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FOUNDATION FOR
PRADER-WILLI
RESEARCH
CANADA

Working Toward an Independent Future

FIRST STEPS

A package for parents/caregivers of newly diagnosed children with Prader-Willi Syndrome

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A Letter of Hope

We are so glad you found us!

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 Prader-Willi Syndrome.

FIRST STEPS was written by parents for parents. It 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 the Prader-Willi journey with our own families.

Our family has come a long way since receiving our daughter's diagnosis. Years ago, while in the NICU, all we saw when we looked at our daughter was a lifeless little girl. We were two scared parents that 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 most, Diagnosis Day is unforgettable in our minds. The heartbreaking day an imagined future for our new baby or child was forever changed. An overwhelming day when we 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 can to provide the best possible care and the brightest future for our child. 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?"

I wish we would have spent less time looking at our daughter as a diagnosis and spent more time just cuddling and enjoying her for who she is. I wish we could've been able to see a clip of what life was going to be like now, back then. We never could have dreamed Giulianna could change our lives the way she has. She has brought more joy and love to our family than we ever imagined possible!

While the list of potential problems associated with Prader-Willi Syndrome is long and sometimes devastating to consider, we 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:
- an incredibly supportive parent network
- an ever-expanding group of medical and research professionals who are working diligently to provide optimal care and treatments for our children
- 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 threatens to overwhelm most 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 help connect you to these resources, alleviate some of your fears about your child's future, and empower you as you coordinate care for your child.

- *Carole Elkhal, Mother to Giulianna;*
Community Director, FPWR Canada
contact: carole.elkhal@fpwr.ca

We're here for you! If you haven't already, please contact us at info@fpwr.ca

What Is Prader-Willi Syndrome?

Prader-Willi syndrome (pws) is a genetic disorder that occurs in one out of every 15,000-25,000 births. PWS affects males and females with equal frequency and affects all races and ethnicities. The symptoms associated with PWS are caused by a lack of active genetic material in a particular region of chromosome 15, but it remains unclear how inactivation of this region leads to the PWS characteristics.

Early on, PWS is characterized by low muscle tone (hypotonia) in infants, with difficulty feeding and the risk of *failure to thrive*. Later, this is replaced by an unregulated appetite and a strong drive to eat. Individuals with PWS lack normal hunger and satiety cues. They usually are not able to control their food intake and will overeat if not closely monitored. Food seeking behaviours are very common. In addition, the metabolic rate of persons with PWS is lower than normal. Without appropriate dietary intervention and constant vigilance, the combination of these problems will lead to early onset childhood obesity and its many complications.

In addition to obesity, a variety of other symptoms are often associated with PWS, including growth hormone deficiency, abnormal body composition, speech impairment, scoliosis, sleep disturbances and learning disorders.

Behavioural difficulties may include symptoms of obsessive-compulsive disorder and difficulty controlling emotions. Behaviour and mental health issues can represent some of the most challenging aspects of caring for an individual with PWS.

Prader-Willi Syndrome is a spectrum disorder and symptoms vary in severity and occurrence among individuals. With the benefit of an early diagnosis, access to growth hormone replacement therapy, and a nurturing environment, those with PWS are accomplishing more than ever! Nevertheless, even as many with PWS have the intellectual capacity to live independently, the challenges of the disorder are limiting, and the vast majority of individuals with PWS are not able to live without constant supervision.

Currently there is no cure for Prader-Willi Syndrome, but our goal is to change that! For many individuals and their families who are affected by the disorder, the elimination of some of the most difficult aspects of the syndrome, such as curbing the insatiable appetite, has the potential to dramatically improve the quality of life and open up an abundance of new opportunities. Through advancement of FPWR Canada's mission, we intend to do just that.

FAQ About PWS

HOW COMMON IS PWS?

PWS occurs in approximately one out of every 15,000 - 25,000 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.

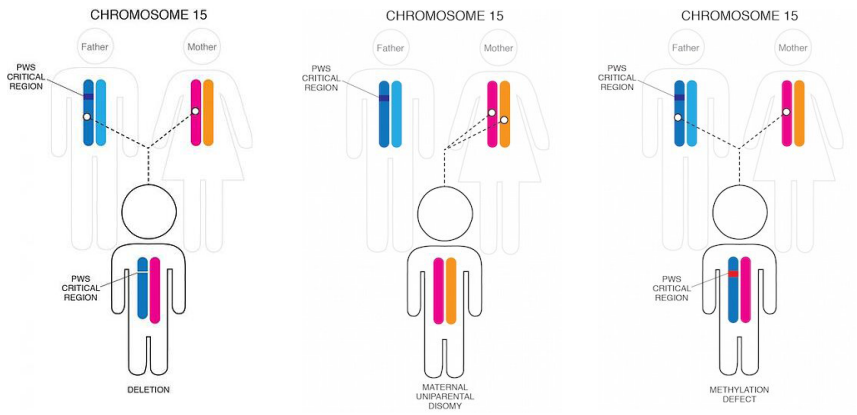
WHAT CAUSES PWS?

PWS occurs when information from one of an individuals two chromosome 15s – the one normally contributed by the father, is missing. This can happen in three ways:

1) Deletion. Most often, part of the chromosome 15 that was inherited from the persons 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) Imprinting Mutation. 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.



Enjoy your precious babies while they are young, and connect with positive families who are a few years ahead of you in their journey.

In the beginning, I spent way too much time thinking about Dante’s future when current research may eliminate some of our biggest fears.

And just like any good parent would do, lead by example and set your expectations high—the sky is the limit :)

- Tanya Johnson
Mississauga, ON
Mother of Dante; Age 17

FAQ About PWS

HOW IS PWS DIAGNOSED?

A suspected diagnosis of Prader-Willi Syndrome (PWS) is usually made by a physician based on clinical symptoms. PWS should be suspected in any infant born with significant hypotonia (muscle weakness or “floppiness”). The diagnosis is confirmed by a blood test. The preferred method of testing is a “methylation analysis,” which detects >99% of cases, including all of the major genetic subtypes of PWS (deletion, uniparental disomy, or imprinting mutation). A “FISH” (fluorescent in-situ hybridization) test will identify those patients with PWS due to a deletion, but it will not identify those who have Prader-Willi Syndrome by “UPD” (uniparental disomy) or an imprinting error.

Almost all cases of PWS can be confirmed by one of the above tests. However, in the rare event that laboratory tests do not confirm PWS, a clinical diagnosis can be helpful for the development of a management plan.

ARE THERE DIFFERENCES IN THE SEVERITY OF PWS BASED ON THE GENETIC SUBTYPE?

There may be some subtle differences in the characteristics of PWS based on genetic subtype: for example, those with deletions may be fair-skinned with light hair compared to other family members and may be more susceptible to seizures; those with PWS by UPD may be at higher risk for mental illness in young adulthood. Overall, however, there is considerable overlap between the different genetic subtypes. It is likely that the thousands of genes outside the PWS region, which exhibit normal variation between individuals, also contributes significantly to the variability in PWS symptoms between those with the disorder.

WHEN DOES THE OVEREATING ASSOCIATED WITH PWS BEGIN?

The symptoms of PWS change over time in individuals with PWS, and a detailed understanding of the nutritional stages of PWS can be found **HERE**. Typically, an increased interest in food begins between 2 and 8 years of age, with hyperphagia typically beginning sometime between the ages of 8 and adulthood.

It’s a whole new world but one with a powerful PWS community behind you that will help every step of the way.

Your child will give you unconditional love, teach you to be patient and appreciate the little things in life. It’s an unfamiliar journey but one that you can make into an adventure. Life is about moments we create and remember - make memories to live your best life.

- Michelle Cordeiro,
Richmond Hill, ON
Mother of Julia; Age 15

FAQ About PWS

IS PRADER-WILLI SYNDROME CURABLE?

Currently, there is no cure for Prader-Willi Syndrome, and most research to date has been targeted towards treating specific symptoms. For many individuals affected by the disorder, the elimination of some of the most difficult aspects of the syndrome, such as the insatiable appetite and obesity, would represent a significant improvement in quality of life and the ability to live independently. The Foundation for Prader-Willi Research Canada is interested in advancing research toward understanding and treating specific aspects of the syndrome, with the goal of an eventual cure 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 (read more about HGH on page 13).

Additional studies have found positive results on development, behavior, and intellectual capability. For help accessing growth hormone in Canada, consult with FPWR Canada by emailing *info@fpwr.ca*.

WHAT DOES THE FUTURE HOLD FOR PEOPLE WITH PWS?

People with PWS can expect to accomplish many of the things their “typical” peers do such as 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.

The whirlwind of diagnosis can seem overwhelming and scary - appointments, adjusting expectations, learning about PWS, but with the help of those who have been there before you, you’ll realize you are not alone in this “new normal”.

No one will better understand your feelings and fears than another PWS parent, and they are an invaluable resourse. Reach out and make connections - this diagnosis will become a lot less scary when you realize you have a very awesome and supportive community behind you who will guide you through the quirks of PWS and show you just how amazing your kiddo is.

- Jennifer Joseph,
The Blue Mountains, ON
Mother of Darwin; Age 7

Real Talk with Real Parents

The things you really want to know.

Belinda & Jack Jones
with Brook
Brampton, ON

Simina & Alex Ardeleanu
with Maya
Pickering, ON

Silvia Rinaldi & Melissa
Mississauga, ON

WHAT WILL MY CHILD LOOK LIKE?

My child looks like every other child who is 9 and is happy and healthy. She has a ton of interests and many hobbies! We've taught her to be strong and brave; to give everything a try no matter how hard it looks. This has given her confidence to embrace her differences while also loving herself for her kindness and compassion, and growing confidence in leadership and individuality.

- Belinda & Jack Jones – Brampton, ON
Parents to Brook, 9

WILL OUR FAMILY BE ABLE TO TRAVEL?

YES! You model and instill experiences that your family and you, as parents enjoy doing - from camping, to museums, to road trips (though road trips with any toddler or child is a literal hassle), to excursions! And the same goes for every child you are welcoming into your life - you are trying to find your new normal and balance in the new family. Maya loves meeting new people, and camping, and exploring, and we get her involved in everything we do as a family. Yes we had to be creative in regards to food and start our own rituals, but now even our friends are on board and their kids are beyond kind and understanding to Maya's specific diet restrictions!

- Simina & Alex Ardeleanu – Pickering, ON
Parents to Maya, 6

YES, you most certainly can travel with your child. I was so convinced after receiving the diagnosis that we would never be able to travel again. But here we are 17 years later, and my daughter has been on a plane +20 times, and we've done numerous long road trips. Family time away is such an important thing, even more when you are raising a child with PWS. Planning and preparation will be key to having a successful trip. Make sure you have lots of favourite toys, security items, blankets and activities to keep your child entertained during travel time. Check ahead to see if your dining destinations can accommodate any special requests you have but always bring a secret stash of your own "snacks" just in case of travel delays. As your child gets older and you start to recognize what may trigger their anxiousness, you can plan your trip to handle these situations (e.g. asking the flight attendant if his/her meal can be served first, pre-seating on the plane, avoiding large tour groups, keeping meal times and bed times consistent). Do not let the fear of travelling with your child hold you back — with planning and preparation you can make some wonderful memories with your family on vacation!

- Silvia Rinaldi – Mississauga, ON
Mother to Melissa, 17

Real Talk with Real Parents

The things you really want to know.

Genevieve Currie & Family with Callum
Calgary, AB

Mandy Young & Sophie
North Vancouver, BC

Brooke Gibson & Daniel
Chestermere, AB

WHAT WILL MY CHILD BE SMART?

Yes your child will be very smart in ways you may not have imagined. Your child may remember conversations and people from years past. Your child may be very good at remembering people's names and where they met them. You child may know every dog and dog owner's name in the neighborhood so you won't fumble when you run into them trying to remember who they are. Your child will hold other's attention with their curiosity about how things are done and why they are done in a certain way. Your child will be unique and very smart in their own way.

- Genevieve Currie – Calgary, AB
Mother to Callum, 13

WILL OUR FAMILY BE ABLE TO GO OUT FOR DINNER?

As a family of foodies receiving the PWS diagnosis, that was the first question I thought. For us as a family to be happy we needed eating out to be a reality. We choose restaurants wisely and go back to our favorites so they get to know us and our unique requests. We generally stick to family run establishments and away from chain restaurants. With the right food security and certainty for our daughter, eating out is not an issue at all for us.

- Mandy Young – North Vancouver, BC
Mother to Sophie, 9

Absolutely. Going out for dinner is still such an enjoyable family experience for us because we set Daniel up for success. We always set expectations going in for what he can and cannot have and the type of behavior we expect. He knows that, depending on what he orders, he may not get to eat the entire meal at the restaurant so some might go home for leftovers. We have made it exciting because he gets 2 meals out of one. Wherever possible we also look at the menu in advance so he has an idea going in of what his options are. Family dinners out are still possible and a wonderful family experience, it's just a matter of having expectations and setting our kiddos up to succeed in advance.

- Brooke Gibson – Chestermere, AB
Mother to Daniel, 9

We just received a diagnosis of Prader-Willi Syndrome.
WHAT SHOULD WE DO NEXT?

FIRST STEPS

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, and you're faced with an infinitesimal 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. CONSIDER STARTING YOUR CHILD ON GROWTH HORMONE THERAPY

There is evidence that there are significant benefits to starting 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

These therapies 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 others' 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.

OTHER SPECIALISTS

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

Early Interventions

Early interventions should begin as soon as a diagnosis has been made. Early intervention in children under age three years, particularly physical therapy, may improve muscle strength and encourage achievement of developmental milestones. Physical, Occupational and Speech therapies are recommended for PWS infants, and research indicates, early diagnosis and therapies may reduce the duration of tube feeding and prevent early obesity in PWS Infants.

PHYSICAL THERAPY

Physical and occupational therapy are 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 receiving the diagnosis later in childhood, a speech and language assessment should be made as soon as the diagnosis is made.

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.

The dietician 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 as 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.

continued on next page...

Early Interventions

HIPPOTHERAPY

Hippotherapy is commonly recommended for kids 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 in the patient that are similar to and essential for walking. In addition, adjusting to and accommodating for the horse's movements increases sensorimotor integration.

You can search for a hippotherapy center near you. Please ensure it is not therapeutic horse ridding which does not provide the same therapeutic benefits.

AQUA THERAPY

Although there is little data supporting the implementation of aqua therapy for those with PWS, it is also 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 (developmental baby 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.

Growth Hormone Therapy

In multiple studies, human growth hormone (HGH) has been found to be beneficial for those 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 behaviour.

There is evidence that GH treatment improves cognitive performance. Children who have been on growth hormone prior to four 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. You can read more about the GH guidelines **HERE**.

Despite HGH treatment, many difficult symptoms associated with PWS remain hard to treat. To date, no effective medications have been found to regulate appetite. Inability to control food intake is often the biggest obstacle keeping those with PWS from living independently. In addition, medical treatment of the psychiatric and behavioural issues associated with PWS has produced inconsistent results.

FPWR CANADA'S CONFERENCE PRESENTATIONS
Dr. Jennifer Miller, M.D.
Pediatric Endocrinology, University of Florida

Video Links:

- **HORMONE DOSING**
- **COGNITIVE BENEFITS**
- **BENEFITS FOR ADULTS**

Growth Hormone Access

Growth Hormone (GH) Access has changed tremendously over the last year. The Foundation for Prader-Willi Research Canada (FPWR Canada) successfully mounted a physician led submission to get provincial funding of GH for children (17 years and under) with a PWS genetic diagnosis.

In June of 2020, Health Canada approved GENOTROPIN for the treatment of paediatric patients who have growth failure due to PWS. As a result, GENOTROPIN is the only GH indicated for PWS at this time.

For more information on possible avenues to GH access in Canada, please click **HERE**.

If you have additional questions, please contact our FPWR Canada GH Advocate celine.lepage@fpwr.ca

Research | Awareness | Fundraising

At some point in your Prader-Willi journey you may want to get more involved in advancing PWS research, awareness and fundraising. We've all benefitted from the mothers, fathers, doctors, researchers, and donors who have built the strong foundation of research, care, and support that has enveloped our families upon diagnosis.

It's our turn to do the same! Not only for our generation of children with PWS, but for all the families who will receive diagnoses after ours.

Click HERE to read Stories of Hope from PWS families.

PWS RESEARCH

There is much knowledge yet to be gained through PWS research - knowledge that will lead to significant medical breakthroughs and new therapeutic options for our children, and will dramatically change the outlook for those diagnosed today. Our logo's tagline is "Working Toward an Independent Future"... and with a continued focus on research, we will be able to see the day when our children will be able to live independent and happy lives, free of the burden of Prader-Willi syndrome.

Visit www.fpwr.ca to learn more about current research.

FPWR CANADA'S ANNUAL CONFERENCE

We host an annual conference where we discuss the latest trends in PWS research and learn how to become effective advocates for our loved ones. Check out the latest conference information **HERE**.

FUNDRAISING FOR RESEARCH

By supporting PWS research, you will help us find effective treatments and, eventually, a cure for PWS. Our children deserve nothing less!

GET INVOLVED WITH FPWR CANADA

There are numerous ways you can help to advance the cause of FPWR Canada!

- Join a committee - work with passionate families and volunteers to make a difference in the lives of people with PWS;
- Share your skills, talents or interests - people are at the heart of our work and we welcome you to share your professional skills with us (e.g. accounting, photography, legal, web design etc.);
- Volunteer at our events - make a difference in your community and lead by example.

Learn how to get involved with FPWR Canada **HERE!**

IT STARTS WITH ONE SMALL STEP

ONE Small STEP for Prader-Willi Syndrome

When you fundraise through ONE SMALL STEP, you directly impact vital research, collaboration, and life-giving clinical care programs for people around the globe suffering from the symptoms of Prader-Willi Syndrome.

One SMALL Step, hosted by the Foundation for Prader-Willi Research Canada, is an international event supported by Prader-Willi organizations and families around the world. Since it's inception, One SMALL Step has raised funds to achieve the mission of FPWR Canada: *"to eliminate the challenges of PWS through the advancement of research."*

CLICK HERE TO GET INVOLVED!

Resources

CLICK ON ANY OF THE FOLLOWING RESOURCES TO LEARN MORE.

Foundation for Prader-Willi Research

www.fpwr.ca

www.fpwr.org

OneSMALLStep

Provincial Organizations

APWQ - Association Prader-Willi Quebec

OPWSA - Ontario Prader-Willi Association

BCPWA - British Columbia Prader-Willi Association

PWSAA - Prader-Willi Syndrome Association of Alberta

PWS Organizations around the World

PWSA-USA - PWS Support Organization

IPWSO - International PWS Organization

Prader-Willi France - Organisation de support pour le SPW de la France

FPWR UK - Foundation for Prader-Willi Research UK

Prader-Willi Research Australia

Facebook

Many of our families are on Facebook!
Join our sponsored pages, make new friends!

- FPWR Canada
- FPWR USA
- OneSMALLStep
- Échange entre familles québécoises
- Canadian PWS Families Group
- PWS lovebugs

PWS Research

Learn all about the many developments taking place in PWS research!

- Research Publications
- Recorded Webinars
- PWS Therapeutics in Development
- Clinical Trials
- Current Projects funded by FPWR Canada

Photos and Video of People with PWS

- Photo Collection
- Video Collection

www.fpwr.ca

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