

General Health in PWS

The following information has been generously contributed by Dr Susanne Blichfeldt, Paediatrician and Scientific Adviser to the International PWS Organisation, and is taken from her powerpoint presentation. This will give a general idea as to the health indicators in a person with PW syndrome.

With no intervening treatment such as growth hormone, or sex hormone treatment, in general, after 18 years of age the mean height of women is 148 cm; men 156 cm, or approximately 14cm shorter than siblings; puberty is incomplete. Before age 30 they look younger: after age 30, they look older.

- This is due to sex steroid deficiency & GHT

What is more obvious:

- Many are overweight
- Many wear glasses
- Many have back and joint problems
- Some have problems with oedema (fluid retention) and skin (dry, picking, sensitive)

What is less obvious:

- Osteoporoses – up to 90% above age 30
- Diabetes – mostly Type II
- Heart and circulation problems
- Respiratory problems
- Stomach problems including constipation
- Sleep problems

What we need to know:

- Different reaction to some medications
- Different temperature sensitivity
- Temperature instability
- Different pain reaction
- Different sleep pattern
- Different behaviour and psychology
- Increased risk in psychiatric behaviours

Eyes and Teeth

- The need for glasses can come early
- Optician needs to be consulted every 2-3 years
- First teeth often have weak enamel. Second teeth can be strong.
- Sticky saliva means more brushing needed
- Dentist every 6 months

APPENDIX
GENERAL HEALTH IN PRADER-WILLI SYNDROME

The back:

- KYPHOSES (curvature of neck and spine) because of weak neck and back muscles
 - Exercises can prevent permanent kyphoses
- SCOLIOSIS ('S' curvature of spine)
 - Brace or operation needed

Legs and Feet

- Often problems with hips, knees, feet - giving pain and having water retention
 - Caused by overweight and weak muscles
- Feet: many walk on outer edges, some need special soles in shoes to prevent pain and worsening 'turn out'

The Skin

- Skin is 'weak' and bruises easily
- Skin is fair – increased risk of sunburn
- Skinpicking – something particularly 'PW'
 - Use bandaids, aloe vera, soft gloves, keep nails short
 - Keep hands busy!
 - Praise often for healed skin
 - Prevent infection

Osteoporosis

- Seen in 90% after age 30 (USA)
- Seen in 30% adults after age 18 (UK)
- Caused from lack of sex steroids and growth hormone
- Risk of fractures – often overlooked
- Special scanning can show osteoporosis
- Vitamin D and Calcium

Heart and Circulation

- Overweight is a disease risk factor
- Future GHT could be useful influence
- Circulation problems with Oedema

Respiratory

- Usually no lung disease, but can have small lung volumes
- GHT has good influence, with deeper breathing
- Apnoea –pauses in breathing – important to have this checked
- May need sleep mask (CPAP) to help breathing when asleep

Stomach and Intestines

- Possible weak intra-abdominal muscles
- Be aware of acute gastric dilation
 - Can happen from overeating
 - Particularly if person is on GHT, this can be fatal if unnoticed
 - Be aware of a dilated abdomen, vomiting, paleness, and refusal to eat

Other Intestinal Problems

- Gallstones
- Constipation
- Bleeding from rectum (picking?)
- Gastric banding for overweight – in PWS this is exceedingly dangerous with risk of bursting, internal toxicity, death

Diabetes

- More often Type II which comes with overweight
- Can be inherited if in family
- Treatment:
 - Medication, weight reduction and diet
 - Once weight reduced, medication can be lowered or ceased
- Untreated is a great health risk: eyes, kidneys, heart

Hormone Deficiency

- Sex steroids for men and women
- Growth hormone
- Based on individual evaluation
- Will help prevent brittle bones, and early aging
- Thyroid hormone deficiency

Medical Checklist

- For PWS, include checks for
 - Weight, blood pressure, respiratory and heart, oedema, skin, teeth, joints, back
 - Blood tests for hormones, diabetes
 - Dietician
 - Physiotherapist
 - Counsellor