PWSA UK
PRADER-WILLI SYNDROME ASSOCIATION UK
pwsa.co.uk

# Your New Baby

## New Diagnosis Pack

# Your New Baby

Firstly, congratulations on the birth of your baby. We know that the news that your baby has Prader-Willi syndrome (PWS) will have come as quite a shock, but we want to reassure you that things are not as bleak as they seem at the moment and that the team at PWSA UK are here to help and hold your hand every step of the way.

## Getting the news

Most parents find out that their baby has PWS soon after birth. It is important for you to know that nothing done before or during pregnancy can cause PWS. It occurs in all races, social classes and in all countries throughout the world. Anyone can have a baby with PWS.

It can take a while for you to come to terms with your own feelings about the diagnosis, and they may well change over time, going through different stages. You could feel that your hopes for your child's future have suddenly been shattered and feel grief for what your child might have been or done in their life. You may feel depressed about what the future holds after your baby is born, - if this persists talk to your GP or health visitor. It could be that you feel anger and bewilderment that things turned out differently from what you had expected. You will need to come to terms with the knowledge that you and your child are now going down a different road to the one you had envisaged.

Many parents tell us that it is best to focus on the here and now and not look too far into the future. There will be good days and bad days, but before you know it, the good days will start to outweigh the bad. As the days and weeks pass you will get to know your baby, you will begin to see them as a unique person with their own individual character.

There's lots of material about PWS on the internet and it is tempting to want to know as much as possible about it, as soon as possible. Some parents gain reassurance from reading information. One of the most reliable sources is our website www.pwsa.co.uk.

"Caring for my daughter was a completely different world to the one I had been preparing for during my pregnancy and all the rules were different.

But I've found it to be a rewarding world where her achievements are greater, and just to see her cheeky smile fills me with the greatest pleasure possible.

I no longer mind the challenges I face. I've also gained confidence and a respect from others I didn't have before."

*Quote from parent.*

We think that between 30-40 babies are born each year in the UK and that there are around 2,000 individuals with the syndrome.

Your baby will have the same needs as all babies. They will need to be fed, they will sleep a lot, they will have a weak cry and need lots of love and cuddles like any other baby. The most important thing that you need to know about PWS is that everyone with the condition is a unique individual.

Children with PWS generally need more time to reach developmental milestones such as sitting-up, walking and talking and they may need some additional support when they go to school. For more information about this, you can visit our website.

## How do I feed my baby?

Many babies with PWS are tube-fed in the early days and weeks. Both breast and bottle feeding can be challenging for your baby, due to weak suck strength and sleepiness, both caused by hypotonia (low muscle tone). Failure to thrive or poor weight gain will be monitored by your health visitor.

## Should I be doing anything special or different with my baby?

In the early weeks and months, enjoy and get to know your baby. Have fun with them, and don't be afraid to be pro-active. Active and engaging play, such as gently moving baby's legs in a cycling motion whilst singing a lullaby, can be really effective. Really engage with eye contact and respond to their efforts to communicate with you, just like all parents do.

## What about the future?

It can be difficult to keep yourself from thinking about the future and, of course, it is very sensible to think about what may happen in the years ahead and to make provision for it; although as time goes by there will be new developments and changes.

One of the most important developments in recent years has been growth hormone treatment, which has transformed the lives of children with PWS. As more research is carried out, we can expect new treatments for other aspects of PWS as well. The enclosed leaflet, entitled Growth Hormone Therapy for children with PWS, gives further details.

## What is life like for families?

Having a baby with PWS usually means that family life is somewhat different to other families. However, depending on circumstances, this does not necessarily mean big changes, and introducing positive structures and boundaries can be very helpful. Balancing your baby's needs and those of other family members can bring unique challenges. Try not to compare your child to others, and never forget your child is a unique individual.

*"Our happy, contented baby smiles a hundred times a day, each one melts us.*

*We are so very lucky to have her and take each day as it comes"*

*Quote from parent.*

## How do I tell my other children about the diagnosis?

If you have other children, decide how much information to pass on to them. This will depend on their ages, understanding and curiosity and they may not remember all you tell them. Just keep listening to them and answering their questions. If you treat PWS as only one aspect of your baby's life then your other children will soon follow your lead and do the same

We have produced a leaflet for siblings which you can find at: https://www.pwsa.co.uk/wider-family-support

## What is life like for people with PWS?

Each child with PWS is individual—some will go to mainstream schools, some to specialist schools and some will start at mainstream and transfer to a special school later on. Individuals with PWS also attend colleges, some do work experience and voluntary work.

## Will my baby be healthy?

Children with PWS are generally as healthy as other children, and are also prone to the same childhood illnesses and diseases. Unless your GP or paediatrician advises otherwise, your child should receive ALL the usual childhood vaccinations against disease—there are unlikely to be any side effects from these, other than those usually experienced.

There are some health issues particular to PWS but these do vary considerably between individuals and not all children with PWS will necessarily have them all.

However, you should be aware that babies with PWS do not have a vomiting reflex and are rarely sick, even when they are not very well. Their baseline temperature may be a few degrees lower than normal so a slightly elevated temperature should be investigated.

Individuals with PWS frequently have a decreased sensitivity to pain and so there is a potential danger of underestimating a problem. As PWS babies may have a weak cry, they may not be able to let you know if they are experiencing pain. We have enclosed leaflets for both GP's and Accident and Emergency staff to make them aware of the more unique aspects of PWS.

## Is there any treatment for PWS?

Our knowledge of PWS is growing all the time and in the years since it was first recognised, treatments such as growth hormone therapy are now available which make our children's lives so much better.

Although there is currently no 'cure' for PWS, lots of research is being carried out, and there are several treatments being trialed for different aspects of PWS. The first treatment for hyperphagia has recently been approved by the FDA in the US and it is likely that other aspects of PWS will eventually become treatable.

*"We've had to deal with more emotions during the first months of our baby's life than many parents face in a lifetime. You've got to take care that PWS doesn't overtake your child's individuality, and become the most important thing about them. We're just happy to have such a loving, beautiful baby who we will never take for granted. And so far the smiles have outweighed the tears*

*We're sure everyone who comes into contact with our little girl benefits. When things get difficult sometimes, we look at her smile or watch her while she sleeps, and we know we have been given a very special person. She may need lots of time and patience, but we all get so much back."*

*Quote from parent.*

## Sharing your feelings

If you are having difficulties with coming to terms with your feelings, and you think they are having a negative effect on you, your child, or other family members, a trained counsellor may be available through your GP. However, it is often parents of other children with disabilities – not just PWS – who can offer the understanding and sympathy you may need; many will have struggled with the same feelings as you. If you would like to be put in touch with other parents who have a child with PWS do let us know.

There are several closed Facebook groups, where our families post queries, experiences and ask for advice. The groups are very supportive and are very welcoming to new parents:

**Facebook:** Prader-Willi Syndrome Association UK

**Instagram:** PWSA UK **Twitter:** PWSA UK

**PWSA UK Facebook Group, exclusively for parents:** PWSA UK Community Hub: Empowering Life with PWS

**Facebook community groups:**

- PWS Families
- Prader-Willi Syndrome Support UK
- Lovebugs of the UK (PWS) **under 5's**

*Lovebugs is a hidden group, please let us know if you would like to be invited as you will not be able to find it.*

We hold events for families in different areas of the country. To hear about these you can complete our membership form here: https://www.pwsa.co.uk/become-a-member

Remember, we're here for you every step of the way.

Our team understands PWS, will answer your questions, and guide you through each stage with understanding and without judgement.

Our office is open 9am–5pm Monday to Thursday, and 9am–1pm on Fridays.

01332 365 676

supportteam@pwsa.co.uk

# Professionals you should see

Children and young people with PWS require multi-professional advice and over-sight from infancy onwards.

This should be provided by local services, and there are a number of specialist PWS clinics —the current specialist Paediatric PWS clinics in the UK are:

- The Royal Alexandra Hospital, Brighton
- Birmingham Children's Hospital, Birmingham
- Chelsea and Westminster Hospital, London
- The Royal London Hospital, London
- The Royal Stoke University Hospital, Stoke-on-Trent
- Queen Elizabeth University Hospital, Glasgow
- Royal Hospital for Sick Children, Edinburgh
- Leeds Children's Hospital, Leeds

You can be referred to these by speaking to you GP, and we can provided a letter of support should it be needed.

Professionals you should see once you have received a diagnosis are:

- Hospital Paediatrician - general overview and referral to other services
- Endocrine Paediatrician - advice on hormones and PWS
- Consultant Community Paediatrician - to oversee local services
- Paediatric dietitian - dietary advice
- Child Development team - general development and support
- Physiotherapist - form muscle tone and core strength
- Speech and language therapist
- Occupational therapist
- Social worker from Children with Disabilities team

We know this list looks overwhelming - you do not need to see all of these professionals all of the time. They should, in time, form a network of support around you to help you through your journey.

# Feeding and weaning your child

## Feeding your baby

Failure to thrive (when a baby doesn't meet recognized standards of growth) is incredibly common in PWS, and can be counted as a characteristic of the syndrome. Your son or daughter's dietitian, speech therapist, health visitor or pediatrician are there to guide you and help you find what works best for you and your child.

## Tube feeding

For some babies with PWS, their swallow can be uncoordinated, or their suck too weak to feed properly. It's not unusual for babies to be fed by a tube, known as a nasal gastric tube (NGT). Although this can be quite overwhelming to see, it allows your son or daughter to get the right amount of nutrition to support their growth and development. Check with your health professional if oral feeding can go alongside tube feeding, and don't be afraid to introduce a dummy – using it during the tube feed will help your baby to connect an oral sensation with food. It'll also help to develop and improve their suck.

Before the tube can be removed, your little ones swallow will need to be assessed – this is usually done by a speech and language therapist and can be as simple as closely supervising your baby while they take some liquid. They may want to carry out a videoflouroscopy, an x-ray that provides a moving image of your baby's swallow in real time

## Weaning

PWS affects everyone differently, and in terms of weaning, what might work for one family may not work for another. There is no single approach to managing PWS, and it's important to manage diet in a way that works for you and your family. If, for example, you would rather your son or daughter didn't eat sweet treats, it's best not to introduce them. We are all different – some parents are happy for their little one to have the occasional treat, and others introduce snacks between meals. However you choose to do it, being consistent is key.

It's also really helpful for those around you to understand your approach – it can be difficult for them to grasp, particularly if your son or daughter doesn't show any obvious signs of PWS at this stage.

The Department of Health recommends weaning at 6 months – if your baby shows signs of being ready earlier, you can start any time after 17 weeks. You may be advised to start earlier because some babies find it easier to take first solids from a spoon rather than sucking. Ask your dietitian, health visitor or paediatrician for advice around when to start weaning if you're unsure.

There are a multitude of choices available when it comes to introducing solids. You can use pre-prepared sachets or pouches, or make your own, and it can be handy to cook several batches and keep them in the freezer. Gradually increase the lumpiness of the food as your baby gets older by mashing food rather than liquidising it, and increase the variety by using pureed lean meats, chicken, fish etc.

By the time your baby is 1, they will likely be eating the same food as the rest of the family, and please don't worry if they haven't reached this stage yet. All children, regardless of PWS, dance to the beat of their own drum and go at their own pace.

# Growth Hormone

The decision to start you child on growth hormone (GH) can feel overwhelming, and parents often have a lot of questions around its use and how it is administered. There have now been many research trials showing clear benefits for children with PWS.

## Benefits of Growth Hormone:

- Increased growth (height)
- Improved motor development, muscle strength and tone and better body composition (ie more mucle mass and less fat mass)
- Improved tolerance for exercise
- Improved bone health
- Improved respiratory function
- Improved congnition

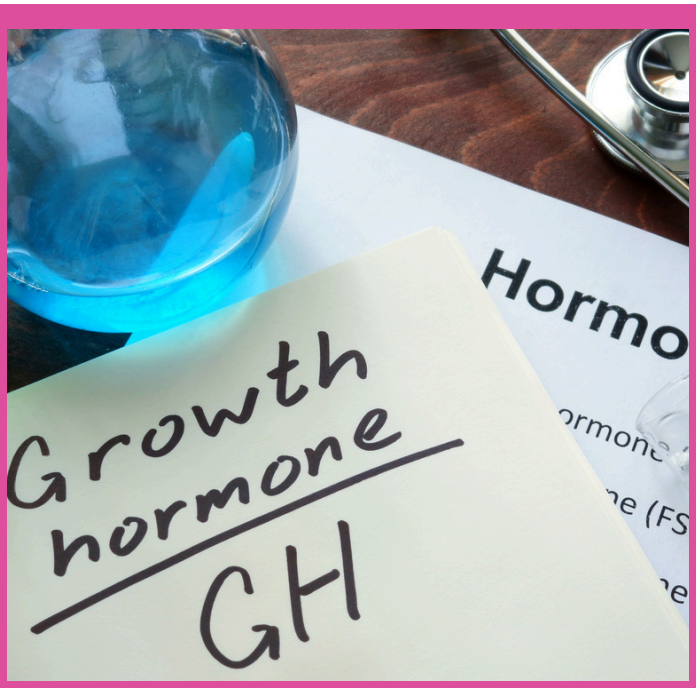
GH is usually given in the form of a daily injection, and parents are offered full training in how do to this correctly,

## Should every child with PWS Have GH treatment?

It should be considered for every child with PWS, and the NHS will provide treatment if their endocrinologist recommends it, and has confirmed it is safe to go ahead. However GH treatment is not compulsory and some families decide not to have it for their child.

There are some medical reasons why treatment with GH cannot be started:

- A significant illness in addition to the PWS
- Severe obesity
- Severe breathing problems
- Untreated central and/or obstructivve sleep apnoea
- Uncontrolled diabetes




## What age should GH treatment start?

It is recommended by our PWS specialist Clinics that GH is started by the age of 12 months.

## How to obtain GH treatment for your child

GH treatment is always set in motion and monitored by a specialist - generally a paediatric endocrineologist. However, the actual steps involved in this process can widely vary between different regions snd hospitals. GP generally prescribe the GH on the instruction of the specialist, but do not usually initiate treatment.

The generic process is as follows:

- A specialist decides when to start GH for a child with PWS. Specific tests to prove GH deficiency are not required, but it is essential to prove the diagnosis of PWS by genetic analysis
- The specialist who initiates the GH will organise carious tests prior to the start of treatment, such as a sleep study and blood tests

- Parents decide which GH preparation to use based on their own personal preferences and the devices available - the specialist should prove information and guide them.
- Training is then organised for the parents and if appropriate the patient. The training is usually provided by a specialist nurse or by the GH company itself.
- Depending on the local processes, the specialist of the patient's usual GP provides the prescriptions for GH on a regular basis. Most GH companies provide additional services like collections of the prescription and direct home delivery.

## Why should your child have a sleep study?

A sleep study is used to determine if there are any underlying respiratory issues, and it's really important that every child has a study carried our before commencing GH treatment. Usually involving an overnight stay in hospital, it is painless and doesn't have any side effects for your child. It will involve placing ECG pads on your child's chest, a probe on their toe or finger and possibly a stretchy band with a sensor across their chest. on occasion, a sleep study may show that tonsils or adenoids need to be removed - if this is the case, a secondary sleep study will need to be carried out after surgery before commencing GH treatment.

## Monitoring treatment

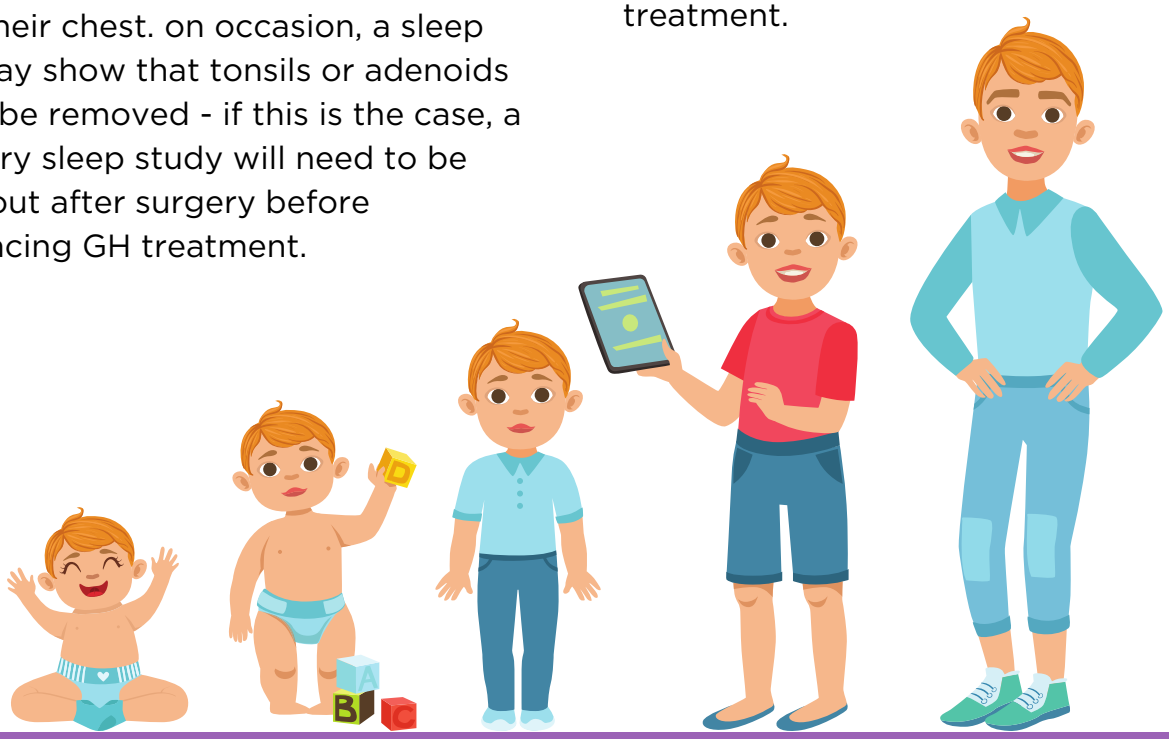
Your child should be monitored every 3-6 months to ensure that the treatment is both safe and effective. This can involve blood tests to look at levels of GH and repeat sleep studies.

## Weight managements during GH treatment

GH treatment does not change the problems of increased appetite in PWS and is not a solution to weight gain. Children with PWS need less calories, approximately 60 - 80% compared to children without PWS, and regular exercise is really important. GH treatment will help develop muscle strength, making exercise not only easier, but more beneficial.

## Length of time for treatment

Unless there are reasons why GH is no longer appropriate, treatment should continue until your child has stopped growing. Some adults with PWS are being offered GH treatment, although at the moment there is less research evidence for this and no NHS guidance for treatment.



# Information for GP's

## Overview

Prader-Willi syndrome: A rare genetic abnormality of Chromosome 15.

## Characteristics

Hyperphagia:

- Overwhelming appetite leading to obesity.
- Food foraging - ingestion of inappropriate items needs to be considered.
- Sometimes excessive fluid intake.

Hypotonia:

- High fat: muscle ratio.

Lack of vomiting reflex:

- Vomiting should be viewed seriously.
- Gastroparesis and constipation may occur after excessive eating

High pain threshold

- Watch out for unreported injuries/fractures.
- Beware abdominal pain.
- Unexplained bruising.

Abnormal temperature control:

- Slight pyrexia must be viewed seriously

Hypogonadism:

- Rare but possible for female to fall pregnant (reported) and male to father a child (none reported).

Intellectual ability

- Range 50-100, with most in 60-65 region.
- Delayed social/emotional skills .
- Challenging behaviour

Capacity:

- The drive to eat overwhelms rational decision making.
- Without supervision life-threatening amounts of food are consumed.
- These characteristics are not uniform but occur over a spectrum.

Other features include:

- Obesity related problems.
- Obstructive sleep apnoea.
- Osteoporosis.
- Diabetes type 2.
- Oedema.
- Skin picking leading to cellulitis.

# Detailed information for GP's

## Characteristics of Prader-Willi Syndrome (PWS)

PWS is a rare and complex genetic syndrome, first described in 1956, which affects people throughout their life. In about 95% of cases it is a de novo genetic abnormality affecting chromosome 15. There are probably no more than 2000 people with the syndrome throughout the UK. It is very rare for an adult with PWS to live totally independently; most still live with parents or relatives, or are in residential care or supported living.

The range and severity of symptoms differs between individuals, but all will have the following characteristics in some degree:

## Hyperphagia (overeating)

This is still not fully understood, but appears to stem from a dysfunction in the hypothalamus which means that the person does not know when they have had enough to eat - hence they feel hungry most of the time. The desire to eat can overwhelm all rational decision-making processes with regard to food.

## Hypotonia (low muscle tone) and abnormal body composition

The person is less active and needs fewer calories than another adult of the same age and height to maintain a healthy weight. Most individuals have a higher fat to muscle ratio than normal. As much as 50% of body weight is fat mass. It is thus advisable to start medications with a lower dose than normal.

## Hypogonadism (immature sexual development)

Some people with PWS may have already experienced the first signs of sexual development as early as 5 years, with pubic and underarm hair appearing. However, in the majority of cases, full sexual development does not occur in either men or women with PWS. Women may not experience the onset of periods, or they may be very erratic, and breast development may be slow. Men's voice may not break and facial hair can be very scanty. Sex hormone treatment will help with these issues – specialist input from an endocrinologist is often helpful. Whilst infertility is generally thought to be the norm within both sexes, there have been reports worldwide of women with PWS having a child (which, if the woman has the deletion form of PWS, has a 50- 50 chance of being born with Angelman syndrome).

## Short stature

Women average around 4' 9" (145 cm) in height and men around 5' 2" (158 cm). However, stature may be closer to that of other family members if the individual has received growth hormone as a child. A few individuals with PWS, however, even without growth hormone, are within normal height ranges.

## Obesity

The combination of hyperphagia and hypotonia (especially when exacerbated by short stature) means that the person with PWS will become morbidly obese if external controls are not in place to prevent them accessing food. In addition, they usually require fewer calories per day than those without PWS to maintain a healthy weight.

## Life expectancy

With restricted access to food, an individual with PWS can live well into middle age and beyond. The oldest known person with PWS died at age 74 - she had lived in residential care for the greater part of her life.

## Immature or delayed social and emotional skills and challenging behaviour

Most people with PWS operate at a lower social and emotional level than their chronological age and intellectual level, sometimes displaying behaviours which can be seen in young children (eg temper outbursts) and refusal to comply with requests. Some can appear very able on first meeting, but this often masks difficulties in understanding.

## Intellectual abilities

Intellectual abilities range from 50 to over 100. The majority are in the 60-65 range.

## Various cognitive deficits

Including:

- Problems with attention-switching, resulting in oppositional behaviour and resistance to change.
- Problems with auditory processing - find it difficult to carry out instructions given verbally.
- Perseverative speech.

# Health care for people with PWS – unusual features

People with PWS are generally as healthy as the general population and are also prone to the same illnesses and diseases. However, there are some unusual features of PWS which require special attention and may not always be evident to those who have little experience of the syndrome. The following is a list of those features. Please note that they vary in degree between individuals with PWS and not all people with PWS will necessarily have all the features.

## High pain threshold

Individuals with PWS frequently have decreased sensitivity to pain. Therefore, known injuries must be assessed for more serious problems, and signs of unreported injuries should be sought. In the absence of a verbal complaint of pain, other symptoms of specific injuries should be evaluated.

## Easy bruising

Many individuals with PWS bruise easily but, because of the high pain threshold, are often unable to say how they came by the bruise.

## Lack of vomiting reflex

Probably due to low muscle tone. If someone with PWS has a history of rare vomiting and presents with this symptom, it should be taken very seriously.

## Abnormal temperature regulation

An individual with PWS may not be pyrexial even when seriously ill and may run dramatically below-normal temperatures at times. Even slight temperature elevations should be

considered a warning sign. It is advisable to keep a record of the person's temperature when healthy, as this is sometimes below the normal average.

## Food foraging

If left unsupervised, an individual with PWS may consume life-threatening amounts of food. A dramatic weight increase within a day - especially if coupled with reports of stomach distress or vomiting - may be a sign that the person is severely ill. Loss of appetite can also be a sign of illness.

People with PWS can be quite indiscriminate in what they eat, eg poisonous berries, out-of-date food, frozen food, food from waste bins or off the ground. The lack of vomiting reflex (see above) may not alert the observer to the fact that the person has ingested items such as these.

## Excessive fluid intake

There have been a few reports of people with PWS drinking excessive amounts of fluid, leading to potentially fatal low sodium and potassium levels.

## Loss of appetite

Most people with PWS will continue to eat during illness and do not seem to experience the same loss of appetite as others might. A report of loss of appetite may be an indication of a serious illness. However, this should not be confused with a refusal to eat as a result of challenging behaviour.

## Skin-picking

Skin picking or spot picking is very common in PWS. Skin picking is often provoked by small spots and grazes, which are picked continually, and thus never allowed to heal. However, sometimes wounds are made where there was no wound previously. Any area of the body can be a target. Most common are the limbs and the head or face, but other areas may be involved. There is an increased risk of infection as a result.

## Undetected bone fractures

Due to the high pain threshold, it is not unusual for a person with PWS to have an undetected broken bone. Following a fall or other injury, a person with PWS should be closely monitored for a change in walking or arm movement. Observe for deformities, swelling or bruising. In addition osteoporosis ('thin bones') is more common (because of the hypotonia and lack of sex hormones) and therefore a fracture may result from relatively minor trauma.

## Abdominal pain

Individuals with PWS do not commonly exhibit a vomiting reflex. If a person with PWS suddenly reports abdominal pain or bloating, is vomiting, or has abdominal distention, there may be life-threatening bowel inflammation or necrosis, and emergency surgery may be needed

## Gastroparesis and constipation

This is a condition that is common with PWS and can be more life threatening then in a typical situation. A person with Prader-Willi syndrome when diagnosed with gastroparesis may need hospitalization. People with PWS also commonly present with constipation, probably due to low muscle tone in the digestive tract.

## Risk of choking

There is a risk of choking arising from people with PWS trying to eat food quickly, either because they habitually do this, or because they are trying to disguise the fact that they have taken food to eat which they should not have done. There have been reports of deaths in adults with PWS from this cause.

## Obesity-related problems

High blood pressure, diabetes, oedema, congestive heart failure and respiratory failure are the most common problems for the adult who is significantly overweight.

## Obstructive and central sleep apnoea

Sleep apnoea is relatively common and but may also occur in those who are not seriously overweight. Sleep checks are advisable every few years.

## Risk from pneumonia

People with PWS may be more liable than the general population to pneumonia, which can have a very sudden onset and sometimes prove fatal. Adults should be offered a pneumonia vaccination.

## Oedema and lymphoedema

Swelling of the legs and feet are common in PWS, especially in those who are overweight. However, it can also occur in those who are not significantly overweight. There is a heightened risk of cellulitis.

## Anaesthesia

There is nothing inherent in PWS which gives cause for concern with the administration of anaesthesia. However, individual health problems related to PWS should be taken into account.

These include:

- Obesity (complications caused by obstructive apnoea, pulmonary hypertension, altered blood oxygen or blood carbon dioxide levels, significant oedema).
- High pain threshold
- Temperature instability - parent or carer should be asked for information about patient's usual temperature
- Thick saliva - may complicate airway management
- Food seeking behaviours - the person may have eaten food even if they say they have not. Unless carer or parent can verify this, the person should be assumed to have food in their stomach.
- Hypotonia may cause difficulties in ability to cough and clear airways
- Excessive post-operative drowsiness in some individuals

# Information for Accident and Emergency (A&E) and other hospital staff

Prader-Willi Syndrome (PWS) is a rare genetic condition which is present from birth. Caused by an abnormality on chromosome 15, it affects the functioning of the hypothalamus and other aspects of the brain and muscles. Main characteristics of the syndrome are:

- Hyperphagia – beginning around 2-4 years
- Hypotonia – from birth
- Hypogonadism
- Short stature, compared with other family members (less evident if receiving growth hormone treatment)
- Delay in and difficulties with emotional and social development
- Developmental delay /learning disabilities ranging from severe to borderline. A minority of people with PWS do not have a general learning disability. Most have specific difficulties eg - slow processing of verbal information etc
- Challenging behaviours, (there is a PWS behavioural phenotype), whether or not the person has a learning disability.
- A significant number of individuals with PWS also have Autistic Spectrum Disorder or some features thereof.

## Unusual features and potential risk factors in PWS

A high pain threshold, poor temperature regulation and lack of vomiting associated with hypothalamic dysfunction can mean that a serious medical problem may present in an unusual way and so could be overlooked.

## Abnormal body composition affecting medication

Individuals with PWS have reduced lean tissue and increased adiposity. This may affect dosages of medication, which may need to be less than usual. Psychotropic medications must always be started at very low doses. Medications that have antidiuretic effects may cause water intoxication.

## Body temperature abnormalities

An individual with PWS may be unaware when they are too hot or too cold, or eat or drink very hot food or liquids. Hyper and hypothermia have been reported. Hyperthermia may occur during minor illness and after anaesthesia. Fever may be absent despite serious infection. In cases of hypothermia, the patient may not report feeling cold. Baseline temperature may be below 98.6˚F/37˚C.

## High pain threshold

The majority of people with PWS have a high pain threshold and hence underreport pain. This may mask fractures, acute abdominal conditions, serious infections and internal injuries. Individuals may have difficulties localising pain.

## Lack of vomiting

Vomiting rarely occurs in PWS but some children with PWS do vomit with minor illnesses. The absence of vomiting cannot therefore be relied upon as reassuring when assessing illness. However, the presence of vomiting, particularly when the individual has a history of never or rarely vomiting, may signal a very serious or life-threatening illness (see below). Emetics may be ineffective.

# General care while an individual with PWS is in hospital treatment

## Anaesthesia

There is nothing inherent in PWS which gives cause for concern with the administration of anaesthesia. However, individual health problems related to PWS should be taken into account.

## Cortisol levels

Individuals with PWS are probably more likely that the rest of the population to have cortisol deficiency. If there is clinical suspicion of cortisol deficiency during acute illness, immediately take a blood sample to check serum cortisol level. This can then be used to help with diagnosis even if you need to start hydrocortisone as an emergency. Please discuss results with your local endocrinology team.

## Medication

People with PWS may have abnormal reactions to standard doses of medication.

## Patients receiving growth hormone replacement

Growth hormone (GH) replacement is often used in patients with PWS. If felt necessary, it can be temporarily stopped during an acute illness without any likely acute adverse consequences. Patients and families may however wish to continue GH to help maintain routine. Please discuss with your local endocrinology team if any questions or concerns.

## Oxygen levels

Oxygen levels may be normally lower than usual. Obese patients who have been chronically hypoxic may not tolerate fully corrective use of oxygen and are likely to start retaining CO2.

### Websites with extensive information about PWS

PWSA UK https://www.pwsa.co.uk/

International PWS Organisation https://www.ipwso.org/

PWSA USA www.pwsausa.org

# Benefits you can claim

There are a number of benefits that you are able to claim from birth onwards. This leaflet is just to give you a brief outline of those available, and when you are ready, we will be more than happy to help you apply for benefits that are relevant to you.

Child benefit - paid every four weeks.

https://www.gov.uk/child-benefit

Universal credit, which may include disabled element.

https://www.gov.uk/universal-credit

Carers allowance (providing you earn less than £120 a week)

https://www.gov.uk/carers-allowance

Disability living allowance (DLA) from the age of 3 months

https://www.gov.uk/disability-living-allowance-children

We have further information to help you apply for the above, and this can be requested by contacting the Support Team on 01332 365676 or by emailing supportteam@pwsa.co.uk.

# Welcome To Holland

*By Emily Perl Kingsley*

I am often asked to describe the experience of raising a child with a disability - to try to help people who have not shared that unique experience to understand it, to imagine how it would feel. It's like this.....

When you're going to have a baby, it's like planning a fabulous vacation trip - to Italy. You buy a bunch of guide books and make your wonderful plans. The Coliseum. The Michelangelo David. The gondolas in Venice. You may learn some handy phrases in Italian. It's all very exciting.

After months of eager anticipation, the day finally arrives. You pack your bags and off you go. Several hours later, the plane lands. The flight attendant comes in and says, "Welcome to Holland."

"Holland?!?" you say. "What do you mean Holland?? I signed up for Italy! I'm supposed to be in Italy. All my life I've dreamed of going to Italy."

But there's been a change in the flight plan. They've landed in Holland and there you must stay.

The important thing is that they haven't taken you to a horrible, disgusting, filthy place, full of pestilence, famine and disease. It's just a different place.

So you must go out and buy new guide books. And you must learn a whole new language. And you will meet a whole new group of people you would never have met.

It's just a different place. It's slower-paced than Italy, less flashy than Italy. But after you've been there for a while and you catch your breath, you look around.... and you begin to notice that Holland has windmills....and Holland has tulips. Holland even has Rembrandts.

But everyone you know is busy coming and going from Italy... and they're all bragging about what a wonderful time they had there. And for the rest of your life, you will say "Yes, that's where I was supposed to go. That's what I had planned."

And the pain of that will never, ever, ever, ever go away... because the loss of that dream is a very very significant loss.

But... if you spend your life mourning the fact that you didn't get to Italy, you may never be free to enjoy the very special, the very lovely things ... about Holland.

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