

“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, 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 & 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.
Bladder	Poor emptying often seen Obesity may increase urinary tract infections (UTI) &/or incontinence	Timed toileting Increase awareness of full bladder feeling / empty bladder feeling Confusion may indicate UTI
Body fat - increased (with less muscle) even with healthy weight	Overweight and obesity can lead to serious complications	Energy intake must be limited Exercise must be a part of life Walking daily for 1 hour is simple & effective
Bone density (BD) (and bone strength)	Often reduced due to reduced hormone levels, decreased muscle mass and too little physical activity. Low BD increases risk of fractures.	Maintain adequate calcium & Vitamin D intake and check blood levels. Check bone mineral density every 2 years. Helped by regular weight bearing exercise.
Bowel	Constipation is common and may lead to bowel or urinary incontinence, rectal picking and slow stomach emptying	Maintain regular fluid intake. May require daily low dose laxative. Helped by exercising after eating. Prevent over-consumption of fibre.
Diabetes Mellitus (DM)	Often develops as a result of obesity but also seen in normal weight	Maintain a healthy weight & regular exercise to prevent DM.

	Poorly controlled DM can cause undesirable weight (muscle) loss, renal failure and loss of vision.	Check fasting blood glucose levels (BGL) every 6-12 months. If known DM exists check HbA1C (average BGL of past 6-8 weeks) every 3-6 months.
Ears and hearing	Lack of concentration or response may indicate the development of hearing loss, or psychoses	Check with ageing as person with PWS may not be able to express loss of hearing. 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 & 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.
Muscle - Reduced strength & tone Reduced muscle mass	Weaker muscles, unstable joints Scoliosis, kyphosis Increased sensitivity to some medications	Poor posture, slower mobilisation, reduced breathing mechanics (worse with obesity) Requires strengthening exercises. Dose of some medications, when newly prescribed, should be lower than usual
Pain tolerance - high	Complaints of real pain are rare Real pain is often indicated by change in behaviour or activity level Check all swollen red areas of body if a fall or trauma has occurred	Undetected illness or injury from accidents All pain complaints must be investigated to exclude a cause & prompt diagnosis Insist on x-rays /other investigations after trauma or appearance of ill health or pain.
Sequential processing	Poor personal hygiene → infections	Encourage thorough washing of body May require assistance or visual cues

Sex hormones Are reduced	Lack of, or reduced sexual maturity Replacement required for bone health	Yearly sex hormone (androgen) blood test from 15 years of age Testosterone replacement (males - start with low dose)/oestrogen replacement - females), as required
Skin picking	Sores & infections	Cut fingernails every week, keep hands busy to distract from picking.
Sleep apnoea (pauses in breathing while sleeping)	Daytime sleepiness (occurs in PWS even without sleep apnoea), lack of concentration, excess irritability Can worsen with weight gain	To be assessed by sleep/respiratory specialist - may need positive airway machine or longer sleep time at night.
Stomach & Intestines Slow emptying of stomach and reduced intestines passage time often occurs in people with PWS	Distended stomach Loss of appetite – no desire to eat (often only symptom – must investigate!) Vomiting, rarely seen in PWS, often indicates serious illness	Complaints of “discomfort” from bloating and dilatation of stomach or loss of appetite <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!