

“Good health” checklist

The basics of a healthy adult life for someone with Prader-Willi syndrome (PWS) include a healthy, appropriate eating plan and regular, effective physical activity to avoid obesity, to keep the body fit and to maintain good mental health. Before any of these can be achieved the people working with the person who has PWS must have a good understanding of the complexities of the syndrome.

This “checklist” is to assist you in maintaining good health for your person with PWS. It is to be shared with other family members, professionals and caregivers who are involved with your person with PWS.

PWS is a genetic disorder which is due to a lack of expression of particular genes on the 15th chromosome. It affects several systems in the body. Below are listed some common problems seen in people with PWS, what the effect of the problem is on health and how it needs to be monitored or treated.

Changes in physical health are often only detected or suspected because of changes in general behaviour so it’s important to know what to be aware of and what to check!

System/organ	Effect	Monitor / Watch / Treat
Appetite regulation	Always wanting to eat / drink. Abnormal interest in food. Overeating.	Food and beverages must be limited . Overweight / obesity.
Behaviour	Often perceived and misunderstood by others to be mood or mental illness. Often occurs because of stress and lack of appropriate support.	Requires appropriate management strategies and rarely, medication, unless due to a diagnosed mental illness, which can occur in people with PWS.

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Bladder	<p>Poor emptying often seen.</p> <p>Obesity may increase urinary tract infections (UTI) and/or incontinence.</p>	<p>Timed toileting.</p> <p>Increase awareness of full bladder feeling / empty bladder feeling.</p> <p>Confusion may indicate UTI .</p>
Body fat - increased (with less muscle) even with healthy weight	<p>Overweight and obesity can lead to serious complications.</p>	<p>Energy intake must be limited.</p> <p>Exercise must be a part of life.</p> <p>Walking daily for 1 hour is simple and effective.</p>
Bone density (BD) (and bone strength)	<p>Often reduced due to reduced hormone levels, decreased muscle mass and too little physical activity.</p> <p>Low BD increases risk of fractures.</p>	<p>Maintain adequate calcium and Vitamin D intake and check blood levels.</p> <p>Check bone mineral density every 2 years.</p> <p>Helped by regular weight bearing exercise.</p>
Bowel	<p>Constipation is common and may lead to bowel or urinary incontinence, rectal picking and slow stomach emptying.</p>	<p>Maintain regular fluid intake.</p> <p>May require daily low dose laxative.</p> <p>Helped by exercising after eating.</p> <p>Prevent over-consumption of fibre.</p>
Diabetes Mellitus (DM)	<p>Often develops as a result of obesity but also seen in normal weight.</p> <p>Poorly controlled DM can cause undesirable weight (muscle) loss, renal failure and loss of vision.</p>	<p>Maintain a healthy weight and regular exercise to prevent DM.</p> <p>Check fasting blood glucose levels (BGL) every 6-12 months.</p> <p>If known DM exists check HbA1C (average BGL of past 6-8 weeks) every 3-6 months.</p>
Ears and hearing	<p>Lack of concentration or response may indicate the development of hearing loss, or psychoses.</p>	<p>Check with ageing as a person with PWS may not be able to express loss of hearing.</p>

		If acutely impaired check for infection first.
Eyes	Strabismus (abnormal alignment of eyes, squint); short/long sightedness.	From birth or a change in vision with age. Check eyes every 2 years .
Feet / legs	Can develop oedema and cellulitis then severe infections, when obese or inactive. Feet , hip, knee abnormalities (from birth) can worsen with weight gain and ageing.	Maintain a healthy weight. Maintain daily activity / exercise. Check regularly for sores and infections especially if swollen or poor circulation.
Lungs	Obesity, reduced breathing mechanics, scoliosis, and kyphosis can cause reduced oxygen consumption. Obesity and inactivity can lead to pneumonia, lung infections and respiratory failure. Asthma can also occur.	Maintain healthy weight. Maintain daily activity /exercise. Medical check is required if exercise tolerance is poor or deteriorates. Blood oxygen levels may need to be checked.
Mental illness	Depression, psychosis, mood disorders.	Require psychiatric assessment and may need treatment with medication. Due to brain dysfunctions in PWS dose-response and side-effect susceptibility are less predictable so lower initial doses are recommended.
Mouth	Reduced saliva causes dry, sticky mouth. Reflux from stomach can harm teeth (may need antacid medication).	Poor dental hygiene – dental erosion. Requires regular, effective cleaning and regular visits to a dentist.

<p>Muscle -</p> <p>Reduced strength and tone</p> <p>Reduced muscle mass</p>	<p>Weaker muscles, unstable joints.</p> <p>Scoliosis, kyphosis.</p> <p>Increased sensitivity to some medications.</p>	<p>Poor posture, slower mobilisation, reduced breathing mechanics (worse with obesity).</p> <p>Requires strengthening exercises.</p> <p>Dose of some medications, when newly prescribed, should be lower than usual.</p>
<p>Pain tolerance - high</p>	<p>Complaints of real pain are rare.</p> <p>Real pain is often indicated by change in behaviour or activity level.</p> <p>Check all swollen red areas of body if a fall or trauma has occurred.</p>	<p>Undetected illness or injury from accidents.</p> <p>All pain complaints must be investigated to exclude a cause and prompt diagnosis.</p> <p>Insist on x-rays /other investigations after trauma or appearance of ill health or pain.</p>
<p>Sequential processing</p>	<p>Poor personal hygiene leads to infections.</p>	<p>Encourage thorough washing of body.</p> <p>May require assistance or visual cues.</p>
<p>Sex hormones</p> <p>Are reduced</p>	<p>Lack of, or reduced, sexual maturity.</p> <p>Replacement required for bone health.</p>	<p>Yearly sex hormone (androgen) blood test from 15 years of age.</p> <p>Testosterone replacement (males - start with low dose)/oestrogen replacement - females), as required .</p>
<p>Skin picking</p>	<p>Sores and infections.</p>	<p>Cut fingernails every week, keep hands busy to distract from picking.</p>
<p>Sleep apnoea</p> <p>(pauses in breathing while sleeping)</p>	<p>Daytime sleepiness (occurs in PWS even without sleep apnoea), lack of concentration, excess irritability.</p> <p>Can worsen with weight gain.</p>	<p>To be assessed by sleep/respiratory specialist - may need positive airway machine or longer sleep time at night.</p>
<p>Stomach and Intestines</p> <p>Slow emptying of stomach and reduced</p>	<p>Distended stomach.</p>	<p>Complaints of “discomfort” from bloating and dilatation of stomach or loss of appetite.</p>

intestines passage time often occurs in people with PWS	Loss of appetite – no desire to eat (often only symptom – must investigate!). Vomiting, rarely seen in PWS, often indicates serious illness.	<i>Risk of gastric necrosis</i> – death of stomach tissue due to reduced blood flow to stomach wall. Requires urgent medical treatment.
Temperature (body) – poor regulation and sensation	Inappropriate clothing for weather. Can suffer from hypo or hyperthermia. Infections, but no fever.	Assist with choice of clothing for hot/cold weather. If generally unwell, CHECK FOR INFECTION with or WITHOUT raised body temperature / fever.
Water intoxication	Will lead to electrolyte imbalance which may cause seizures.	Do not allow unlimited fluid intake.
Yearly medical checks	May need to be insisted on by family members for their person with PWS.	Check weight, waist measurements, blood pressure, lung function, teeth, posture. Annual blood tests are recommended from the age of 15 years. Ask for: biochemistry (including calcium, cholesterol, glucose), iron studies (including iron), endocrine (including sex hormones, thyroid, Vitamin D).

Do not be afraid to seek a medical assessment for your person with PWS if you are concerned in any way about their health, due to changes in their manner or behaviour. Always provide information about PWS for the doctor or medical professional who is seeing your person with PWS, remembering that PWS is not well-known and includes specific traits that must be understood before treatment is prescribed. People with PWS need the ears, eyes and voice of the person with whom they live, to maintain good health and longevity!

This article was written by IPWSO's Famcare Board.

