Parents Guide

PRADER WILLI SYNDROME

0 to 12 months

pwsai
PRADER WILLI SYNDROME
ASSOCIATION IRELAND

Congratulations on the birth of your beautiful baby

This booklet has been designed to give you some basic information about Prader Willi Syndrome (PWS) and to offer you practical advice to support you and your baby.

It focuses on birth to 12 months. Having a newborn baby can be challenging, having a newborn baby with PWS can bring additional challenges.

The information will inform you about the diagnosis from a parent's perspective to allow you to focus being a parent and enjoying the newborn period.

It is important to note that PWS is a spectrum disorder.

A child can have some or all of the symptoms, each in varying degrees.

Most children will not have every symptom.

What is Prader Willi Syndrome Association of Ireland (PWSAI)?

PWSAI is a charity that was founded by parents of children with PWS to provide support and advocate for people with PWS, their families and carers. As parents, we appreciate what you are going through, we have been there ourselves. PWSAI and its members are available any time you feel ready. You can connect with us via the new diagnosis section of our website. See details on the back.

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What is Prader Willi Syndrome or PWS?
PWS is a rare and complex chromosomal disorder that a child is born with and has for life. It occurs in approximately 1:15,000 births, roughly 5-6 births annually in Ireland. PWS occurs randomly, there is nothing a pregnant woman does, or does not do, to cause PWS.

There are three causes for PWS, these are called sub types. The sub type results from the way in which chromosome 15 developed in utero. It may be important in later life to know which sub type of PWS your baby has. So, it is good to get a copy of the genetic report (Methylation Testing) that has been completed at diagnosis The genetic consultant can help with this.

PWS Sub Types:
Chromosomal Deletion, approx. 70% of cases.
Uniparental Disomy (UPD), approx. 30% of cases.
Methylation Defect (Imprinting), approx. 1-3% of cases

Typically, PWS is a neurological condition that causes hypothalamic dysfunction. The hypothalamus links the endocrine and nervous systems via the pituitary gland. It is responsible for producing many of the body's hormones and controls endocrine functions such as metabolism and growth. PWS is medically complicated and affects many governing functions such as temperature regulation, thirst, hunger, the sleep/wake cycle, mood, cognition and learning, the expression of emotions and the feeling of pain.

Medical referrals required from 0-12 months

The treatment of PWS has changed over the past few decades. Although there is no cure for PWS, huge progress has been made both medically and within Early Intervention Services, that managing this Syndrome has changed dramatically.

The future is bright for our kids.

The treatment of PWS requires a multi-disciplinary approach, meaning there will be many different medical disciplines involved in the treatment of your child. This booklet will detail what is considered best practice from birth to 12 months, breaking it down to manageable steps.

1

Your child will need a referral to an Endocrinologist

Most people with PWS have insufficient Growth Hormone. Human Growth Hormone (GH) has been administered as a nightly injection to children with PWS since the 1990s. There is evidence that starting GH before 1 year of age has significant benefits.

An Endocrinologist specialises in conditions relating to Hormones. To get GH prescribed, your child will need to see an Endocrinologist, one with experience in PWS is best. Currently most PWS patients in Ireland see a Paediatric Endocrinologist at Children's Hospital Ireland (CHI) at Tallaght.

2

Your child will need a referral to a Respiratory Consultant.

Best practice states that a Sleep Study (called a Polysomnography) should be carried out prior to starting GH and again three months after.

At present the only Children's Hospital in Ireland that performs these detailed sleep studies on infants is CHI Crumlin. This sleep study is preformed overnight while your child sleeps. Most of the time the sleep study can be preformed at home.

During the sleep study your child's breathing and sleep pattern will be recorded and analysed. A respiratory consultant will send a report of the study to your Endocrinologist. If the sleep study is abnormal your child may need to be reviewed by a respiratory consultant. If no concerns are raised, then your Endocrinologist can proceed to prescribing GH. Your local Paediatric Endocrinologist can refer to respiratory and commence GH.

3

Your child will need a referral to Early Intervention

Early Intervention – Disability Network Ireland

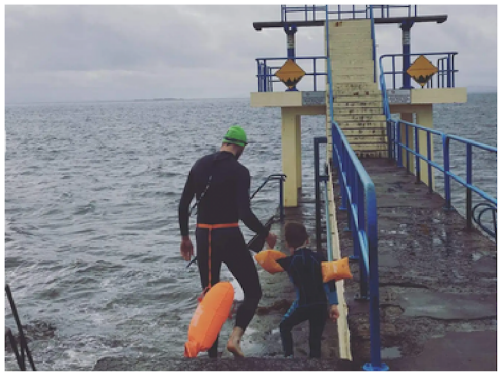
Early Intervention (EI) Services provide so much therapeutic support for us and our children that they have become a key player in this multi-disciplinary approach. EI therapies may include Physical Therapy (PT), Occupational Therapy (OT), Speech and Language Therapy (SLT, speech therapist also provide feeding therapy), Dietician.

Build a Network

Connecting with others who understand what you are going through can be immensely valuable. Those who are a little further into the journey can provide support and advice.

Making that first step to connect can be daunting, hopefully in time you will feel ready. PWSAI can connect you with another parent, someone a little further into this journey, to help get you started on building a network. We can also help you to connect with any of the many private social media groups for parents, carers or grandparents that are out there.

The early months are a treasure that go by so fast, amidst all the fear you may be feeling, please try to remember to stop and cuddle your baby.



When you are ready... we are here

It might not seem like it now, but your child will be able to achieve so much more than you currently imagine. Milestones will be achieved, each in their own time.

Watch their eyes beam up with pride as you cheer them on every achievement. Your child will bring so much love, joy and happiness to your family.

They will grow and shape you as a parent like never before. Take this journey day by day, and most importantly, enjoy your baby.

We are part of your support network.
If we can help, we will.
If we can't, we will listen and try and understand.
We are here.
PWSAI

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