



# FIRST STEPS

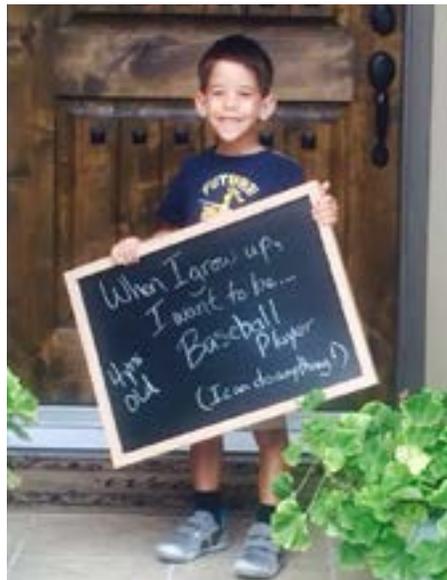
A PARENT'S GUIDE TO PRADER-WILLI SYNDROME

# First Steps: A Parent's Guide to Prader-Willi Syndrome

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# A Letter of Hope

**First Steps: A Parent's Guide to Prader-Willi syndrome was written by parents for parents.** We are so glad you found us! This book is a way to share the gift of hindsight: what we wish we'd known when we first received our child's diagnosis, or what we did learn that was so very valuable and helpful as we began our journey with Prader-Willi syndrome within our own families.

Most likely, you are the parent or family member of a child recently diagnosed with Prader-Willi syndrome. In this moment, two things are true: (1) You have a beautiful child who will bring you great joy and has the opportunity to experience a beautiful life, (2) You are not alone in your journey nor with the challenges of PWS.

My own family has come a long way since receiving our son's diagnosis. When we were in the NICU, we were two scared parents who felt as if life was over as we knew it. Our dreams for our family had been shattered, and we were mourning the child we had dreamed of having.

For us and for most parents of children with PWS, diagnosis day has become unforgettable. It was the heartbreaking day an imagined future for our new baby or child was forever changed. It was an overwhelming day when we first heard terms like "failure to thrive," "hypotonia," and "hyperphagia" and struggled to consider how they might manifest in our little boy or girl.

It was also a day of discovering a fierce determination to do whatever we could to provide the best possible care and the brightest future for our child. It was a day of many questions: What do we need to do? Who do we need to talk to? What does this diagnosis mean for our child and family?

I wish we would have spent less time looking at our son as a diagnosis and spent more time just cuddling



and enjoying him for who he is. I wish we could have been able to see a clip of what life was going to be like now, back then. We never could have dreamed Jayden could change our lives the way he has. He has brought more joy and love to our family than we ever imagined possible!

While the list of potential challenges associated with Prader-Willi syndrome is long and sometimes difficult to consider, the parents whose voices you'll hear in this book are committed to doing all we can to ensure our children live happy, healthy, fulfilling lives. Luckily, we have three tremendous resources working in our favor:

1. an incredibly supportive parent network
2. an ever-expanding group of medical and research professionals who are working diligently to provide optimal care and treatments for our children
3. access to the most current information about effective practices for managing Prader-Willi syndrome.

If there is one piece of advice that most of us would give to families with a new diagnosis of PWS, it is to not believe everything you read about PWS. All too often, the picture painted of PWS is so bleak that it can overwhelm families to the point of despair. If that is where you are, we are here to say that THERE IS HOPE! Our hope is that First Steps will connect you to helpful resources, alleviate some of your fears about your child's future, and empower you as you move forward.

—Susan Hedstrom  
Executive Director, Foundation for Prader-Willi Research

# What Is Prader-Willi Syndrome?

Prader-Willi syndrome (PWS) is a rare genetic disorder that occurs in approximately one out of every 15,000 births. PWS affects males and females with equal frequency and affects all races and ethnicities. PWS results from an abnormality of chromosome 15, and it is a chance occurrence. *Nothing that parents do either prior to conception or during the pregnancy causes PWS.*

**When reading about the symptoms that are associated with PWS, it's important to keep in mind that PWS has a broad spectrum.** Although many symptoms can be associated with PWS, most individuals do not experience all of the possible symptoms. In addition, the range of severity is very broad. Many parents understandably want to know where their child will fall along the spectrum of PWS, but right now it's not possible to tell ahead of time which symptoms will happen in any particular individual, or to what degree. We do know that all children with PWS benefit from early intervention, a loving environment, good social interaction and an informed and caring medical team.

Infants with PWS often have low muscle tone (hypotonia). They may have a weak cry and a poor suck reflex, and can be very sleepy. Babies with PWS usually are unable to breastfeed and may require special bottles or tube feeding. As babies with PWS grow older, strength and muscle tone generally improve. Motor milestones are achieved, but may be delayed.

Unregulated appetite (hyperphagia) and easy weight gain commonly begin between ages 3 and 8 years old, but are quite variable in onset and intensity. In addition, the metabolic rate of persons with PWS is lower than normal. Maintaining a healthy weight can be challenging, but is achievable if there are consistent rules about food, and if family, friends and teachers work together to maintain a healthy and controlled food environment.



## What Is Prader-Willi Syndrome?, continued

A variety of other clinical features can also be associated with PWS. Individuals typically exhibit cognitive challenges, with IQs ranging from the low normal range to moderate intellectual disability. Those with normal IQs usually have some learning disabilities. Other clinical problems may include growth hormone deficiency/short stature, scoliosis, sleep disturbances with excessive daytime sleepiness, high pain threshold, speech apraxia/dyspraxia, and infertility. Behavioral challenges can also arise, and may include increased anxiety, obsessive-compulsive symptoms, skin picking, and difficulty controlling emotions. Adults with PWS are at increased risk for mental health issues.

Although the symptoms associated with PWS are daunting, there are many reasons to be hopeful. Acting proactively to become educated about PWS and to set up a good environment will help your child thrive. In addition, research offers new possibilities to eliminate the challenges of PWS. There are a variety of [new drugs currently in development](#) for control of appetite in PWS and most of this progress has occurred in the last few years. There have also been recent advances in technologies to manipulate DNA, which have led to the possibility of gene-based strategies to treat PWS at the molecular level. [Conventional drugs and devices](#) are being developed and tested to alleviate some of the more troubling aspects of PWS including hypotonia, hyperphagia, and behavioral problems. Research has brought us a wealth of opportunities and hope for the days ahead, and the number of potential new therapies and intensity of investigation is promising.

More information on the symptoms of PWS can be found on the [“About Prader-Willi Syndrome”](#) page on the FPWR website.



# Frequently Asked Questions

## How common is PWS?

PWS occurs in approximately one out of every 15,000 births. Although considered a “rare” disorder, Prader-Willi syndrome is one of the most identified cause of severe childhood obesity. PWS is found in people of both sexes and all races.

## Is PWS inherited?

Most cases of PWS are random occurrences and generally are not associated with an increased risk of reoccurrence in future pregnancies. In the case of an imprinting mutation, which is the rarest form of PWS, PWS can recur within a family. A family should see a geneticist to discuss their particular situation and to seek information on their personal recurrence risk.

## What causes PWS?

PWS occurs when information from one of an individual’s two chromosome 15s—the one normally contributed by the father—is missing. This can happen in three ways: (1) Most often, part of the chromosome 15 that was inherited from the person’s father is missing or deleted in this critical region. This small deletion occurs in approximately 70% of cases and usually is not detectable with routine genetic analysis such as amniocentesis. (2) Another 30% or so of cases occur when an individual inherits two chromosome 15s from their mother and none from the father. This scenario is termed uniparental disomy (UPD). (3) In a very small percentage of cases, a small genetic mutation in the Prader-Willi region causes the genetic material in that area to be inactive. This is referred to as an imprinting mutation.

## How is PWS diagnosed?

A suspected PWS diagnosis is confirmed with a blood test that looks for the genetic abnormalities that are specific to PWS. A DNA methylation test of the PWS critical region on chromosome 15 will identify all types of PWS and is the preferred test for diagnosis. The FISH (fluorescent in-situ hybridization) test identifies PWS by deletion but does not diagnose other forms of PWS. In cases where an imprinting mutation is suspected, blood may also be drawn from the parents. You can find information on genetic testing on the [FPWR website](#).



“Zach was barely 2 days old when we first heard the words Prader-Willi syndrome. We had very experienced doctors that said that he fit the newborn profile for PWS perfectly. It only took a little over a week to get the preliminary results back confirming what the doctors had told us. It was hard to tell our families, and I will never forget those moments. Having a place to go online to learn about PWS and having other parents to email and chat with about their children was priceless in the first crucial months as we began our journey.”

—Lindsay Mattingly, mom to Zach



“Ella was 4 weeks old when we received her diagnosis. I felt embarrassed, alone and afraid. We immediately began looking for answers — who was the best doctor to see? What were the interventions we should be putting in place? How could we help Ella? In our search we found FPWR and a community of people that could not only give us the answers we needed, but also HOPE.”

—Dana Capobianco, mom to Ella

## Frequently Asked Questions, continued

### Are there differences in the severity of PWS based on the genetic subtype?

At this time, there is no consistent evidence that the severity of PWS is related to genetic subtype.

### When does the overeating associated with PWS begin?

This is highly variable, however, an increased interest in food typically begins between 2 and 8 years old, with hyperphagia typically beginning sometime between the ages of 8 and adulthood.

### Is Prader-Willi syndrome curable?

Currently, there is no cure for PWS. Early diagnosis and optimal clinical care can improve outcomes, but only increased research offers hope for more effective treatments and an eventual cure. There are many reasons to be hopeful, though. FPWR is working to accelerate the development of new therapies for PWS, and a working list of therapies in development can be found here: <http://www.fpwr.org/therapeutics-in-development-for-pws/>

### Are any treatments available for Prader-Willi syndrome?

In multiple studies, human growth hormone (HGH) has been found to be beneficial in treating Prader-Willi syndrome. In June of 2000, HGH was officially approved by the Federal Drug Administration (FDA) in the United States for use in children with Prader-Willi syndrome. Additional studies have found positive results on development, behavior, and intellectual capability. In addition to hGH, early interventions and careful management of symptoms can improve development and decrease clinical problems.

### What does the future hold for people with PWS?

People with PWS can expect to accomplish many of the things their “normal” peers do—complete school, achieve in their outside areas of interest, be successfully employed, even move away from their family home. They do, however, need a significant amount of support from their families and from school, work, and residential service providers to both achieve these goals and avoid obesity and the serious health consequences that accompany it.



### If Only I Knew

*Keegan Johnson*

**If only I knew  
he could reach the top.**

**If only I knew  
how focused he would be  
on achieving his objectives.**

**If only I knew  
how competitive he would be.**

**If only I knew  
how happy Dante  
would be and how much  
happiness he would bring our family.**

**If only I knew  
how lovable he would be.**

**If only I knew Dante,  
I could have skipped a lot of the  
heartache and got right to loving him.**

**... but I guess that's the challenge of  
life. Most of the time we don't "know"  
until it is too late. The key, therefore, is  
to find something you can believe in.**

*Keegan Johnson, Board Chair of FPWR Canada and father of Dante, wrote this as part of a blog post when his son was 7 years old. [Read the entire blog post here.](#)*

# Parents Answer Your Questions

*The parents quoted in this section are past members of the FPWR New Parents Committee.*

## What will my child look like?

Children with PWS are beautiful. At first, I was so sad thinking that my child would “look a certain way because he has a syndrome.” Well, he looks a lot like his dad! Yes, there are certain features that are present that are certainly a result of PWS, but he is so incredibly handsome that it doesn’t bother us one little bit. My husband and I often talk about how beautiful kids with PWS really are.

— *Lindsay Mattingly*

When Cade was an infant, I use to always see “PWS,” but as time went on, he has changed so much, and while some characteristics of the syndrome are still there, a lot are not. He is turning into such a sweet little guy and so full of life!

— *Maegan Richard*

Your child is beautiful and will remain beautiful as he or she gets older. Because of growth hormone therapy, our children look just like any typical child. Personally, my Julianna looks like the beautiful little princess I always dreamed my little girl will be. — *Carole Elkhall*

## Will our family be able to go out to dinner?

We still go out to eat as often as a family with two small children would (not very often) but only because I like to cook! When we do go out, we always choose to go somewhere with healthy whole food options like a salad bar or grilled chicken and veggies.

— *Jen Bender*

I always wondered how we would manage going out for dinner and, well, the answer is, we just go prepared. We still go, and there is almost always something Cade can have ... we just adjust to his needs and all goes fine. It hasn’t been complicated for us yet.

— *Maegan Richard*

Yes! We choose healthy restaurants and come prepared. Jayden is grain-free and sugar-free so we choose options like a cobb salad or chicken with vegetables. We usually bring along some tomatoes and avocado just in case! — *Susan Hedstrom*



**Heather Osterman and Max**



**Sarah Peden with  
Lillian and Eric**



**Perla Mondriguez  
with Evan, Andres and Emilio**

## Our Parents Answer Your Questions, continued

### Will our family be able to travel again?

Yes! My husband's parents live seven to eight hours away from us, and we usually travel to see them twice a year. "Nana & Papaw's farm" is Zach's favorite place on earth! We have also been on a beach vacation every year since Zach was born. His first trip to the beach was when he was 9 months old! He sleeps with oxygen now because of sleep apnea, and we even just throw that in the car with us now and take off. He hasn't flown yet, but I am sure that is in his future. — *Lindsay Mattingly*

We've taken Lillian vacationing many times. When she was an infant, we traveled with her apnea monitor, feeding tube supplies, and growth hormone device. We flew to Florida to visit family and Arizona to see the Grand Canyon. She's been to Disney World, Hilton Head Island, Washington D.C., St. Louis and several beach vacations on the east coast. She's a great traveler! We even plan to take her on a trip out west this summer to see Yellow Stone National Park. Our family has always enjoyed traveling and plans to continue to do so. — *Sarah Peden*

### Will my child be smart?

We think Zach is very smart! He certainly has areas that he struggles with, but he does learn well for his age. He knew all of his colors, numbers, letters, and shapes at an early age. Even his therapists have always thought that he was a very smart boy! He is very social and loving, which makes our hearts smile too. — *Lindsay Mattingly*

Leah is extremely intelligent. She is 22 months and knows about 30 signs and uses words too. She can point out pictures in books, make people laugh with her witty personality, and tries to manipulate like any other toddler going through the terrible two's! I believe early intervention, a home rich in language, and loving your baby fosters intelligence in our kids. — *Jen Bender*

Your child will be as smart as you allow him/her to be. I think when we set high standards, they will follow through ... Cade has proved this true many times. — *Maegan Richard*



**Johnson Family**



**Unger Family**



**Hammill Family**

# 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 for the first few months/year to help you tackle immediate priorities.

1. **Love your baby!** Provide stimulation and valuable input into your baby's sensory system by holding him, bouncing her, swaying her, talking and singing with him, massaging her, and waking your baby at regular intervals for feedings. Our babies could sleep most of the day and night uninterrupted, don't limit yourself to loving them while they are awake!
2. **Stabilize feeding and weight gain.** Consider working with a team of professionals, including a speech therapist and nutritionist, who can provide valuable advice about feeding techniques and equipment such as bottles and nipples.
3. **Begin growth hormone therapy for your child.** There is evidence that there are significant benefits to starting growth hormone (GH) as early as possible. Start by making an appointment with an endocrinologist who is familiar with Prader-Willi syndrome and growth hormone therapy.
4. **Implement early intervention therapies,** which may include occupational, physical, and speech therapy.
5. **Build your parent network.** You will discover quickly how welcoming, helpful, and compassionate the Prader-Willi family is. We celebrate each other's

children's successes, support one another through the challenges, and share information about best practices and treatments to help build our collective knowledge about managing PWS effectively.

6. **Consult other specialists.** Depending on your child's needs, you may need to be seen by additional specialists, such as a pulmonologist, gastroenterologist, ENT specialist, nutritionist, geneticist, or developmental pediatrician.



"My PWS mantra is (no pun intended), 'Don't bite off tomorrow's troubles today'. When I find myself distracted, worrying about my son's life five, ten, fifteen, or twenty-five years from now, I remind myself that his future is yet unwritten. That the problems or issues I've read about online may not become his issues. That I can significantly affect his outcomes by diligently coordinating his care, by arming myself and his team of doctors with the most current information, and by helping advance the research that could identify cures or treatments for the scariest stuff. Remembering to stay in the present helps me redirect energy to productive activities, focus on what needs to be done now, face the unknown with a positive outlook, and most importantly, enjoy my beautiful baby boy today and every day." — Megan Catalfamo, mom to Benjamin

# Early Interventions

Early interventions should begin as soon as a diagnosis has been made. Early intervention in children under age 3 years, particularly physical therapy, may improve muscle strength and encourage achievement of developmental milestones. Physical, occupational, and speech therapies are recommended for infants with PWS.

Research indicates early diagnosis and therapies may reduce the duration of tube feeding and prevent early obesity in Infants with PWS.

## Physical Therapy

Physical is helpful to improve balance, coordination, and strength. Your physical therapist will work with your child to improve gross motor skills such as lifting his head, sitting, crawling, and walking.

## Occupational Therapy

Occupational therapy primarily assists with fine motor skill development and hand control. Occupational therapists will also perform activities to help with sensory integration and may perform oral motor exercises to improve sucking strength in preparation for pre-speech.

## Speech Therapy

Early assessment and intervention are critical to the development of functional communication. Parents are strongly encouraged to begin oral-motor therapy in infancy to assist with feeding and the acquisition of the oral-motor skills necessary for babbling and speech. If your child is receiving a diagnosis later in childhood, a speech and language assessment should be made as soon as the diagnosis is made. The Prader-Willi California Foundation provides an excellent [brochure on speech and PWS](#).

## Nutrition Therapy

Nutrition therapy can assist you in developing and adjusting a feeding plan for your child and is helpful in monitoring your child's growth. At these visits, a registered dietician or nutritionist will weigh and measure head circumference and body length. This ensures that the same person is calculating these measurements monthly and that these numbers are accurately recorded on your child's growth chart. This practitioner can then communicate these numbers to your child's primary care physician. Having an accurate recording of your child's growth is extremely important when he or she starts growth hormone therapy. A nutritionist or dietician experienced with Prader-Willi syndrome can also help you set up a meal plan to ensure your child is receiving the vitamins and nutrients needed for optimal development, while also considering your child's specific caloric needs.



# Early Interventions

## Hippotherapy

Hippotherapy is commonly recommended for children ages 2 and above. The horse's rhythmic, repetitive movements work to improve muscle tone, balance, posture, coordination, strength, flexibility, and cognitive skills. The movements also generate responses that are similar to and essential for walking. In addition, adjusting to and accommodating for the horse's movements increases sensorimotor integration. You can learn more at the [American Hippotherapy Association website](#).



## Aqua Therapy

Although there is little data supporting the implementation of aqua therapy for those with PWS, it is often recommended for strengthening muscles. Aquatic therapy is especially beneficial for people who have difficulty with weight bearing activities. Benefits of aquatic therapy include improved muscle tone and strength, endurance, cardiovascular function, balance, and coordination.



## Infant Massage

Research shows infant massage is beneficial in improving blood circulation, aiding digestion, enhancing the development of the nervous system, stimulating neurological development, increasing alertness, and improving immune function.

[View a video demonstration of infant massage here.](#)



# Growth Hormone Therapy

In multiple studies, human growth hormone (HGH) has been found to be beneficial for those with Prader-Willi syndrome. In June of 2000, HGH was officially approved by the Federal Drug Administration (FDA) in the United States and other countries for use in patients with Prader-Willi syndrome.

HGH is effective not only in increasing height, but also in decreasing body fat, increasing muscle mass, improving weight distribution, increasing stamina, and increasing bone mineral density. In addition, studies suggest positive effects on development and behavior.

There is evidence that HGH treatment improves cognitive performance. Children who have been on growth hormone prior to 4 years of age show a significant increase in IQ over the historic average. Adults treated with growth hormone have shown improvement in mental speed, mental flexibility, and motor performance.

International consensus guidelines for the use of Growth Hormone in PWS were published in 2011. This publication can be used as a guide for treatment using GH.

<https://www.fpwr.org/wp-content/uploads/2014/02/GHguidelinesJCEM2013.pdf>



## Related Videos

FPWR conference presentations by endocrinologist Jennifer Miller M.D.:

[Hormone Dosing](#)

[Cognitive Benefits](#)

[Benefits for Adults](#)

# Getting Support, Getting Involved

Here are some opportunities for you to get and give support in your PWS journey, and to help advance PWS research, awareness, and fundraising—when the time is right for you:

## Facebook

PWS families use our sponsored pages to connect, make new friends, and exchange support. Visit and like the [FPWR](#) and [One SMALL Step](#) Facebook pages to get connected.

## Conference

FPWR hosts an [annual conference](#) where we discuss the latest PWS research, learn how to become effective advocates for our loved ones, and connect with each other for support.

## Volunteering

FPWR volunteers help with conference planning and fundraisers, connect with other families via social media, and help advocate for PWS research, funding, awareness, and legislative support. [Fill out this form to get started.](#)

## Fundraising

PWS families host One Small Step walks and other events to raise funds to support PWS research. [Fill out this form to get started.](#)



“Our daughter was diagnosed with PWS at 7 weeks of age. When she was 4 months old, our family made the decision to host a One SMALL Step event. This decision was an investment in our daughter’s future, and it allowed our family to heal. Educate, advocate, celebrate, and empower...this is what One Small STEP for PWS means to our family. We were humbled by the outpouring love and support from our community, and can’t wait for our next event!”

— David, Gwyn, & Ellie Spearman



# Additional Resources

## Foundation for Prader-Willi Research

[www.fpwr.org](http://www.fpwr.org)

[www.fpwr.ca](http://www.fpwr.ca)

[OneSMALLStep.fpwr.org](http://OneSMALLStep.fpwr.org)

## Other PWS Organizations

[PWSA-USA](#)—PWS Support Organization

[IPWSO](#)—International PWS Organization

## Articles

[Health Supervision for Children with Prader-Willi Syndrome](#)

[Endocrine Manifestations and management of Prader-Willi syndrome](#)

[Nutritional Phases in Prader-Willi Syndrome](#)

[Growth Hormone Consensus Guidelines](#)

## Photos and Videos of People with PWS

[FPWR Photo Collection](#)

[FPWR Video Collection](#)



The Foundation for Prader-Willi Research (FPWR) was established in 2003 by a small group of parents who saw the need to foster research that would help their children with Prader-Willi syndrome lead more healthy and fulfilling lives. Today, FPWR is composed of hundreds of parents, family members, researchers, and others who are interested in addressing the many issues related to PWS, including childhood obesity, developmental delay, psychiatric disorders, and autism spectrum disorders.

The mission of FPWR is to eliminate the challenges of Prader-Willi syndrome through the advancement of research. High-quality research will lead to more effective treatments and an eventual cure for this disorder. By working together, we intend to free our loved ones from the burdens of PWS, allowing them to lead full and independent lives.

[Stay in touch! Subscribe to our blog.](#)



Foundation for Prader-Willi Research

340 S Lemon Ave #3620

Walnut, CA 91789

888-322-5487

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