



Health care in PWS Adults



Introduction

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 and not all people with PWS will necessarily have them all.

Food seeking

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. Stomach rupture is possible. Diarrhoea or significant fluid retention are also reasons for concern.

People with PWS can be quite indiscriminate in what they eat, eg poisonous berries, out-of-date food, frozen food, food from waste bins or off the ground. 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.

The lack of vomiting reflex may not alert the observer to the fact that the person has ingested items such as these.



Note:

For ease of reading, the text refers to "he/him" but all the information is equally applicable to girls.

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.

There have been several reports of individuals with PWS who have broken bones which have not been immediately identified, due to the person's high pain threshold.



Vomiting/abdominal pain

Individuals with PWS do not commonly exhibit a vomiting reflex. If your son or daughter suddenly reports abdominal pain or bloating, is vomiting, or has abdominal distension, there may be life-threatening digestive problems, and emergency surgery may be needed. It also may be a warning that they have ingested a large amount of food. In either case, death can occur in hours.

Recent reports have highlighted deaths due to "binge-eating", especially when the person with PWS usually has their food intake well-controlled and does not have a significant weight problem. Lack of vomiting cannot be taken as a sign that little is wrong with your son or daughter. (See also the section on **Temperature** below).

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 hospitalisation. Eating while the stomach is distended with gastroparesis can be very dangerous.

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.



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, your son or daughter 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.

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** <https://www.pwsa.co.uk/assets/files/behaviour-adults.pdf>

article, 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.



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. Treatment is usually by bandaging the legs and/or wearing special support stockings.

Sometimes the skin can get infected and cellulitis (inflammation and infection) can occur. This requires immediate treatment to prevent septicæmia.

Respiratory problems/Obstructive sleep apnoea

Excessive weight in addition to the hypotonia of PWS can lead to serious respiratory problems. Sleep apnoea is common and may also occur in those who are not seriously overweight. If sleep apnoea is suspected, 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. There have been reports of deaths in adults from this cause. 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.



Vaccinations

Unless your GP or consultant advises otherwise, your son or daughter can receive all the usual vaccinations against disease, including flu and pneumonia - there are unlikely to be any side effects from these, other than those usually experienced.

Obesity-related problems

Obstructive sleep apnoea and breathing difficulties (respiratory failure) are the most common problems for the 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 in place for all adults with PWS.



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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.



Risk of 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.



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.



Sexual development

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 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 should be sought.

Whilst infertility is generally thought to be the norm, there have been at least four 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). There are no reports of a man with PWS fathering a child.

We currently know very little about whether with PWS are affected by the menopause., but as more women with PWS reach and pass middle age, our knowledge about this is likely to increase.

Mental health problems

Some adults with PWS may also experience mental health problems. Researchers have found that those with the disomy type of PWS are more liable to these problems, although they can still be found in those with the deletion type. Problems can include: depression, severe anxiety, lethargy, visual and auditory hallucinations (seeing things that are not there and hearing voices), and acute psychotic episodes. Professional psychiatric help should be sought in these cases.

Annual health checks for people with PWS



Prader-Willi Syndrome has a number of characteristics and potential health problems which may not be included within the framework of a standard annual health check for adults with learning disabilities, as recommended in the government document "Valuing People Now". These are:

- Weight
- Sleep apnoea (obstructive, central or mixed) - even where obesity is not a problem
- Blood pressure
- Cholesterol levels
- Scoliosis, kyphosis
- Osteoporosis (in both males and females, at young age, due to lack of sex hormones)
- Oedema
- Cellulitis
- Hypothyroidism
- Gastroparesis
- Diabetes (type 2)
- Skin infections (due to skin-picking)
- Vitamin D deficiency

A more extensive checklist has been produced by the International PWS Organisation and can be found at <https://www.ipwso.org/famcare>

Cortisol hormone insufficiency

There has been a suggestion that some people with PWS may be at increased risk of stress hormone (cortisol) deficiency due to hypothalamic problems but this has not been found in all studies and probably occurs only rarely. If there is a clinical suspicion of cortisol deficiency during an acute illness such as an infection (e.g. low blood sugar, low and variable blood pressure), then consideration should be made to urgently measure cortisol levels and if there is concern hydrocortisone should be administered until the results are available.



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** (complications caused by obstructive apnoea, pulmonary hypertension, altered blood oxygen or blood carbon dioxide levels, significant oedema).
- **High pain threshold** (see front page).
- **Temperature instability** - you should inform the anaesthetist about your son or daughter's usual temperature.
- **Food seeking behaviour** - an individual may have eaten food even they say they have not. This should be assumed, unless you can verify otherwise.
- **Low muscle tone** may cause difficulties in their ability to cough and clear secretions from their airway.
- **Thick saliva**—may complicate airway management.
- **Excessive post-operative drowsiness** may be present in some people with PWS.

Further information about anaesthesia

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



Information for health professionals

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

Health care Adults

Life expectancy

Early death as a result of morbid obesity is not uncommon, and unfortunately can occur in young adults. However, with restricted access to food, the person can live well into middle age and beyond.

The oldest known person with PWS died at age 74 - she had lived in an institution for the greater part of her life.

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