PWSA UK
PRADER-WILLI SYNDROME ASSOCIATION UK

About PWS – the basic facts

Introduction

Prader-Willi Syndrome (PWS) was first described in 1956 by Swiss doctors, Prof. A Prader, Dr A Labhart and Dr H Willi, It can feel overwhelming to learn about this condition, and the information below can seem daunting. It's really important to remember that every child is an individual and Prader-Willi Syndrome is a condition your son or daughter has, and not who they are. PWS is a spectrum, which means that your son or daughter may not show every characteristic.

PWS is a rare, genetic disorder caused by a defect of chromosome 15. Features can include:

- Hyperphagia –excessive and overwhelming appetite
- Hypotonia—low muscle tone
- Hypogonadism—immature sexual development
- Learning disabilities
- Immature emotional and social development
- Speech and language difficulties
- Mobility issues

Behavioural phenotype

As well as the above features, there are a set of behaviours associated with PWS which can include:

- Obsessive and/or compulsive behaviour
- Perseveration—repeating the same question continually or get
- Difficulty in adapting to change of any sort
- Stubborn or oppositional behaviour
- Emotional outbursts and dysregulation

Cause

Most cases (65-70%) are caused by a deletion on the chromosome 15 inherited from the father (known as deletion), and 25-30% are caused by inheriting two chromosome 15s from the mother (known as uniparental or maternal disomy).

Less than 1% of people have a translocation or imprinting irregularity on chromosome 15 – your geneticist can tell you more if your son or daughter has this.

PWS happens purely by accident – it's not a result of anything you or your partner have or haven't done. As of 2023, we believe that the birth rate is between 1 in 10,000-30,000, meaning you are part of a very rare, unique and welcoming community. The chances of having further children with PWS is the same as any other member of the public.

Most infants are now diagnosed at or near birth, although there can still be a delay in diagnosis for some families. Diagnosis is usually through a methylation blood test—this will identify deletion, maternal disomy and translocation. We estimate that there are around 2000 people with PWS in the UK of all ages, and you do not have to do any of this alone.

Below, we take a look at how PWS may affect your child throughout their life – remember, not every child is the same and some may show more characteristics than others.

Babies

Your baby is exactly who they were before diagnosis – they are still yours, still adorable, still amazing. Enjoy getting to know them. Babies with PWS are very floppy when they're born (hypotonia) and usually have a really weak suck and/or are not able to swallow. It's typical for babies to be tube fed for the first few weeks or months, and although this can be daunting, it's just to make sure they are getting the right amount of nutrition. It's not unusual for babies to show no interest in feeding in the first few months, and you'll find that they sleep most of the time and have a very weak cry. For sons, you might notice they have undescended testes and small genitalia. It can be the same for daughters in terms of genitalia but this can be harder to notice. All of this is completely normal.

A quick note about twins – if they are identical, both will have PWS. If they are non-identical, usually only one will have PWS.

Childhood and hyperphagia

Most children with PWS begin to show more interest in food between the ages of 1 and 4, although some might not show this until much later. Remember that it's normal for nursery age children to become more interested in food and use it in play, regardless of whether or not they have PWS. At some point, all children with PWS will develop an overwhelming drive to eat and an insatiable appetite. This can sound really scary, and the degree to which this happens can vary, but there is always a preoccupation with food in play and talk. It can be helped by good management, dietary control and educating your son or daughter about their diet.

If food intake isn't carefully monitored, weight gain is rapid. There are many healthy eating plans you can follow, which in itself can be confusing. The most important thing is to be consistent in your approach, and in our experience, a calorie controlled diet has been the most successful. Due to the low muscle tone and slow metabolism, children and adults with PWS require just 60% of the calories of those the same age. Exercise is also important—it will not only help to keep weight levels down, it will also improve muscle strength. Exercise can take many forms, and it doesn't have to be in a gym or exercise class. Being active with walks, swimming and cycling can all be beneficial. It's really common for individuals with PWS to gain weight on the buttocks, stomach, lower trunk and thighs. There may be some behaviours that challenge around access to food, and again this can be managed with really clear expectations and communication.

Learning abilities

Most children and young people with PWS have a definite learning need, and some have a learning disability. Your son or daughter will be able to learn and achieve great things, provided they are taught in the way they learn. You should be able to get support for your son or daughter from a range of professionals, including educational psychology, occupational therapy, physiotherapy, and speech and language therapy. There will always be an area or activity where your son or daughter really excel, whether this be jigsaws, growing plants, drawing or caring for animals, and it's important to focus on these in their learning journey.

It's not unusual for those with PWS to need SEN support – known as the following in different areas of the UK:

- Education, Health and Care Plan (EHCP) in England
- Individual Education Plan (EP) in Northern Ireland
- Co-ordinated Support Plan (CSP) in Scotland
- Additional Learning Needs (ALN) in Wales

We have information on all of the above, to help take you through the process, along with a range of educational information for families and teachers. We are here to help and can provide support letters for assessment, read and make suggestions about sections of your draft plan, and can attend annual reviews.

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