



pmsf.org



# *Next Steps*

**A Parent's Guide** to Living with  
Phelan-McDermid Syndrome



# There is Hope

## A LETTER FROM A MOM:

*Nothing can prepare you for your child's diagnosis. It doesn't matter their age or if you saw it coming, it's painful either way. My daughter was diagnosed at 15 months old, but I saw the red flags at 4 months. The day we heard "Phelan McDermid syndrome" for the first time is forever etched in our minds and hearts. My husband and I felt a myriad of emotions; oddly enough, it was relief we felt first. I spent months hardly eating, trying to get her doctor to take my concerns seriously, and falling down the rabbit hole researching symptoms at all hours of the night. I didn't know it then, but I was already becoming my daughter's advocate, which has become my greatest passion... You'll find your fighting voice and learn you often have to go with your gut on this journey.*

*The PMS community is like coming home. We were so lost before and immediately after diagnosis. We didn't have any friends or family who knew what we were going through. When I requested to join the family support group on Facebook, I immediately unlocked friendships from all over the world! We found our people, our village, our home. I'm eternally grateful for the PMS families who picked us up from the trenches in the early days. Everyone is so willing to help share their experiences, give advice, and answer questions no matter the time of day. This community of parents, caregivers, siblings, and other family members will take your burdens and carry them with you.*

*I want you to know it's okay to feel grief and joy at the same time. Try to process that again because it is SO important. I didn't know that in the beginning. It's okay to be human. You'll love harder, cheer louder, and take nothing for granted. Your new "normal" will be beautiful.*

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# First Steps After Diagnosis

Often, when you first receive your child's diagnosis, it can feel as if his or her whole life is flashing in front of you, with an infinite list of concerns. We've created a suggested "to-do" list to help you tackle immediate priorities.

## 1 LOVE YOUR BABY!

Your child will feel your love when you cuddle, laugh, and play. The diagnosis does not define your child, they are still the child you love.

## 2 BUILD YOUR PARENT NETWORK WITHIN PMSF AND LOCALLY.

You will discover quickly how welcoming, helpful, and compassionate the PMSF family is. We celebrate each other's children's successes, support one another through the challenges, and share information so you know you are supported as you build a family life with PMS.

## 3 BUILD YOUR CARE TEAM.

Depending on your child's needs, your child may need to be seen by additional specialists, such as a pulmonologist, gastroenterologist, ENT specialist, nutritionist, geneticist, or developmental pediatrician. You need not schedule all of these medical appointments at once. Work with your medical providers to come up with a plan.

## 4 SEEK EARLY INTERVENTION.

Ask your child's pediatrician or primary care professional about early intervention therapies that are available in your area. These may include occupational, physical, speech, and play therapy. If your child is older, there may be other options such as Hippo-therapy or aqua-therapy. You will meet wonderful caring professionals who will support your child and your family in a way that is fun and nurturing.

## 5 START KEEPING NOTES AND RECORDS AND BRING THEM WITH YOU TO MEDICAL APPOINTMENTS.

Keep a progress journal. Our kids advance at their own pace and sometimes it feels like they aren't progressing at all. Then, all of a sudden, they surprise you and make strides. You will have a sense of great joy when they take a first step or make a first sound, or first word.



# Frequently Asked Questions

## ABOUT PHELAN-MCDERMID SYNDROME

### 1 WHAT IS PHELAN-MCDERMID SYNDROME (PMS)?

Phelan-McDermid syndrome is a rare genetic condition caused by a deletion or variation of the long arm (terminal end) of chromosome 22 in the 22q13 location most often including the *SHANK3* gene, or a disease-causing (pathogenic) change (variant) of the *SHANK3* gene.

### 2 HOW DOES PMS AFFECT PEOPLE?

PMS may cause several different health conditions, including intellectual disability, absent or delayed speech, sensory or autistic behaviors (repetitive behavior, jumping, clapping, and lack of eye contact), low muscle tone, problems eating, constipation, and having high pain tolerance. Less common health concerns may include kidney abnormalities, seizures, gastroesophageal reflux (acid reflux or GERD), and heart defects. PMS is a highly variable condition, meaning those affected with PMS will likely show different symptoms of the condition. The symptoms may be present in varying levels of severity, making it difficult to predict which symptoms a person diagnosed with PMS will have.

There are some specific facial features and body features that are seen more commonly in those with PMS. Many of these features have no impact on someone's overall health but are things a doctor may look for when considering a diagnosis of PMS. For a complete list of these features and symptoms, which vary across individuals, go to [this link](#). Not everyone with PMS has the same features.

Some of the primary concerns caregivers have about having a child with PMS include behavioral difficulties, sleep issues, and communication limitations. Other concerns include regression of skills and seizures. Despite these challenges, children and adults diagnosed with Phelan-McDermid are often described as beautiful, courageous people with engaging personalities.

### 3 HOW IS PHELAN-MCDERMID SYNDROME DIAGNOSED?

A suspected PMS diagnosis is confirmed with a blood test that looks for the genetic abnormalities that are specific to Phelan-McDermid syndrome. Two types of tests are used to identify all types of PMS: chromosomal microarray (CMA) or Whole Exome Sequencing (WES). Blood may also be drawn from the parents to confirm a new (*de novo*) condition vs. a mosaic or inherited cause. You can find [information on genetic testing](#) on the PMSF website.

### 4 BASIC GENETIC TERMS EXPLAINED

To better understand the testing, a brief review of genetics may be helpful. Within each cell of the body, there are chromosomes, where genetic information (DNA) is stored. There are 46 total **chromosomes** in each cell that can be further divided into 23 distinct pairs. One copy of each pair is inherited from the mother, and the other copy is passed on from the father. On each chromosome are stretches of DNA that contain **genes**. Genes are important in helping the body develop, grow and function. Genes are responsible for each person's unique features, such as hair color and eye color. Some genes are very important in the development and function of the brain from very early in pregnancy. If these important genes do not work properly, the brain may not work as expected.

### 5 WHAT TYPE OF GENETIC TESTS ARE USED TO DIAGNOSE PMS?

The diagnosis of PMS requires genetic testing. Most people are diagnosed using a genetic test called chromosomal microarray analysis (also called SNP array, array-CGH or comparative genomic hybridization). This test is responsible for finding extra or missing pieces of genetic information by scanning each chromosome of an individual's genetic make-up. For a diagnosis of PMS, the microarray testing will show a deletion (loss) of genetic information on chromosome 22q13 that includes part, or all, of the *SHANK3* gene.

Another helpful test used to diagnose PMS is the chromosomal karyotype. This type of genetic testing provides a picture of the structure of the chromosomes. Chromosomes are typically in a "stick" or "line" form. About 10% of the time, individuals with PMS will have a ring (circular) structure of chromosome 22. When the ring forms, a portion of genetic information may be deleted from the area of 22q13. Individuals diagnosed with PMS due to a ring chromosome 22 should be followed more closely by their doctors for other potential health concerns. A genetic test called fluorescent *in situ* hybridization, or FISH, may also be used when looking at chromosome structure. FISH can identify a deletion of genetic information or a swapping of genetic information between chromosomes (called a "translocation") that can result in a diagnosis of PMS.

The last type of genetic testing that is commonly used to diagnose PMS is called sequencing. This type of testing involves reading the *SHANK3* gene from start to finish, looking for any spelling changes (variants) that may cause

the gene not to work properly. The *SHANK3* gene is important in the function of the brain. If a disease-causing variant is found, a diagnosis of PMS can be made.

## 6 HOW COMMON IS PMS?

In the United States, a rare disease is defined as a condition that affects fewer than 200,000 people in the US. Other countries have their own official definitions of a rare disease. In the European Union, a disease is defined as rare when it affects fewer than 1 in 2,000 people. Although considered a “rare” disorder, Phelan-McDermid syndrome is one of the most identified genetic causes of autism accounting for an estimated 1-2% of autism diagnoses. PMS is found in people of both sexes and all races.

## 7 IS PMS INHERITED?

Most cases of PMS are random occurrences (called *de novo*), meaning “new” and not inherited. These cases are generally not associated with an increased risk of recurrence in future pregnancies. A family should see a geneticist to discuss their particular situation and to seek information on their personal recurrence risk.

## 8 ARE THERE DIFFERENCES IN THE SEVERITY OF PMS BASED ON THE GENETIC DELETION SIZE?

At this time, there is no consistent evidence that the severity of PMS is related to genetic subtypes. Natural History research and genotype/phenotype research is evolving.

## 9 IS PHELAN-MCDERMID SYNDROME CURABLE?

Currently, there is no cure for PMS. Early diagnosis and optimal clinical care can improve outcomes, but increased research, funding, and understanding of PMS offer hope for more effective treatments that may lead to a cure. The PMSF is committed to bringing the family voice to the table as active members of the scientific, clinical, legislative and industry communities.

## 10 ARE ANY TREATMENTS AVAILABLE FOR PHELAN-MCDERMID SYNDROME?

Individuals with PMS are treated for specific symptoms and co-existing conditions. Treatment and management of symptoms may come from a team of medical specialists including neurology, developmental pediatrics, gastroenterology, nephrology, endocrinology, sleep specialist, or cardiology. Your child or adult with PMS may also receive supportive therapies such as physical therapy for gross motor development, occupational therapy to assist with fine motor skills (including feeding difficulties), and speech therapy for communication. Children with PMS may receive some, or all, of these services either through insurance or their school programs.

## 11 WHAT SHOULD I DO MEDICALLY?

Search for a primary care doctor who is willing to learn more about PMS and help you get referrals and specialty care when needed. First, familiarize yourself with the PMS Clinical Care Practice Parameters that are linked in the resource section of this guide and on the PMSF website. You may share the simple infographic with your care providers to give an overview of the possible ways PMS may affect people. Remember, everyone is different, so not all children have the same presentation of PMS.

Some suggestions from our medical advisors are to have a renal ultrasound and echocardiogram within 3 months of diagnosis. Test your child’s hearing and vision within 6 months of diagnosis, but within 3 months if the child is less than 4 years old or parents have concerns about vision or hearing. Developmental and neurological evaluations should be done within 6-12 months. A neurologist can then order an EEG and an MRI, if needed.

Be flexible on the timeframe to avoid undue stress to an already stressful situation.

## 12 WHAT CAN I EXPECT OF MY CHILD?

No two children are exactly alike, so remember to see your child and try not to be overwhelmed with the potential limits of the diagnosis. Read our family stories about our children who are diagnosed with Phelan-McDermid syndrome.

People with PMS enrich the world. They attend school, develop outside areas of interest, have friends, participate in the community and family events, even move away from their family home. They do, however, need a significant amount of support from their families and from school, work, and adult service providers throughout their lives.

## 13 WHERE DO I GO LOCALLY FOR HELP?

If you’re the parent of an infant or young child, you’ll want to contact your local health department to see what services you qualify for as a family that is caring for someone with special needs. Ask about Early Intervention services for infants and pre-school age children. Ask about your state’s Autism Waiver program if your child is diagnosed with autism spectrum disorder.

For school-age children, check with your school district and school social worker about what types of supports and therapies are available for children even before they attend school. Your school district will set up Individual Educational Plans that set out how your child will be taught and which therapies will be available for each year they’re in school.

For newly-diagnosed adults, educational/vocational transition plans, legal planning, adult living and working situations become important. Start with our family FB pages, your local regional REP and local parent groups, to help guide you through these important steps.



# FAQ CONT.

## 14 WHAT CAN I DO TO HELP ADVOCATE FOR MY CHILD?

Educating yourself about your child's rights to education and your state's education and waiver programs is a good place to start. Remember, you don't have to do everything at once, so pace yourself. Try to join local parent support groups that are recommended by your pediatrician or support service provider. Other parents can be not only a great source of support, but can help guide you to services that have been helpful to them in their journey.

## Advice FROM OTHER PARENTS OF PMS CHILDREN:

- *Don't compare your child to typical children their age. Let them progress at their own pace and enjoy every triumph!!!!*
- *Allow yourself to grieve for the child you thought you'd be having. This doesn't mean that this little treasure you have is any less loved! But there is sadness and that's OK.*
- *Ask for help! This is one of the most difficult things for people who are usually self-reliant. However, remember that this is new territory for your family and it will require some help to create your new normal. Ask for someone to watch your other children for a while, get a mothers-helper for an hour while you make dinner, have groceries delivered once in a while.*
- *Be kind to yourself. You did not do anything to cause your child to have Phelan-Mcdermid syndrome. You will go through ebbs and flows of emotions, so let them come and give yourself and others the time and kindness to process.*
- *Remember that this is a marathon not a sprint, so take the time you need for every step. Don't wear yourself out trying to get everything done at once.*
- *Keep seeing your friends. You have entered a whole new world, and your friends don't understand it. But in years to come when the therapists have left and you just feel like being your fun old self, you will be so happy if you can still call your friends to socialize.*





# There is *Help*

*We are always an email or phone call away. PMSF will help you connect with other families in your area and worldwide. Our network of families will become a valuable resource to you as you navigate your way through creating a life for your child and your family. You will be assigned to a geographical region and will be invited to local gatherings where you will be able to meet other families who share your experiences with your own child and who are familiar with local resources.*

*Families tell us that gathering either virtually or in-person is one of the most helpful benefits of being in this community. You will grow to know you are among friends and even “family” as many refer to our community. PMSF organizes an international family conference where families, clinicians, researchers, and others in the PMS support community gather every other year. It is a life-changing event for families and we always have an overwhelmingly positive response.*

*PMSF has Scientific and Medical Advisory Committees who support and inform our community about the latest research opportunities and best clinical care practices.*

*The PMSF website includes abundant resources for families, we publish a monthly newsletter, we have published video resources including family stories, scientific and medical presentations, and webinars. We always seek input from families to know what you need. We celebrate the accomplishments of each individual and realize that we are all stronger when we work together for a common cause.*





# Get Involved



## FAMILY SUPPORT

Phelan-McDermid syndrome is a rare disorder, but we are committed to expanding global knowledge about PMS. PMSF has created a peer-network support model that encourages experienced family members to share with others through use of social media, regional gatherings and international conferences. Our community is growing quickly with the advent and availability of genetic testing and increased awareness. We encourage anyone with a genetic variant of the 22q13 region or a *SHANK3* variant to [join PMSF](#) to become part of our support network.

## RESEARCH

PMSF, our patient advocacy group, has a seat at the table with researchers and industry to represent the patient voice of our community. One step you can take toward becoming involved and informed about research is to [enroll in the DataHub](#). There are many other ways you can get involved. Reach out if you are interested.

## ADVOCACY

PMSF is the largest PMS patient advocacy network. Our membership and our voice reach around the world. We have engaged with other rare disease groups to amplify our voice and ensure we are at the forefront of the developments in legislation, funding and social actions that affect our families.

## FUNDRAISING AND AWARENESS

The Phelan-McDermid Syndrome Foundation is committed to raising much needed funds to improve the lives of everyone affected by PMS. Many of our families tell us that because our lives can be so out of control, participating in a group or individual effort to raise funds or awareness can offer some sense of relief and knowledge that you can contribute to hope and solutions.

If or when your family is ready, there are many awareness and fundraising opportunities to help support our mission. Each family can choose how they participate and when. Here are two examples of how our community celebrates all of our PMS children:

- October 22nd, PMS Awareness Day, is one day a year that you will be certain you are not alone. When we SHINE Green, it feels like a big world-wide hug. [View video](#)
- Our premier Phelan-Lucky t-shirt campaign was started by a mom for her son, Jack. She wanted to do something with what felt natural to her family. She turned an annual St. Patrick's Day gathering with a few friends into a global event. [Learn more](#)

Fundraising is a major lifeline of our organization. We are forever grateful to those that support PMSF. Have an idea or need help setting up a fundraiser? Contact our [Director of Development](#).

## RESOURCES

These resources and graphics may be helpful for medical appointments and educational planning.

- [Phelan-McDermid Syndrome Foundation](#)
- [Clinical Care Infographic](#)
- [About PMS and Genetic Testing Infographic](#)
- [Genetic Follow-on Testing](#)