We’ve created a suggested “timeline” to help you navigate through the process of your journey with PWS. Our message to new parents is that you enjoy your child. Live the life you were meant to live and your child will also live the life he or she was meant to live. Trust your perceptions and observations of your child’s abilities and disabilities. Your knowledge is a critical component in the diagnosis and understanding of PWS. You will become empowered to become the best expert and advocate concerning your child.
CONGRATULATIONS!

Congratulations on the birth of your baby! You are now the protector of a new and amazing little person who is bursting with world-changing potential!

Receiving a diagnosis of Prader-Willi syndrome may have ignited a wildly overwhelming number of concerns, fears, and questions about what the future holds for your child and family. The emotional chaos caused by this diagnosis may be overshadowing the excitement of welcoming your new arrival and THAT’S NORMAL.

Right now, you may struggle with fear of “the unknown.” Please take comfort in knowing that as this journey unfolds, you will discover that there are undoubtedly far more similarities between your child and children without this genetic disorder than there are differences. Your child has the same needs as a typically developing child, but may simply require a different route or method of fulfilling them, achieving a desired goal, or acquiring a given skillset. PWS simply defines your child’s medical condition; it does not define your baby’s capabilities or who he or she will grow to be.

Despite the cache of challenges PWS presents diagnosed children and their families, there are countless invaluable lessons in love, determination, acceptance, and advocacy to claim. Though your child had less than a 1% chance of receiving this diagnosis, there is no shortage of wonderful resources to help guide you as you seek exceptional care for your new bundle. In fact, there is an entire community comprised of other families who have taken the path you are about to embark on and who are a well of information and support, in conjunction with the Prader-Willi Syndrome Association (USA), which strives to address the needs of families spurred by each facet to this rare and complex genetic disorder.

I will be very candid with you by sharing that our family, like so many others, was deeply devastated by the Prader-Willi syndrome diagnosis and the devastation made us very slow moving to begin utilizing the resources offered by PWSA (USA). Reflecting on what exactly it was that held us back from information and support seeking has led me to think that maybe in our struggle to accept Grayson suffering from this genetic disorder, we somehow convinced ourselves that ignorance was bliss, that if we didn’t plant roots in the PWS community, it somehow made Grayson’s diagnosis false, and that we wouldn’t need the resources PWSA (USA) had available.

In our journey with Grayson, my reluctance in the decision to defer information and support seeking through PWSA (USA) has been my deepest regret. Hindsight being what it is, I wish I had dove headfirst into all the resources this wonderful organization has to offer because the information and support we acquired through the new parent package, contact with other families of children with PWS, and threads posted on PWSA (USA) Facebook pages, has lifted the cloud of loneliness and helplessness that can sometimes hover over families in receipt of this diagnosis. Once we became comfortable, we began using PWSA (USA) to find answers to our questions surrounding Prader-Willi syndrome; We gained a sense of direction and with the guiding support from PWSA (USA) and the PWS community were able to significantly improve on Grayson’s quality of care. We are so grateful for the support of PWSA (USA) because it has changed Grayson’s life.

We did, in fact, need this organization and community more than we could have ever imagined. And because there is strength in numbers, they need YOU! We encourage you to dive into all the resources PWSA (USA) has gathered – Ask questions, share information, and reach out to other PWS families so that our beautiful children can live life to the “fullest!”

-Sara

The Grosso Family
WHAT IS PRADER-WILLI SYNDROME (PWS)?

PWS is a rare genetic disorder that occurs in approximately 1 in every 12,000 to 15,000 births. PWS affects both males and females, and all races worldwide. It is due to the lack of several genes on a small area of chromosome 15 (15q11-q13). At birth, PWS is characterized by low muscle tone, feeding difficulties and failure to thrive. During childhood, this may be replaced by a continuous drive to eat.

PWS is a spectrum disorder, with a wide range of abilities and challenges. There are common health problems in PWS such as growth hormone deficiency and muscle weakness, which can impair breathing and feeding skills, and slow physical development and learning.

Currently, there is no cure for PWS. In the past, medical care targeted specific symptoms, but now exciting research breakthroughs are occurring. These are providing a better understanding of the roots of the challenges; drug trials are underway. Until these treatments are available, we want to offer the best early start for your child.

Prader-Willi Syndrome Association (USA) offers support, education, advocacy, and funds for research to enhance the quality of life of those affected by Prader-Willi syndrome. We want to help you become an expert in your child’s rare disorder, and work toward optimal health.

HOW IS PWS DIAGNOSED?

Even though PWS is a genetic disorder, it is almost never “inherited” or “passed down” from the parents. It is a random genetic error. It is not caused by anything the parents did; it can occur in any family regardless of their diet, health, or social and educational advantages. In the process of creating a new life, genes and chromosomes must split and divide, and errors can occur.

PWS has three subtypes; the most common is called paternal deletion; the second is UPD, and the rarest is an inherited imprinting defect.

Healthy individuals rely upon their father’s chromosome 15 to be active, and the mother’s gene is “silent”. If the father’s gene is missing, this is called paternal deletion. It is not because the father has a genetic problem or did anything to cause this. It is totally random.

UPD is when the mother’s chromosome gets doubled – and then the father’s gene is eliminated by chance. Again, this is not the fault of the mother.

Your geneticist should explain which subtype your child has.

DNA Methylation Testing – The BEST INITIAL TEST

All persons suspected of having PWS should be tested with a DNA methylation analysis. This test detects essentially all (>99.9%) cases of PWS. If this test is POSITIVE, the FISH test will also be needed to detect which subtype.

FISH

Even if this test is negative, the individual could have PWS. It does not detect UPD or the rare imprinting defect.

For more information on the genetics of PWS, visit: http://www.pwsausa.org/genetics-of-pws/.
The priority for a newborn with PWS is weight gain and growth. Babies are sleepy and often have no hunger. They are too weak to suck. Smaller, weaker muscles are a part of PWS, and create hypotonia – low muscle tone. Most infants require some tube feeding, and have trouble sucking from breast or bottle. Many babies can take a part of each feeding by mouth, and the rest by ng (nasogastric) tube. Tube feeding helps them save energy. Rarely, a baby may need most of their feeding by tube, and a gastric tube (GT) may be recommended. As the baby gets older and stronger, with special nipples and therapies to help with feeding techniques, these tubes will no longer be needed.

A dietician is essential to create the perfect diet for each baby – sometimes adding supplements to breast milk, for example. Your baby will be fed on a schedule, and may need to be woken up to feed. Those calories are essential to good brain growth.

Growth hormone (GH) was approved by the FDA in 2000 to improve growth in children with PWS. Growth hormone deficiency is part of PWS, but early treatment provides children with much more than just better growth. GH improves muscle strength, which is the key to early learning; babies learn by movement and using their bodies. Research studies have clearly indicated that early treatment leads to the best outcomes in strength, development, and body composition. GH is the only drug approved for PWS, but it is a game changer.

Sleep and breathing problems can be detected in the NICU. Low muscle tone means that breathing can be weaker. Babies with PWS often have central (brain) sleep apnea, which is generally outgrown. This may require oxygen at home.
EARLY INTERVENTION SERVICES

These should be started as soon as you arrive home with your baby. This is the key for children with developmental disabilities and results in significant improvements in cognitive, academic and social outcomes. Therapy should begin shortly after birth, even as you are juggling feedings and life with a baby. Infants with PWS will achieve each milestone in the same order as other children, but at their own slower pace.

Babies are often undemanding, which means parents must try harder to entertain them, to stimulate learning and brain growth. Your therapists can help with a plan that is just right for your child.

Speech therapy
ST, SPT, or SLP - Sucking and feeding are skills that lead to speech, so this is a critical therapist right from the beginning. The SLP focuses on pre-speech and pre-language skills that must be acquired before an infant says his or her first words. While many children with PWS may have delays with speech, they have normal receptive skills for understanding speech. For more information, see the American Speech-Language-Hearing Association at www.asha.org.

Physical therapy
PT should start as soon as possible for improvement of mobility and strength. PT helps with development of motor milestones, improved mobility, and can help decrease the risk of scoliosis. PT should focus on core strength beginning in infancy. Tummy time and head control are often challenges, and PT will help. With appropriate PT, milestones can be achieved; staying consistent throughout their lifetime can help prevent obesity in the future. For more information, see the American Physical Therapy Association, Inc. at www.apta.org.

Occupational therapy
OT during early development is crucial. OT focuses on fine motor skills and sensory development. These skills include opening and closing things, picking up and releasing toys of various sizes and shapes, improvement of manipulating buttons, shapes and stacking. In some areas, the OT is a feeding specialist. For more information, see the American Occupational Therapy Association, Inc. at www.aota.org.
Once you have settled into a routine, you will rely on your medical team to address any health issues, and to look for the problems that are common in PWS. Your pediatrician may take the lead, or your endocrine or genetics team. Create a notebook with pockets to keep track of medical papers and reports; have a paper calendar to follow the many appointments in this first year. Write down your questions for each specialist.

Your baby may remain sleepy and need to be woken up to feed. Be patient. Be aware that people with PWS have trouble controlling their temperature, so avoid overdressing in the summer and underdressing in winter months.

Immunizations for children with PWS should follow the current AAP guidelines. Doses should be the same and the schedule should not be adjusted. Influenza vaccination is particularly important given the higher risk for respiratory problems. Children with PWS are not more likely to acquire a contagious illness, but with lower muscle tone and fewer signals that they are ill (high pain tolerance, temperature dysregulation) lung infections are higher risk.

Issues to discuss with your doctors in the first year include:

- When to start growth hormone
- When does the baby need a sleep study
- Respiratory Problems in PWS
- Recommendations for Evaluation of Breathing Abnormalities Associated with Sleep in PWS
- Is my baby gaining weight and growing correctly?
- Frequent diet adjustments with the help of a dietician
- Should my baby have a swallowing study?
- For boys – are the testicles positioned correctly?
- Is the penis unusually small?
- Are the hips correctly positioned? How is the back growing?
- Eye check – weak muscles can lead to a “lazy eye”.

See a pediatric ophthalmologist to evaluate any strabismus (eye-crossing). Strabismus is common in children with PWS, and is often addressed with patching, eyeglasses and/or minor muscle surgery.

https://www.pwsa.co.uk/assets/docs/vision_and_care_of_the_eyes.pdf
Pediatrician – Maintain regular contact with your pediatrician; this person is a valuable advocate for your child. These regular physical exams and assessments are important, even as you see multiple specialists. PWS-specific growth charts are available, free of charge, on our website (www.pwsausa.org) or by request.

Pediatric dentist – preferably with experience in sensory issues and special needs. Children with PWS can have thick, sticky saliva, and thin tooth enamel, which makes them more prone to early wear and cavities. Good oral hygiene and regular exams by an experienced pediatric dentist can help ensure your child will have strong, healthy teeth for his or her lifetime.

Pediatric orthopedist for an evaluation of scoliosis and hip health. Over 40% of children with PWS develop spinal curves. They often show fewer outward signs of a curve than typical children so a back Xray is essential.

Sleep specialist – Sleep studies (polysomnography) diagnose and monitor all types of breathing problems common in PWS.

ENT – if recommended by your physician. GH can make tonsils grow; weaker muscles in the neck can press on the windpipe during sleep; and some children with PWS have smaller mouths – all can cause obstructive sleep apnea, and might make a tonsillectomy and/or adenoidectomy necessary.

Dietician – The prevention of obesity is one of the most important goals and it can be achieved, but not without a careful plan.
Hippotherapy
The term “hippotherapy” comes from the Greek word hippos, (horse), and simply means therapy with the assistance of a horse. Hippotherapy is performed by a specially trained physical or occupational therapist working with an experienced horse handler, and can be started as young as two years of age. The action of the horse stimulates muscle tone, mobilizes joints, increases sensorineural integration, assists with the development of balance and equilibrium, and may improve speech production and cognition. For more information, ask your Early Intervention Therapist, or read more at this link: www.americanhippotherapyassociation.org.

Early Behavior Management
Many children with Prader-Willi syndrome thrive in predictable routines. Knowing what to expect can often head off behavior issues and relieve anxiety. It is important from the very beginning to reinforce the positive behavior you want to see. Children and toddlers with PWS can be “hyper-reactive”, so a calm environment, and calm reactions and responses bring out the best in them. These children want to please us, so disapproval may be very painful to them. Ignore small mistakes and praise small steps, like being patient, or smiling when waiting.

Transitions and changes may be hard, so parents may need to change their own style, becoming less spontaneous. Children may process words more slowly, so give your child time to think and don’t expect a fast response.

Techniques such as ABA therapy, (http://www.appliedbehavioralstrategies.com/what-is-aba.html) or other behavioral therapies are helpful as your child grows. Consult your pediatrician or Early Intervention program for more information.

Cognitive Assessment
As little ones with PWS grow from toddler to young child, parents begin to wonder where they fall on the cognition/intelligence spectrum. It is commonly expected that children with PWS will have some degree of cognitive or intellectual deficit, but learning ability varies widely between individuals, and can depend on many factors. Cognitive development screenings are often part of developmental assessments by pediatricians and therapists, and if there are indications of delay, more formal evaluations can be scheduled. Remember, IQ scores and other cognitive profile results do not define your child – they are simply a tool to help identify strengths and weaknesses, and help decide whether special help may be needed.
Nutrition and obesity prevention

Children with PWS are genetically predisposed to becoming overfat – and overweight. After the first year of life, they may need only 60% of typical caloric intake to maintain good growth and normal weight gain. Metabolism is slower, even with high levels of activity.

Consistent exercise and activity should become part of your family routine. Not only does it benefit the child with PWS, but keeps the entire family healthier. Stronger, bigger muscles help burn calories in all people.

Meals should be predictable, served as a family meal, and always served at a table. Portions should be provided by the parent, not self-served. No snacks should be given “on demand.” Controlling the food environment from the beginning is the best way to ensure good behavior and good weight control. Parents must be very strong and not give in to any plea for “more” – and educate others to ignore any requests for food. Behavior control is the key to obesity control.

PWS Nutrition Phases

Children with PWS may follow this common pattern. The most recent generations of children with PWS may not have as much hunger or food seeking as was seen in the past.

**Phase 1a:**
*Patients have no desire to eat as infants, often with failure to thrive infancy*

**Phase 1b:**
*Period of normal appetite and normal weight gain (until approximately age 18 months – 36 months)*

**Phase 2a:**
*Weight begins to increase without a change in appetite or calories (age 18-36 months and lasts until approximately age 5)*

**Phase 2b:**
*Increasing interest in food (ages 5-8 years)*

**Phase 3:**
*Insatiable appetite (average age on onset 8 years)*

**Phase 4:**
*Appetite is no longer insatiable and is able to feel full (may occur in adulthood)*
RESOURCES/SUPPORT SERVICES

PWSA (USA) is the only national organization dedicated to improving the lives of all persons with Prader-Willi syndrome. We provide support in every stage of life through research, education, support, and advocacy. Hospitals, physicians, organizations, and parents from all over the world consult with PWSA (USA) regarding medical problems and other PWS-related issues daily.

- **Parent Mentoring** – Parent to Parent support with a trained parent mentor. Through PWSA (USA) we have extensive age-appropriate materials that we provide to every family, and for physicians which includes up-to-date materials paramount to treatment for PWS.

- **Medical and Crisis** – PWSA (USA) Medical, Family and Crisis Counselors on staff for support with medical, behavioral, and educational needs. Information and referral services, and advocacy to assist with benefits and services including SSI and essential therapies. Special Education Advocacy on behalf of the student with PWS including information about IEPs, and PWS-specific school strategies for educators.

- **PWSA (USA) State Chapters** – PWSA (USA) Chapters offer local advocacy, resources and family connections, and coordinates partnership with national support services.

- **National Convention** – Held every two years, often a life-changing event for parents and their children.

- **MEDICAL ALERT BOOKLET** – download link HERE

“As parents when you hear the heartbreaking news that your child has been diagnosed with Prader-Willi syndrome, you are flooded with a range of emotions. At first we felt alone and didn’t know what to expect. While sitting in the NICU, I reached out to PWSA and was immediately contacted by a new parent support coordinator, who took time to talk with me many, many times to answer any questions I had and put me in touch with other parents going through the same things. But most importantly, she was there to let me vent and talk to someone who knew exactly what I was going through. The PWS community is a close-knit family. Most of us have or will go through the same challenging obstacles. Having support groups and family support advocates are a great addition to help give us and our loved ones the help they deserve.”

~ Blythe

Averie Wolksendorfer

The Gregg Family
GETTING INVOLVED IN PWSA (USA), AND HELPING THE CAUSE...

"What you do makes a difference, and you get to decide what kind of difference you want to make."
- Jane Goodhall

Key Roles You Can Play
We all benefit from the involvement of others, whether you think of it as "many hands lighten the load," or "there is strength in numbers." Being a community volunteer working with others from PWSA (USA) has additional benefits, ranging from gaining knowledge that can help you, to making fast and lasting friendships.

Many have learned that it is a "win-win" partnership when you decide to:

• Be a Volunteer
• Support a Fund-Raising Event
• Attend Chapter, Regional, and National Meetings
• Host a Fun Fund-Raising and Friend-Raising Event

Certainly, an increase in awareness and advocacy on behalf of those with Prader-Willi syndrome is a top priority.

Increasing the dollars available to fund the family support, research, and related activities of PWSA (USA) goes hand in hand with expanding the ranks of supporters.

You Can Make A Positive Impact By:

• Supporting the eWalk Virtual Fundraiser
eWalk is Prader-Willi Syndrome Association (USA)’s online virtual walk that allows anyone, from anywhere, to participate and help raise vital funds. Remember, PWSA (USA) is the only organization that provides new diagnosis support, research, family/crisis support, medical support, advocacy, and education.

eWalk is a simple way to raise money for Prader-Willi syndrome. All you have to do is register, personalize your page, send it out, and watch donations come in. The Internet magnifies our ability to spread awareness about PWS, and you can personalize your page with a picture and story. Once you send out the Web Page link through social media and e-mail, all recipients have to do is click on your link, go to your page where they see your story and picture, and they can click on the donate button. And, there are even prizes awarded to you as you raise various amounts of critically needed donations!

For more information, please click:
https://www.pwsausa.org/ewalk.

Raising funds for PWSA (USA) is soothing for my soul. As a mom of a 5 year old daughter with PWS (and 3 other children), life is crazy. Fund raising or any event I attend or participate in allows me to focus on my PWS journey and lets my family do the same. Meeting other parents, siblings and individuals with PWS makes my journey feel so much less lonely and gives me hope and inspiration. PWSA (USA) has so many resources for us and I find comfort in knowing they are there for that emergency that might happen.
~ Ruth Livak
• Organize an “On The Move” Event
Both creative and flexible, “On The Move” is a campaign designed to increase awareness of PWS and to raise funds for PWSA (USA) and state chapters. It strengthens support for the PWS community as a whole and PWSA’s numerous family support services.

On The Move events often include an element of physical activity ranging from walks to bowling to golf outings, but also can be dinners, auctions, and more. Anyone can organize – with key information and assistance from the PWSA national office – and participate in an “OTM” event, and they can take place throughout the year.

Here are the top ten reasons to conduct an On The Move Event:

1. Raise critical funds for vital research, family support, awareness, and education.
2. Win great prizes!
3. Includes elements of physical activity and community camaraderie.
4. Great way to get friends, family, neighbors, and co-workers together for a fun event.
5. Can be held by individuals, state chapters, or local groups.
6. Terrific way to spread awareness and support advocacy goals.
7. PWSA (USA) development team is ready to help you move through the steps.
8. We can set up a personalized Web Page for you and your participants; donations can be collected online.
9. All of the funds raised help those affected by Prader-Willi syndrome.
10. Our event kit and staff support provide you the direction to be efficient and successful.

For more information, please click http://www.pwsausa.org/on-the-move/

“Today, give yourself permission to be outrageously kind, irrationally warm, and improbably generous. I promise it will be a blast.”
- Sasha Dichter
• **Attend the PWSA (USA) Biennial Convention/Conference**

This one-of-a-kind gathering is where research, support services, and families come together with the common goal of improving the lives of individuals with Prader-Willi syndrome and their families. This amazing event:

• Offers programs tailored for parents, people of all ages with Prader-Willi syndrome, medical and professional providers, many of whom will travel from all over the world to attend;

• Provides high quality professional care for persons with PWS so that family members can focus on receiving the latest information;

• Allows the experience of networking with others who understand topics related to PWS, which is priceless for our participants.

For details about upcoming PWSA (USA) national conventions, please click: [http://www.pwsausa.org](http://www.pwsausa.org)

“**A hero is someone who has given his or her life to something bigger than oneself.**”
- Joseph Campbell

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**Your Involvement Accelerates All Our Progress**

Together we can and will address the challenges related to Prader-Willi syndrome and improve the quality of life for all of those impacted by it.

To get involved, please call 1-800-926-4797 and ask for the Development Department.

Jaelle LaPlante with Declynn, Katie Bey with Ania, Amanda Ordonez with Alejandro, Katie Moureau with Cade
As always, **THANK YOU** very much for your involvement and support!