PWSA UK
PRADER-WILLI SYNDROME ASSOCIATION UK

Healthcare in PWS
Birth to 2 years

Introduction

Children with PWS are generally as healthy as other children, but are also prone to the same childhood 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 or when the unusual features are subtle or mild and hence more difficult to identify.

These features are included in this article, along with some health issues which are quite common in children with PWS.

Please note that the issues vary considerably between individuals with PWS and not all children with PWS will necessarily have them all. As time goes by, you will be able to identify which of those issues may apply to your child.

NOTE

For ease of reading, the text refers to "he, him, his", but the details are equally relevant to girls, unless otherwise stated.

Squint (strabismus)

A squint, where one or both eyes point in different directions, is common in babies and children with PWS, and is most likely due to poor muscle tone. Corrective treatment is by patching or a simple surgical operation.

Your paediatrician or eye specialist will advise you on timing and best form of treatment. See also Vision and Care of the Eyes in PWS https://www.pwsa.co.uk/assets/files/eyes-birth-13.pdf

Information for hospitals and GPs

You will find further information to give to hospitals and GPs included in this pack or on our website at https://www.pwsa.co.uk/information-for-professionals/health

Issues which occur only in infancy

Hypotonia (low muscle tone) makes sucking more difficult and failure to thrive or poor weight gain is the major problem in many PWS infants. Most infants also have a weak cry and are quite sleepy.

Respiratory problems are common in these infants and assistance with clearing secretions may be needed. Insufficient fluid intake, due to feeding difficulties, can occur.

An infant fed by nasogastric tube needs close monitoring, due to risk of reflux and aspiration.

Orthopaedic problems

Scoliosis

Children with PWS have a high risk of developing scoliosis (curvature of the spine to one side). Hence your child's spine or back should be examined regularly (or at least once a year as a minimum) to identify if he has scoliosis and to institute any intervention if needed.

Scoliosis develops in children with PWS for a number of reasons. Children do have poor muscle tone and poor muscle power; this often leads to poor posture and development of scoliosis. Often associated obesity or increased weight can make the scoliosis worse.

Parents worry that treatment with growth hormone can precipitate or worsen the scoliosis. However, growth hormone is generally safe and does not cause an increased incidence of scoliosis. However, in those who already have scoliosis, it may worsen and hence should be monitored more closely. If a particular child has severe scoliosis, then growth hormone therapy is contra-indicated.

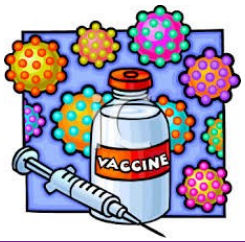
Conservative measures like good posture, good care of the back and physiotherapy are all helpful and useful. Generally no other treatment is required for mild scoliosis. However for moderate to severe scoliosis, interventions are needed (such as a spinal brace). In very severe and complex cases of scoliosis (and additional kyphosis or forward bending of the spine) surgical intervention may be needed.

Hip dysplasia

Babies and young children should also have hip checks from an early age to look for symptoms of hip dysplasia. If treated early, there should be no long term problems.

Vaccinations

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.



Respiratory problems/Obstructive sleep apnoea

Children with PWS may have respiratory problems and are prone to chest infections. Sleep apnoea (where the child momentary stops breathing while asleep) is sometimes seen in children with PWS; this is exacerbated when the child also has increased weight gain or obesity. When sleep apnoea is suspected, please alert your doctor because this will require further specialist investigations and treatment as appropriate.

Risk of choking

Choking happens when food or fluids gets into the airway, rather than going into the stomach. This can happen in any child, but lack of vomiting reflex makes it more difficult and tricky to identify in a child with PWS. Hence, do not try to feed your child faster than he is able to cope with. Always ensure that the feeding position for your child is appropriate at each feed.

Undescended testes in boys

Undescended testes are quite common in boys with PWS. Your paediatrician will advise you on the timing for the best course of treatment, which often involves a simple operation to bring down one or both testes.

Bruising

Many children with PWS can bruise easily; additionally they may not cry out at the time of injury or accident, due to high pain threshold. Hence the presence of a bruise should alert you to actively look for any other signs of an injury in the child.

Temperature

A child with PWS may have below-normal temperatures at times and may not have a high temperature even when seriously ill. Even slight temperature elevations should be considered as a warning sign and to observe your child closely for any other clues. If in doubt, please ask your doctor to examine him.

Vomiting/abdominal pain

Individuals with PWS do not commonly exhibit a vomiting reflex. Many babies with PWS are rarely sick, even when unwell. If your child has a fever, or is not responding as they usually do, you should seek your doctor's help, to check them out.

Lack of vomiting cannot be taken as a sign that little is wrong with your child.

PWS Specialist Clinics

In a few areas of England and Scotland, hospitals run specialist clinics for children with PWS.

Specialist clinics currently run at:

- **Chelsea & Westminster Hospital, London**
- **Royal Alexandra Hospital, Brighton**
- **Birmingham Children's Hospital**
- **Royal Hospital for Sick Children, Glasgow**
- **Royal Stoke University Hospital, Stoke on Trent**

These clinics allow you to see several specialists in one day and are staffed by professionals with a special interest in PWS.

PWSA UK representatives attend the clinics, when possible, to offer non-medical support.

High pain threshold

Individuals with PWS frequently have decreased sensitivity to pain and thus there is a potential danger of underestimating the problem.

Also, as PWS babies may have a weak cry, they may not be able to alert you if they are experiencing pain.

Thus, all known injuries must be assessed by a GP or paediatrician to exclude any serious problems. In time, you will be able to recognise subtle signs when your child is out of sorts or unwell.

Following a significant fall or other injury, your child should be closely monitored for a change in posture, walking or movement of limbs.

Observe for deformities, swelling or bruising as these may indicate an undetected broken bone or fracture.

Low cortisol levels

Cortisol is an important stress hormone which the body produces all the time, but increases the production to help deal with illness, infections or stress. There have been several studies looking at adrenal function in individuals with PWS. A study from Holland raised the possibility that deficiency of cortisol was much more common than expected in PWS; however later studies have not reproduced this result.

Individuals with PWS are probably more likely that the rest of the population to have cortisol deficiency, and the current guideline suggests that individuals are not tested routinely, but a test should be done if there is any concern. Symptoms of low cortisol include tiredness, low blood pressure, episodes of low glucose or fainting.

Anaesthesia

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

- **Obesity** (may be associated with obstructive sleep apnoea, high blood pressure, poor breathing pattern (hypo-ventilation) which is further exacerbated by hypotonia or poor muscle tone etc.).
- **High pain threshold** (see previous page).
- **Temperature instability** - you should inform the anaesthetist about your child's usual temperature.
- **Food seeking behaviour** is common in children with PWS; though generally not in infants. However, the anaesthetist or health professional should assume that your child has eaten and has food in his stomach, unless you can verify otherwise.
- **Low muscle tone (especially in infants)** may cause difficulties in their ability to cough and clear secretions from their airway. In addition, thick saliva may further complicate airway management.
- **Excessive post-operative drowsiness** may be present in some children.

PWSA UK

Suite 4.4 , Litchurch Plaza
Litchurch Lane
Derby DE24 8AA

T: 01332 365676
E: admin@pwsa.co.uk
W: www.pwsa.co.uk

Reg Charity No: 1155846

Further information about anaesthesia

You can download information about anaesthesia and PWS to hand to your child's anaesthetist at:
www.orpha.net/data/patho/Pro/en/Prader_Willi_EN.pdf

Thank you

Thank you to Dr Shankar Kanumakala from the PWS Clinic, Royal Alexandra Hospital, Brighton, and to Dr Nicola Bridges from the PWS Clinic, Chelsea & Westminster Hospital, for their help in checking this document.