

Healthcare in PWS 13–18 years



Photo by Kate Lloyd
Photography

Introduction

Teenagers with PWS are generally as healthy as other teenagers, and are also prone to the same teenage 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 people with PWS. Please

note that the issues vary considerably between individuals with PWS and not all teenagers with PWS will necessarily have them all.

Food seeking

As your son or daughter gets older, they are likely to experience varying degrees of independence; this can lead them to having access to food and food outlets.

Going to college is a typical example of when this occurs. Some cope with this better than others, but generally speaking it will mean that others within your son or daughter's circle should be made aware that access to food should be controlled.

Individuals with PWS may be quite indiscriminate in what they eat, e.g. poisonous berries, out-of-date food, frozen food, food from waste bins or off the ground. There are reports of people with PWS eating large quantities of items like salt or shampoo, though this is not thought to be very common.



NOTE

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

Vomiting

Individuals with PWS do not commonly exhibit a vomiting reflex. Many people with PWS are rarely sick, even when unwell.

If your son or daughter 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 son or daughter. See also the section on TEMPERATURE

Continued ...



Food seeking (continued)

The lack of vomiting reflex (see page 1) may mask or hide the fact that the person has ingested inappropriate items. A dramatic weight increase within a day - especially if coupled with reports of abdominal distress (and occasional vomiting) - may be a sign that the person is severely ill. Similarly, symptoms such as loss of appetite, diarrhoea or significant fluid retention are also reasons for concern. (See also gastric problems on page 4).

Skin lesions/oedema/cellulitis

A common feature of PWS is skin-picking, which may seriously worsen at this age, and can be exacerbated by anxiety. The Behaviour Management article, included in this pack, gives you information about how to manage this. It is important to monitor any wounds for infection and obtain appropriate treatment from your GP if this occurs.

If your son or daughter is very overweight, they may also have oedema or lymphoedema (fluid in the limbs, usually legs and feet), which produces swelling. Sometimes the skin can get infected and cellulitis (inflammation and infection) can occur. This requires immediate treatment.

Vaccinations

Unless your GP or paediatrician advises otherwise, your child should receive ALL the usual childhood vaccinations against disease, including flu- there are unlikely to be any side effects from these, other than those usually experienced.



Orthopaedic problems

Young people with PWS have a high risk of developing scoliosis (curvature of the spine to one side). Hence their spine or back should be examined regularly (or at least once a year as a minimum) to identify if they have scoliosis and to institute any intervention if needed.

Scoliosis develops in PWS for a number of reasons. Individuals 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 young person 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.

Respiratory problems/Obstructive sleep apnoea

Teenagers with PWS may have respiratory problems and are prone to chest infections. Sleep apnoea (where the person momentarily stops breathing while asleep) can become more of a problem in teenagers with PWS, especially if they are obese. If sleep apnoea is suspected, alert your doctor, because this will require further specialist investigations and treatment as appropriate.



Health care in PWS

High pain threshold

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

Thus, all known injuries must be assessed by a GP or paediatrician to exclude any serious problems. If your son or daughter shows symptoms that are out of the ordinary, it is also a good idea to have him or her checked over.

Following a significant fall or other injury, your son or daughter 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.

Bruising

Many people with PWS can bruise easily; additionally they may not notice 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 your son or daughter.

Temperature



Individuals 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 son or daughter closely for any other clues. If in doubt, please ask your doctor to examine your son or daughter. It is also a good idea to ask your doctor to make a note of their temperature when they are healthy so that you have a base line guide for variations

Risk of choking

Choking happens when food or fluids gets into the airway, rather than going into the stomach. This can happen in any person, but lack of vomiting reflex makes it more difficult and tricky to identify in a person with PWS. There is a risk of choking when people with PWS try to eat food quickly, either because they habitually do this, or because they are trying to disguise the fact that they eating when they shouldn't. Encourage your son or daughter to eat slowly and chew food properly before **swallowing**.



PWS Specialist Clinics

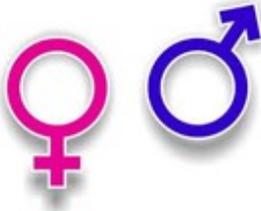
In a few areas of England and Scotland, hospitals run specialist clinics for children with PWS.

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.



Sexual development

Some teenagers may have already experienced the first signs of sexual development, sometimes starting 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.

Teenage girls may not experience the onset of periods and breast development may be slow. Teenage boys' voice may not break. Sex hormone treatment will help with these issues – specialist input from a paediatric endocrinologist should be sought.

Obesity-related problems

Obstructive sleep apnoea and breathing difficulties (respiratory failure) are the most common problems for the young person who is significantly overweight. Congestive heart failure and pulmonary hypertension may also occur at this age due to significant obesity.



Good and pro-active dietary management can help prevent obesity or obesity related problems getting worse. However, it is extremely important to have regular check-ups to ensure weight is within the normal range; also this will help to put measures in place if weight starts to creep up. Regular checks to exclude diabetes and high blood pressure should be considered.

Mental health problems

Some teenagers with PWS may experience mental health problems. These can include: depression, lethargy, severe anxiety, and occasionally acute psychotic episodes. Professional psychiatric help should be sought in these cases.

Researchers have found that those with the maternal disomy form of PWS are more liable to psychosis, so if your son or daughter has this type of PWS, and begins to show unusual behaviour patterns, you should obtain a referral to a psychologist or psychiatrist.

Severe gastric illness

Abdominal distention or bloating, pain, and/or vomiting may be signs of life-threatening gastric inflammation or necrosis, more common in PWS than in the general population. Rather than localized pain, there may be a general feeling of unwellness. If an individual with PWS has these symptoms, close observation is needed. An X-ray and an endoscopy with biopsy may be necessary to determine degree of the problem and possible need for emergency surgery.

Gastroparesis

Another consideration is gastroparesis, a weakness of the stomach that causes delayed stomach emptying. This is a condition that is common with PWS and can be more life threatening than in a typical situation. A person with Prader-Willi syndrome when diagnosed with gastroparesis may need hospitalization. Eating while the stomach is distended with gastroparesis can be very dangerous.

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 than 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 range.
- **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 younger children)** 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.

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



Health care in PWS

Information for health professionals

You can use articles for hospital staff and GPs to tell them more about the health aspects and risks in Prader-Willi syndrome.
<https://www.pwsa.co.uk/information-for-professionals/health>

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PWSA UK

Suite 4.4 ,
Litchurch Plaza

Litchurch Lane

Derby DE24 8AA

01332 365676

admin@pwsa.co.uk

www.pwsa.co.uk

Reg Charity No:
1155846

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