibilities for trial and treatment 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—one where dozens of companies are invested in developing treatments to stop or slow the progression of Duchenne and Becker. 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 new path which will lead you to new places. It will lead you to people you never expected to meet. Some of those places will be very helpful and you will find good, caring people. They will help keep you steady and guide your decisions as you explore the world of Duchenne and Becker.

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 and however unstable and uncertain you feel, you will regain some balance, and your strength will return.

Some people in your life may not understand Duchenne and Becker. They may slowly drift away. Others will welcome you and your family 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 new friends. They will come into your life and help in unexpected ways. They will become family.

The world of Duchenne and Becker 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 that you will once again find your smile.

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

A handwritten signature in black ink, appearing to read 'Pat Furlong'. The signature is stylized and fluid.

Pat Furlong

Founding President, Parent Project Muscular Dystrophy

NOTES

SECTION 1

What is Duchenne & Becker
Muscular Dystrophy?



SECTION 1

What is Duchenne & Becker Muscular Dystrophy?

This section provides an in-depth overview of Duchenne and Becker muscular dystrophy. Because Duchenne is a genetic condition, 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 and Becker 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 and Becker muscular dystrophy are on a spectrum of muscle diseases known as *dystrophinopathies*. 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 a dystrophinopathy with many similarities to Duchenne. And while Becker can be variable, depending on when symptoms first present, typically the symptoms progress more slowly 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 condition 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*. More information on manifesting carriers is provided in section 3 of this guide.

Hope remains strong. While there is currently no cure, 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 condition. 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 people 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.

What causes the symptoms of Duchenne?

Duchenne is caused by changes in the *DMD* gene, which codes for or instructs the body to make the muscle protein called dystrophin. 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 result 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 observe 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 a bit clumsy, experiencing frequent falls. Additionally, 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 condition, 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.

It may be helpful to review some basic genetics as we discuss inheritance of Duchenne. Duchenne is an *X-linked disorder*, which means the dystrophin gene is on the X chromosome. Every person has 23 pairs of chromosomes, which contain all of their genes. The last pair of chromosomes is referred to as the sex chromosomes and can determine if someone develops as male or female. One chromosome in each pair is inherited from each parent. Typically, someone who develops as female will have two X chromosomes, and someone who has one X and one Y develops as male.

Since the *DMD* gene is on the X chromosome, we see a difference in effect for females and males. Males typically only have one X chromosome, so if the *DMD* gene on that chromosome has the variant, they will have Duchenne. Since females have two X chromosomes, they may or may not experience symptoms since there is a "back-up" copy of the *DMD* gene. Females with one X chromosome with a *DMD* gene with a variant are known as carriers.

Since Duchenne is genetic, was it inherited from me?

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

Duchenne can also occur even if no one in your family has had it before. The *DMD* gene is very large, which increases the possibility of a new variant happening during the formation of an egg. This is why approximately one-third ($\frac{1}{3}$) of Duchenne cases are caused by a *spontaneous variant*, 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 variant or has a new variant is for the mother to have carrier testing. Read more about carrier testing on the next page.

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 variants, 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 variant in the DNA. It is important to know the specific genetic variant 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 your child has Duchenne or Becker, or if they may have another type of muscular dystrophy that shares some of the same features.
2. **To enable testing of family members:** Once the specific variant in a family is known, then other family members can be tested to determine if they are carriers of the variant. Targeted genetic testing is the best method for performing accurate carrier testing.
3. **To determine what variant-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 variants. You must know your particular variant in order to participate in a clinical trial and to access any current or future variant-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 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 and should undergo carrier testing. This is important because carriers of a *DMD* variant 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, women can make informed decisions regarding future pregnancies and be aware of potential health implications of being a carrier.

Most female carriers of Duchenne are unaware that they are carriers until they have a child with Duchenne. The genetic variant 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 at-risk 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 *DMD* variants 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 some degree of muscle weakness. Care considerations for female carriers is discussed in section 3.

The decision to have siblings tested, and the timing for testing, varies from family to family. This decision can be affected by factors such as age (especially if unable to legally consent), potential symptoms, and the family's personal beliefs. However, it is always recommended that siblings have testing at some point, regardless of their parent's carrier status. 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@duchenregistry.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 5,500 members. All data is entered through The Duchenne Registry mobile app, making the collection of data easy and 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, Becker, and carriers, 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 duchenregistry.org.

NOTES

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. Your first impulse may be to keep this information to yourself to try to protect others and keep things feeling “normal.” While this reaction is understandable, it can be incredibly burdensome to feel like the keeper of serious information, and sharing the news of the diagnosis may help to relieve some of this stress. Talking about it will also allow you to 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 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.

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 that 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.

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.

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.

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. It can also be helpful to support siblings by identifying additional resources for them as they process their feelings, such as with a school counselor or therapist, normalizing that talking about these things is healthy.

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. Establishing this support early on in your journey will be a gift to yourself and your family, especially during times of stress.
- 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.
- Be aware of the high risk for caregiver burnout when caring for a child with complex needs. In prioritizing setting aside time for your interests or hobbies, you can maintain your physical and mental health in order to better take care of your family.

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.

Mental health support & counseling

It is often difficult to find a therapist who is familiar with Duchenne. However, it can be helpful to identify a therapist who has experience in working with caregivers of patients with chronic medical conditions, or parents of special needs children. They are more likely to understand your circumstances, and you can educate them about the specifics of Duchenne. Counseling can be done online if needed or in person. Counseling is particularly important if you think you might be developing depression or overwhelming anxiety.

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. This guide is available alongside additional 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 helping 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).

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. We also have groups for siblings, grandparents, and extended family members; Duchenne affects the entire family, and we recognize that everyone may need their own support network. 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, such as our virtual meet-and-greets. You will receive regular newsletters informing you of all of these topics and more.
- 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 oversaturation. PPMD’s Facebook page ([facebook.com/parentprojectmd](https://www.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 to apply today (parentprojectmd.org/conference).

Attend a local event

PPMD convenes families in-person through personal and locally run events. The **PPMD Together** program prioritizes community, connection, quality of life, and togetherness through education and interactive discussion. **PPMD Connect** groups also host in-person social gatherings, which can be a great way to meet other families and be connected to your local community. To see if there is an upcoming event at a location near you, please visit parentprojectmd.org/events.

Get involved

PPMD offers many opportunities to become involved in the Duchenne community. We understand that engaging may not be the right fit for every family, but many find these initiatives empowering and a way to cope. Getting involved is also a great way to meet families in your area.

Annual Conference

As mentioned previously 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. To learn more, please visit parentprojectmd.org/conference.

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 endurance events all over the country. Whether you are a novice or experienced athlete, we have an event 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:** Fundraising in honor of your birthday, a wedding, or another special moment in your life.
- **Create Your Own Event:** Golf outings, garage sales, basket raffles—from small-scale to large-scale events, PPMD will guide you and give you all the tools for success!

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 the last Saturday in September, AFCA coaches nationwide wear patches 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 the care of Duchenne requires the coordinated 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

- Register with PPMD as a newly diagnosed family to be connected with helpful resources.
- Find a **neuromuscular care center** with a team who knows how to care for patients living with Duchenne.
- Talk to your child's healthcare provider about **physical activity**. Too much of some types of strenuous activity may be harmful to your child's muscles.
- Ask your neuromuscular provider or **physical therapist about stretching** and learn how to stretch your child's muscles appropriately.
- Ask about the risks and benefits of **starting corticosteroids** (most children start before age 5).
- Ask your neuromuscular provider if there are **approved therapies** your child may be eligible for and discuss the risks and benefits.
- Your child may be eligible for support services at school through an **IEP or 504 plan**.
- Duchenne is a progressive condition (meaning that it gets worse over time), but it gets worse very, very SLOWLY and **patients progress at different rates**.
- Schedule time to meet with PPMD's care team and/or genetic counselors through **PPMD For You** to talk about your child's diagnosis and answer any questions you may have (parentprojectmd.org/foryou).
- Take a moment to pause. This diagnosis is overwhelming, and it can be difficult to know where to start or how to absorb all the new information. **You are not alone in this journey, PPMD is here to come alongside you every step of the way.**



This information can also be found at parentprojectmd.org/newlydiagnosed.

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.

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)
- Neuropsychologist, 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.

Delayed milestones & early intervention

Young children with Duchenne may appear developmentally typical at young ages or may present with some early developmental delays. Some young children with dystrophinopathies begin walking later than typical ages (15–18 months). Some may cruise or crawl later or with different patterns. Some toddlers with Duchenne may also have language, cognitive or behavior delays. On a physical exam, some toddlers will have larger calf muscles, wide-based gait, and difficulties with balance.

If any signs of developmental delay are present, it may be helpful to seek an evaluation through your state's early intervention program for services such as physical therapy, occupational therapy, speech/language/feeding therapy and/or early education support. You may ask your child's PCP to connect you to your state's early intervention program, but a doctor's referral is not necessary to seek an evaluation.

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 parentprojectmd.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. Steroids are the only medication known to help slow down the muscle damage and weakness caused by 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 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 in addition to the specific drug prescribed 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 often 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).

Explore variant-specific & genetic therapies

In recent years, there have been several FDA approvals for Duchenne-specific medications. Many of these therapies aim to work by restoring or replacing dystrophin in the body, which may slow muscle breakdown and the progression of Duchenne. Due to the mechanism of these drugs, their labels define strict eligibility criteria including age restrictions, genetic variant amenability, and other necessary testing. If you have questions about your child's variant and therapy eligibility, you can discuss your questions with PPMD's genetic counselors at coordinator@parentprojectmd.org.

Due to the nature of these medications, they come with a high price tag and families may experience barriers to receiving access for their child. Your prescribing physician, who is often your NMS, will work with you to submit for insurance coverage.

Exon-skipping therapy

One of the most common types of variants in the dystrophin gene occurs when a piece of the code in the gene is missing or deleted. By skipping additional segments of the dystrophin code called exons, the deletion can shift from an out-of-frame deletion to an in-frame deletion. Typically, an in-frame deletion results in a smaller, but partially functional, dystrophin protein. This shortened protein is expected to help stabilize muscle cells and therefore slow progression of symptoms of Duchenne.

Gene therapy

Gene therapy for Duchenne is centered on the goal of successfully introducing into a muscle cell the correct genetic code, or recipe, necessary to make the dystrophin protein. Because dystrophin is such a large protein, smaller versions (referred to as micro-dystrophins) are inserted into the delivery vehicle. Viral delivery using the adeno-associated virus (AAV) harnesses the virus's natural ability to deposit genetic material right into the muscle cell. The result of this viral "infection" would be the successful delivery of the microdystrophin gene into muscle cells in the person's body so that a smaller but functional dystrophin protein could be made. Gene therapies may not be specific to certain genetic variants, and people with many different genetic variants may consider gene therapy.

Access & reimbursement

Because these newer treatments can be very expensive, you will need to work with your insurance company and healthcare provider on achieving access. Every insurance plan is different so it's important to understand the coverage and policies in place for your child's plan. Additionally, most insurance companies have case managers on staff that can be a helpful resource for families to understand their benefits. Frequently, drug companies will also offer case management services to help obtain approval.

When your doctor prescribes a treatment, oftentimes insurance companies will require a "prior authorization". This process will require paperwork from your doctor explaining why the treatment is medically necessary and how it will help. If a prior authorization is approved, that does not always mean insurance will cover the full cost so it's important to understand what you will be responsible for out of pocket and what resources may exist to help offset any out-of-pocket expenses.

If your insurance company denies a medication, you can appeal this decision with the help of your neuromuscular team. They will work with you on attempting to overturn this decision. Unfortunately, this process can take a long time but there are plenty of resources that can help with this process. PPMD has put together an access page that provides sample letters of medical necessity, relevant publications for approved therapies, and more. You can also schedule a time to talk to PPMD's care team for advice on navigating this process (parentprojectmd.org/foryou).

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.

Stretching & positioning routines

Gradually introducing stretching and positioning 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, making it easier for your child to move. Your rehabilitation team will advise on stretching and positioning regimens that are specific to your child. Eventually, stretching and positioning 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 ankle foot orthotics or AFOs) at an early age as one means of positioning. Night splints are great for maintaining the flexibility of the calf muscles, 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.

Staying active

Busy children love to play and move! While it is important for your child to be active, it is also important to protect their muscles. Therefore, we recommend that you discuss safe play and exercise with your rehabilitation team. Exercise should have a low intensity and focus on stretching and maintaining range of motion without being too hard on the muscles. Some high intensity exercise, such as jumping, 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 activities and exercise that has been recommended by your physical therapist or PM&R doctor. Below is a table of activities that are generally considered safe or unsafe:

Safe & recommended activities	Unsafe activities (avoid if possible)
Swimming & aquatic therapy	Jumping on a trampoline or in a bouncy house
Assisted cycling	Contact sports
Boy/Girl Scouts	
Martial arts (Taekwondo or karate)	
Yoga	

Tips for AFO compliance

- Wear moisture wicking socks underneath to help with reducing heat and sweat.
- Establish a reward system to encourage regular night splint wear.
- If your child still has trouble tolerating the night splints, then have them only wear one a night and alternate legs each night.
- If they still have trouble tolerating them, use them for positioning during the daytime when seated or lying (they should not be used during walking activities).

Energy & fatigue

We often receive questions from families regarding energy levels such as: “how much is too much activity?” or “what should I do when my child says they are tired?” Your rehabilitation team may recommend thinking about energy conservation in your daily routine, which involves doing activities with the least amount of effort and conserving energy for activities that are more important to your child. For example, plan ahead to allow for some rest breaks and have your child rest before they get tired. An example of this may be riding in a wagon on the way to the park, so at the park they have energy to play with their friends. We want your child to have energy for the things that are important to them in their daily life!

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 overeat. 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.



Hydration

Staying hydrated is an important part of caring for Duchenne, too. In Duchenne, the fragile membranes around muscle fibers are at higher risk for breakdown. As a result, illness or increased physical activity can lead to breakdown of muscle that can progress to rhabdomyolysis, the symptoms of which can worsen if an individual is dehydrated. To help your child stay hydrated, be sure to offer frequent sips of water, and offer additional fluids with extra activity, warmer weather, or during times of illness.

If your child experiences myoglobinuria, which is when urine turns a dark brown color like tea or Coca-Cola (which is different from dark yellow urine, which is a sign of dehydration), call your child’s neuromuscular doctor for advice and be sure to aggressively hydrate.

Usually, with aggressive hydration, the urine will return to its normal color by the 3rd pee. If the urine does not return to its normal color by the 3rd pee, and rhabdomyolysis is suspected, make your way to the emergency room. Rhabdomyolysis should be treated as a medical emergency. As with any emergency, be sure to bring

your PPMD emergency card or download the PPMD mobile app to show to the emergency room staff. It is also important to call your NMS to ensure they are aware of the emergency and can work with the emergency room staff to develop a care plan.

See more information about how to prepare for a medical emergency on page 32.

Bone health

Weakened muscles in Duchenne along with steroids put children at low bone density, or osteoporosis. Your RDN can review your child's diet to make sure they are getting enough vitamin D and calcium. 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
- Complains of sensory issues such as discomfort with clothing or equipment

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 several 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 school'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.

A note about sensory issues

Many children with Duchenne are sensitive to sensory stimuli. You may notice your child being particular about wearing certain clothing or accessories (i.e., socks, shoes, AFOs). It can be explained as once your child is “stuck” on something sensory, they can't easily shift their attentional focus, which results in them hyperfocusing on whatever sensory experience they are noticing (i.e., their socks not feeling quite right). This is the same process as your nose itching when you are prevented from scratching it—until you get distracted and your attention shifts. Children with Duchenne can have trouble with attentional control, which leads to difficulty managing sensory experiences. This can be improved by working on eliminating “comfort chasing” with support from a psychologist or counselor.



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, can lead to lower-than-normal heart function and heart failure over time. When the heart function is lower-than-normal, there is also a risk for an abnormal heart rhythm, called an arrhythmia. Heart failure means the heart is having difficulty meeting the energy needs of the body on its own and importantly, can be managed with medication and monitoring for a very long time. Because of the effect of Duchenne on the heart, it is important that your child is followed by a heart doctor (cardiologist) on a regular basis.

Cardiac testing, including an electrocardiogram (ECG) and echocardiogram is recommended at diagnosis, and then once per year. Your child's cardiologist will discuss a cardiac MRI to evaluate your child's heart muscle as they get older. If there are any changes found on cardiac testing, your child's cardiologist will reevaluate whether they need to be seen at more frequent intervals. An ECG looks at the electrical conduction of your child's heart. Both the echocardiogram and cardiac MRI imaging look at the structure and function of your child's heart muscle. Cardiac MRIs are considered the “gold standard” for cardiac imaging in Duchenne, as your child ages, because your cardiologist is able to see early changes in the heart muscle called fibrosis (scarring), and if found, this may change medical management.

If your child's heart function decreases, their cardiologist will discuss starting cardiac medication. There are several types of cardiac medication used to help the heart muscle function. We have learned that in Duchenne, starting cardiac medication before the heart function decreases can be helpful in delaying this change from occurring. Given this knowledge, your cardiologist is likely to discuss starting cardiac medication for your child when they are young, typically before 10 years of age.

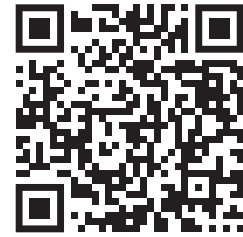
Breathing muscle health

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 every 6 to 12 months after 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 lung volumes and 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. Your pulmonologist will also recommend a baseline sleep study to be performed a few years after the time of diagnosis.

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 should I 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. When you are in an emergency situation, remember to THINK (parentprojectmd.org/think) and keep this information with you at all times:



When you are in an emergency situation, remember to THINK.

- **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.



DUCHENNE MUSCULAR DYSTROPHY EMERGENCY CARE INFORMATION FOR HEALTHCARE PROVIDERS

RESPIRATORY CARE
Risk of respiratory failure. Do not give Oxygen without close monitoring of CO2 levels. Breathing may need to be supported (non-invasive ventilation). Use cough assist machine if needed and available.

LEG FRACTURE TREATMENT
Risk of pain, loss of ambulation, FES. If ambulatory before leg fracture, surgery is preferred over casting to preserve ambulation (i.e., internal fixation with rapid weight bearing). Following a fracture or body trauma, watch for signs of Fat Embolism Syndrome (FES) including fast breathing and/or confusion.

STERIODS
Risk of adrenal crisis. Please refer to the P.J Nicholoff Steroid Protocol (parentprojectmd.org/pj) for stress dosing. Watch for signs of adrenal crisis during times of severe illness, injury, or surgery.

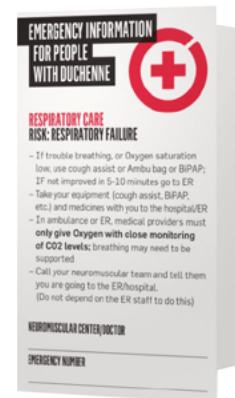
ANESTHESIA PRECAUTIONS
Risk of rhabdomyolysis. Inhaled anesthesia can cause rhabdomyolysis among other serious complications (i.e., cardiac arrest) in patients with Duchenne. When possible, inhaled anesthesia should be avoided. IV anesthesia is considered safe. Use all anesthesia with extreme caution after discussing with the anesthesiologist. Succinylcholine should never be used. Local anesthesia and nitrous oxide are generally safe for minor dental procedures.

GENERAL RECOMMENDATIONS

- Consider long term steroid therapy when administering live vaccinations.
- AST/ALT are normally elevated in patients with Duchenne and need no further evaluation.

FOR HEALTHCARE PROVIDERS

Parent Project Muscular Dystrophy



EMERGENCY INFORMATION FOR PEOPLE WITH DUCHENNE

RESPIRATORY CARE RISK: RESPIRATORY FAILURE

- If trouble breathing, or Oxygen saturation low, use cough assist or Ambu bag or BiPAP. If not improved in 5-10 minutes go to ER.
- Take your equipment (cough assist, BiPAP, etc.) and medicines with you to the hospital/ER.
- In ambulance or ER, medical providers must only give Oxygen with close monitoring of CO2 levels; breathing may need to be supported.
- Call your neuromuscular team and tell them you are going to the ER/hospital. (Do not depend on the ER staff to do this)

NEUROMUSCULAR CENTER/CLINIC

EMERGENCY NUMBER

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 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. People with Duchenne are at risk for low bone density and prone to fractures, and this risk increases with steroid use. If your child has a leg fracture, ask for a referral to an orthopedic physician with experience in treating patients with 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 in the **consider steroids** section (page 25), 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 it 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).

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 manifesting carriers.

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.



Planning for your future

PPMD is working to advance care, therapy options, and innovative research for all people living with Duchenne and Becker. We are hopeful for children being diagnosed and believe the trajectory of their diagnosis will be impacted by these advancements. However, it is never too soon to start planning ahead to set your child up with success to lead a full, independent life.

Facilitating independence

Facilitating independence in your child from a young age can have a huge impact on their quality of life. Teaching your child to advocate for themselves at home, school, and in public is a great way to start. Consider including caregivers outside the immediate family (i.e., babysitters, grandparents, friends) regularly and allowing your child to direct their own care. This will help them understand that people besides mom, dad, and siblings are able and willing to help.

Home safety & modification

We often receive questions about home modifications that should be considered with young kids living with Duchenne in mind. Below are a few general recommendations:

- Remove throw rugs, unsecured cords, or anything that is potentially a tripping hazard.
- Limit overuse of stairs if possible.
- Consider purchasing foldable temporary ramps for small steps around the house, such as deck and/or garage access. These ramps can be taken in the family car for access in public, as well.

Financial planning

There are several ways to ensure that your child receives services for needs in the future. Medicaid waiver programs exist from state and federal governments that offer support and services to disabled individuals. These programs often have wait lists many years long, so it is recommended you put your child on the interest list early even if they will not require services until much later. Your clinic's social worker can help you apply for and navigate these programs, as every state is different. You and your family are also encouraged to speak with PPMD staff, as we are happy to help you navigate benefit planning that is in the best interest of your loved ones.

Once your child is an adult, there is an income threshold to be eligible for benefits such as Medicaid and SSI. In order to protect your child from losing these benefits and services while still saving for their future, you may choose to open a supplemental needs trust. There are several different types, and it is recommended that an estate attorney assist in choosing which one(s) is best for your family.

Live in the now

Although it may be helpful to think about the future, it's important to remember your child is just that, a child. Children living with Duchenne and Becker are resilient and will find creative ways to explore their interests and continue to play and participate with their peers. Modern technology allows for more accessibility than ever before at home, school, and in your daily life. Do not hesitate to reach out to careteam@parentprojectmd.org with any questions, concerns, or to seek guidance on your individual child and their unique needs.

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 can seem daunting, but your care team will be able to discuss potential options for your child. 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 (often referred to as an 'investigational product'). 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 the progression of Duchenne, 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.

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](https://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 of a potential therapy 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.

DUCHENNE AND BECKER CLINICAL TRIAL WORKSHEET

RATE IT: PRO

	How likely is this, in your own opinion?	How good is this to you?	How much does this matter to you?
Scientists may gain important knowledge about Duchenne.	Very	★★★★★	★★★★★
The researchers may find a treatment that works.	Very	★★★★★	★★★★★
	Very	★★★★★	★★★★★
	Somewhat	★★★★★	★★★★★
	Not at all	★★★★★	★★★★★
	Not at all	★★★★★	★★★★★
	Not at all	★★★★★	★★★★★
	Not at all	★★★★★	★★★★★

DUCHENNE AND BECKER CLINICAL TRIAL WORKSHEET

IS A CLINICAL TRIAL RIGHT FOR MY CHILD?

Many parents who enroll their child in a clinical trial say they are satisfied and glad they participated. We want you to be satisfied with your decision, too. Thinking about what matters most to you and your family will help you decide whether to join a trial or not.

Brainstorm

What are some possible pros and cons of joining a clinical trial? Take a moment to list the things that matter most to you. Use one box for each pro or con you list. You can create as many boxes as you need.

Pros
Enter your pro
Add pro

Cons
Enter your con
Add con

Next

The deliberation tool development was supported through an award from the Patient-Centered Outcomes Research Institute (PCORI), PPRN-1309-0400.
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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. If you are seeking more detailed information on a specific potential therapy, PPMD maintains a detailed pipeline of all actively studied Duchenne therapies on our Interactive Drug Development Pipeline page, detailed in the following sections.

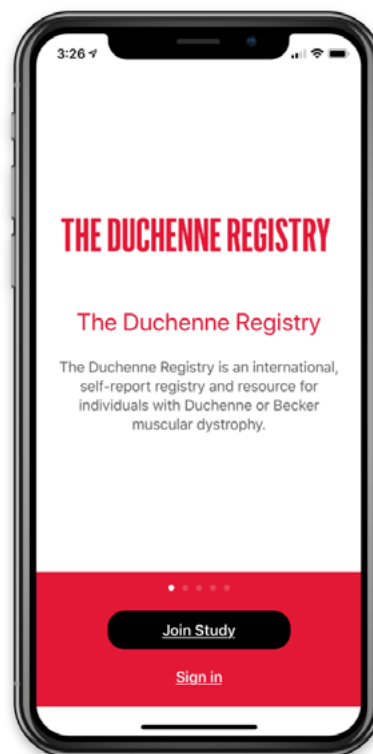
PPMD has many valuable resources that aim to connect people with Duchenne with clinical trial opportunities if they choose. There is a Clinical Trial Video Library available that includes several videos that explain the various aspects of exploring the participation of clinical trials. To visit this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



NOTES

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.

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.

—Pat Furlong, Founding President & CEO



New Diagnosis & Early Care Guide Contributors

Pat Furlong, *Parent Project Muscular Dystrophy*

Amanda Wilkison, RN, BSN, *Parent Project Muscular Dystrophy*

Rachel Schrader, MS, APRN, CPNP-PC, *Parent Project Muscular Dystrophy*

Alexis Hazlett, MSN, RN, CPN, *Parent Project Muscular Dystrophy*

Ann Martin, MS, CGC, *Parent Project Muscular Dystrophy*

Kayla Quirin, MS, CGC, *Parent Project Muscular Dystrophy*

Niki Armstrong, MS, CGC, *Parent Project Muscular Dystrophy*

Eric Camino, PhD, *Parent Project Muscular Dystrophy*

Nicole Herring, *Parent Project Muscular Dystrophy*

Patrick Moeschen, *Parent Project Muscular Dystrophy*

Brian Denger, *Parent Project Muscular Dystrophy*

Mena Scavina, DO, *Parent Project Muscular Dystrophy*

James Poysky, PhD, *Katy Child Psychology Associates*

Lauren Treat, MD, *Children's Hospital Colorado*

Rebecca Scharf, MD, *University of Virginia*

Michelle McGuire, PT, MPT, *Cincinnati Children's Hospital Medical Center*

Anne Connolly, MD, *Nationwide Children's Hospital*

Natalie Truba, PhD, *Nationwide Children's Hospital*

Carol Wittlieb-Weber, MD, *Children's Hospital of Philadelphia*

Mai ElMallah, MD, *Duke Children's Hospital*

Parent JOIN THE FIGHT.
Project END DUCHENNE.
Muscular
Dystrophy

1012 14th Street NW, Suite 500

Washington, DC 20005

800.714.5437

ENDDUCHENNE.ORG

