

*Managing routine medical checks
and other regular requirements
in PWS*

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Presentation Susanne Blichfeldt

- Pediatrician, specializing in pediatric neurology
- Among many activities at job:
- Treatment of children and adults with PWS
- Two sons 44 and 42, youngest has PWS

Other PWS activities:

- Talks and meetings with staff in kindergardens, schools, homes for PWS
- PWS advisory board in DK: Since 1996:
- We are all kinds of professionals
- CSAB in IPWSO: publications and many contacts from many countries
- Former member of the IPWSO board and also the PPCB

The presentation to day

- Today in this talk I focus on the body in PWS
- What to know when you work with PWS
- What to know when securing the needed daily help
- To be aware of special abnormalities a person might have
 - perhaps not talked about or noted before
- To know what can give rise to serious situations

Food, diet and physical activity

- These are very important subjects. This is not the main subject for today
- I recommend the previous ECHO sessions for caregivers:
 - Constanze Lämmer : Nutrition and Diet in PWS
 - Georgina Loughnan : The Power of Exercise for People with PWS
- See [IPWSO Caregivers' ECHO® Resources - IPWSO](#)

PWS compared to other syndromes

- Congenital malformation of organs is rare.
- Children are born floppy after normal pregnancies. Parents are healthy.
- Symptoms in PWS are caused by lack of important genetic signals. (15q)
- All symptoms we see are related to dysfunctions in various areas of the brain and in the autonomic nervous system, which is the nervous system outside the brain that regulate functions in the organs.

So:

- The child is born with all the normal organs incl muscle cells, but the regulation of organ function from the nervous system is the problem and it affects:

Glands (hormones), muscles, intestines, sensation, temperature, cognition.

The brain is different in many ways in PWS

- Regulation of hunger and satiety (for the body the biggest problem)
- Temperature regulation and feeling, and pain sensation are often affected
- Motor development and functions are affected. Balance is poor
- Body perception is often not normal
- Scans of brain show when parts of the brain reacts differently to normal:
 - At sudden changes of plans: nerve cell activity in the brain is different to normal
- Also connections between nerve cells in the brain are different or less
- Autonomic nervous system is affected

The autonomic nerve system

- Works without your control and regulates many body functions.
- Sympathetic and parasympathetic: stimulating and inhibiting nerve signals
- Stomach and intestines
- Urinary tract
- Level of stress
- Blood pressure, and circulation in organs and skin
- Sleep
- Temperature control with diseases: fever-no fever
- Sensation of temperature: warm and cold

Medical, observations, service and diseases

Medical problems in PWS:

- Because of the syndrome itself. That we have to treat
- Diseases arising after years, when medical needs are not known, is to prevent
- Because of overweight (and PWS) also to prevent

Investigations of adults with PWS

A Dutch investigation of 115 adults with PWS (Pellikan et al 2020) showed

- 70 had one non diagnosed physical problem, possible to treat
- 28 had two undiagnosed problems
- 10: had 3 or more undiagnosed problems

- 26 lived in a specific PWS home (age 21-32 years and BMI 22-30 kg/m²)
- 36 in a non-specific PWS home (age 28-50 years and BMI 27-40 kg/m²)
- 19 with parents (age 19-22 years and BMI 26-36 kg/m²)

About the 115 Dutch adults with PWS

In general :

- The eldest and those not living in a specific PWS home had more diseases and also more undiagnosed physical problems

In PWS homes:

- There were more contacts to dietician, more plans for diet, and the adults there were in general more physically active.

Body composition in PWS and food

- Muscles and bones are slender.
- Fat layer is bigger than normal, also with normal weight.
- A DEXA scan of the body will show the body composition.

- Spontaneous muscle activity is often less than normal.

- The calorie needs are primarily based on muscle volume and activity.
- Result of all this: calorie needs are lower than normal.
- So, we recommend to calculate calorie needs based on cm height not weight and adjust after weekly weights

Hypothalamic dysfunction: causes hormone deficiencies

- Hypogonadism:
 - Means that the body does not “produce” sex steroids normally
 - Men: testosterone
 - Women: oestrogene.
 - Blood tests will show if supplements are needed from puberty age
- Hypothyroidism
 - When the thyroid gland produces too little hormone, and you become tired
 - Seen in ca 10% of adults (PWS) blood test will show if supplements are needed

Growth hormone (GH) and PWS

Since 2000:

- GH has been recommended for children with PWS in many countries. not all
- GH is important for growth and muscle development.
- GH does not change appetite. Cannot prevent overweight.
- Many children with PWS have been diagnosed with a total or partial GH deficiency and GH treatment has secured a better growth.
- There is a debate if GH treatment should be continued in adulthood and actually there are differences between countries

Diabetes mellitus in PWS

- Type II Diabetes mellitus- (DM)
- Comes with overweight
- Can be seen already in teenagers

Treatment:

- Weight loss, diet, tablets, in some cases insulin
- In many cases weight loss can be enough to cure
- DM can cause damage on eyes, kidneys, circulation and cause early death

Sleep in PWS

- Regulation of sleep can be abnormal
- Sleep apnea is common: central or obstructive.
- Can be treated with a face mask during sleep
- Daytime sleepiness can be because of disturbed sleep during night
- A sleep study can be a possibility.

Recommendation: ask the doctor if you want to know more

Temperature abnormalities

- Many can have even severe infections without fever.
- Often a reason for delayed treatment, even for sudden death.

Recommendation:

- Have documents ready for you doctor about this.
- Consider the person's attitude or condition: any changes
- Choosing clothes: When the person cannot feel warm or cold you must guide about the clothes

Calcium and vitamin D

- Vitamin D is important for the calcification of bones
- Vitamin D deficiency in PWS has been observed in many countries
- In the Dutch survey mentioned they noticed that treatment of Vitamin D deficiency generally gave a better health

Recommendation: measure D vitamin level in blood test and give supplements every day. 10-25 microgram per day

What more to be aware of in PWS

- Vision
- Hearing
- Mouth, teeth, chewing, swallowing
- Stomach and intestines. No vomits
- Urinary system
- Heart and lungs
- Back and legs and feet
- Skin
- Hygiene, body perception

Registration of symptoms and changes in PWS

- The responsible is the caregiver...
- Many with PWS do not feel changes in their body function.
- Or: they do not talk about, do not complain.
- And sometimes complaints are because of needs of attention.
- Life can be difficult for all.

Vision and hearing

- Many have a squint, many are operated on for that in childhood.
- Many need glasses.
- Vision can change also in young adults.
- Especially after the age of 40.

Recommended for all : Check vision every 2-3 years

- No research done about hearing
- Impression that hearing is often good.

Recommended: Check when there are speech problems, and after age 50 years

Mouth, teeth, chewing, swallowing

- Little saliva: dry mouth, teeth are not cleaned normally by the saliva .
- Teeth:
 - Food remains on the teeth after the meal.
 - Reflux of gastric acid is often not recognized, no complaints, harms the teeth
- Chewing:
 - Motor function can be poor, big pieces of food then swallowed
- Swallowing:
 - Can be incomplete, muscle function can be poor, food remains in the esophagus
 - All this means a risk of choking (especially if the person tries to eat quickly)

Dentist and eating situation

Dentist :

- Check recommended every ½ year, or more often
- Especially to check acid harm on teeth
- Some try to ruminate

Eating situation

- In general it can be recommended that people with PWS do not sit alone when eating, especially if chewing and swallowing is observed as not normal
- Choking is a well-known cause of death in PWS

Stomach and intestines

Stomach:

- often reflux to esophagus, unrecognized inflammation can be seen
- Stomach is easily distended, with overeating, with gastroenteritis
- Vomiting is difficult, never try to provoke this with medications
- A distended stomach can be “rescued” with a tube, relieves the pressure

- Often few complaints with a distended stomach,
- Only observation can be: no wish to eat:
- This demands immediate medical evaluation, often with ultrasound scan.








Intestines

- Slow passage time, often 1½-2 times slower than normal
- Constipation is therefore common in PWS but often not known by the caregivers/parents
- When referred to hospital because distended stomach: many are constipated
- Rectal picking: sometimes starts because of constipation.

Recommended:

- Be aware of time of toilet visits.
- See the stool if possible, and consider needs of laxatives

Bristol Stool Chart

Type 1		Separate hard lumps, like nuts (hard to pass)
Type 2		Sausage-shaped but lumpy
Type 3		Like a sausage but with cracks on the surface
Type 4		Like a sausage or snake, smooth and soft
Type 5		Soft blobs with clear-cut edges
Type 6		Fluffy pieces with ragged edges, a mushy stool
Type 7		Watery, no solid pieces. Entirely Liquid

Urinary system

- Most children become continent
- Incontinence can come with overweight and constipation

- Rare toilet visits to urinate, is often reported
- When asked they say they do not feel a full bladder
- Then the bladder becomes distended
- and when at toilet, up to more liters are “delivered”, in some slowly

Recommendation:

- have a plan for toilet visits, before each meal, before bedtime.

Heart and Lungs

- Problems comes with overweight:

Heart:

- Elevated blood pressure

Lungs:

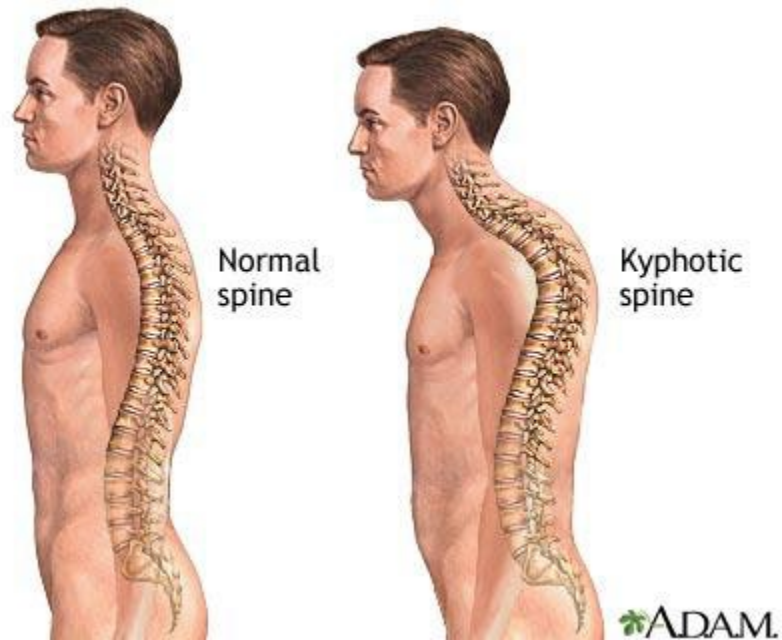
- With severe overweight: difficulties with breathing
- Edema in lungs

Bones

- Bones are slender.
 - Osteoporosis (fragile bones) is often seen in adults.
 - Especially with vitamin D deficiency and inactivity.
 - A DEXA scan will show.
-
- Bone fractures can be a risk.
 - Most often complaints are few when a fracture, because of high pain threshold.
- Recommended: always ask for X ray when a fracture is a possibility:
- Ex; does not use an extremity, swallowing, signs of blood, recent fall

Kyphoses and Scolioses in PWS

- Kyphoses



- Scolioses



Back Scolioses and Kyphoses

- Are because of PWS. Many are operated as teenagers,

Recommended

- Look for it: in the Dutch survey many with scoliosis were not diagnosed

Can give rise to:

- Muscle pain
- Inactivity
- Sleep problems
- In some cases, also adults are operated on

Feet and legs

- Swollen legs and edema: comes with overweight
- Treatment is weight loss and walking. Not diuretics
- Leg ulcers: take care that infections are prevented. Penicillin can be needed
- The feet: Many need help to cut toenails
- Shoes:
- Special soles or support needed?
- Often the case, and can help to a better motor function

Skin

- The skin can be fragile and often need extra care
- Skin picking can be a severe problem.
- Often arises when a person is bored or stressed
- Can be a habit that is difficult to stop

Recommended:

- For busy hands have something to tear up
- Avoid broken skin and scars

A few words about hygiene

- Many need fixed rules.
- Not all will understand the backgrounds. But can learn.
- Have a fixed plan for changing clothes, bath, time in bathroom.
- A timer can be needed in many situations.
- Many need help with hygiene: bath, amount of soap, washing, cutting nails, combing hair.
- But much can be learned. If not learned in parents' home, can take time.

Psychology in PWS

- Behavior and psychology is another important subject in PWS, and special care is needed when introducing routines, for taking care of the body.
- Has to be done gently and with careful knowledge of the persons understanding and cognition.

Advice about how you inform:

- Use illustrations to describe and be sure that information is understood.
- What is seen is remembered, only told is easily forgotten.
- Reasons for instructions are not always understood, but can be learned.
- Example is hygiene rules: like washing hands before meals.

Summary of recommendations

- Medical visits every ½ year
- Weight and blood pressure measured
- Evaluation of heart and lungs
- Evaluation of back, legs, feet, skin
- How is sleep?
- Blood tests every year or every 2 years for: Vitamin D, Thyroid hormones (T3, T4)
- Sex steroids if needed or treatment is given
- Individually evaluated for other tests, also if psychotropic medications are given

More recommendations

- Dentist every $\frac{1}{2}$ year
- Check of vision every 2-3 year
- Hearing test when needed, and after age 50
- Help given when needed: clothes, hygiene, cutting nails, etc.

Always remember www.ipwso.org

- [Guides for Doctors: Consensus Documents – IPWSO](#)
- Print the consensus documents and have it ready at all medical contacts
- [Information for Professional Caregivers – IPWSO.](#)
- And see you all in Ireland in July. Thank you.

Mikkel and Aslan. The dog is the biggest

