AGING IN PRADER-WILLI SYNDROME
Medical conditions and diseases to be treated
to avoid too early aging

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My wish and goal for this presentation

• To tell that early physical aging is not a rule for all with PWS

• To give information about medical facts and possible treatments (PWS.)

• To give information about how healthy adults with PWS can be

• How important awareness of the possible treatable symptoms is
What we know to day about PWS

• More and more adults are living healthy, happy with normal weight

• It is possible to have a long, happy and healthy life with PWS

  Special support is always needed, together with realistic expectations

• Family, caregivers and health professionals must know PWS:
• And know about diseases in PWS and react when specific help is needed.
And when you are old: PWS and all others

• Looking older
  Less muscle power.
  Balance can be weak. More weak than before (PWS)

• More risks for diseases.
• Need for help can change, when you are not able to do as before

• Mentally: you are in many aspects the same... and have more experience
• But it can be difficult to manage the changes that come with age
What we also know

• PWS is a very complex disease.
• Many physical problems and symptoms typical for PWS can arise.

• To day we know how to treat many of the well known medical problems in PWS

• It happens that the medical problems are not seen, and then not treated, which can cause early aging or even early death

• A recent Dutch investigation showed that among 115 adults 61% had one or more untreated health problems, and 25% had multiple untreated health problems
The 115 adults with PWS (Netherland)

- Seventy patients (61%) had undiagnosed health problems
- 1 in every 4 patients had multiple undiagnosed health problems simultaneously.
- All males and 93% of females had hypogonadism
- 74% had scoliosis, 18% had hypertension,
- 19% had hypercholesterolemia,
- 17% had type 2 diabetes mellitus,
- 17% had hypothyroidism.
- Unfavorable lifestyles were common: 22% exercised too little
- 37% did not see a dietitian
Diseases in PWS

- Symptoms can be different in PWS compared to others
- The symptoms seen can be different than expected with diseases
- Ex:
  - Less pain with fractures, and abdominal diseases, no fever with infections
- People with PWS cannot always explain their symptoms when they are ill
- So diseases can be overlooked
- Means: a risk that diseases are diagnosed late, or even too late
What is sometimes wrongly expected

• Adults with PWS cannot survive healthy until “old age”

• Early death is to be expected, as it is for many congenital syndromes

• People with intellectual disabilities like PWS will develop dementia

• Why:
  • We do not have any published data on healthy aging in PWS
  • No systematic information about the medical conditions of those who died
Causes of death most often referred in adults with PWS

- **Overweight**
- **Respiratory problems:** lung infections not treated, apnea
- **Abdominal diseases:** infections and stomach rupture
- **Accidents:** also in traffic
- **Choking.** Breathing is blocked by food in the throat.
- **Sudden unexpected death**, and often no reason found and described
More research is needed

- The body and the brain in PWS:
  - Is aging different? also when correct medical treatment is given?

- Persons with PWS are different individuals. And their social conditions and support varies a lot. This makes it difficult to do research

- So now we must rely on:
  - the knowledge we have about symptoms we see, and give correct treatment
  - and avoid early aging because of overlooked medical condition
So what to be aware of: Weight....

- **Weight**: Overweight can result in diseases in PWS:
  
- 1. Diabetes that causes:
  - circulation problems, edema, heart, kidney, eye diseases

- 2. Joint problems, difficult to walk
- 3. Breathing (lung) problems, low oxygen, poor general condition
- 5. Sleep problems with apnea, daytime sleeping
- 6. Bladder control and intestinal problems, obstipation
- 7. Infections, cancer
And how to treat overweight in PWS

- Gradual weight loss based on “correct food” and moving
- Supported by a dietician who knows (or can learn) about PWS.
- To secure a correct diet plan: protein, carb, fat, vitamin D.
- And having a plan for structure of administration of food

- Daily program for moving:
  - walking is possible for all.
  - other activities that demands physical activity, plays etc
Hormonal diseases caused by hypothalamic dysfunction PWS

- Growth hormone deficiency (many adults need treatment?)
- Hypogonadism (all men, most women)
- Hypothyreodism (15%-20%)
- Adrenal insufficiency (rare)
Growth hormone (GH) deficiency in PWS

- Many young adults have been treated with growth hormone during childhood; some have stopped at age 18, some are still treated.
- GH is important for muscles and organs, also for adults. Does not affect appetite.
- There are different rules about GH treatment for adults in countries.
- In some countries, some special tests are needed before start.
- We know that adults (not PWS) with GH deficiency can have medical problems and earlier aging and earlier death if untreated is seen.
- GH is given as daily injections, and the dose is adjusted after regular blood tests.
Hypogonadism (low testosterone) in men

- Small genitals since birth and no normal maturation of genitals in puberty:
- Insufficient testosterone in blood (produced in the testicles): results in:
  - No growth spurt in puberty
  - No normal muscle growth in puberty, smaller than normal muscles in adults
  - High pitched voice as adults
  - Small genitals as adults, little body hair
  - Looks young in the first adult years, but early aging, looks old, seen around age 30
  - Osteoporosis will arise, risk of fractures
  - Anemia can arise

  Early aging and weak muscles are seen in men with low testosterone (hypogonadism)
Testosterone treatment in PWS

- In puberty or later: blood tests will show if treatment is needed
- An individual assessment if treatment is to be given

- Can be given as daily gel(cream) on the skin
- Or as injections every 3 months

- Recommendation: start with low dose and follow with blood tests

- Testosterone deficiency in men was seen in all of the Dutch cases, many were not treated, and in 50% it was not diagnosed before
Hypogonadism in women, low estrogen

• Lack of female hormones (estrogen and progesterone) result in:
  • No growth spurt in puberty.
  • Not normal breast development.
  • No or late or irregular menstruation periods.
• Early aging,
• Early osteoporosis

• Caution: a few women will have menstruations, often late and irregular and few
  (if no treatment is given)
• all women (PWS) are in theory at risk of becoming pregnant.
Treatment of women with hypogonadisme

• Blood tests can be done to evaluate the degree of hypogonadism

• Estrogen supplement must be considered in women with no / few menstruations

• An individual assessment if medical treatment is needed,

• Evaluate possible side effects.

• Hormone treatment can be given as P pills

• Important to know and tell the possible risk of pregnancy

• Gynecologist contact is to be recommended for all women with PWS
Hypothyroidism

- Hypothyroidism is seen in around 15% of persons with PWS.
- The hormone is normally produced in thyroid gland.
- Hypothyroidism can arise in childhood or later
- Hypothyroidism can result in
  - Less muscle power.
  - Loosing weight (muscles)
  - Feeling cold
  - Slow heart rate
  - Being slow, no energy,
  - Early aging, physically and mentally
Hypothyroidism diagnoses and treatment

- Regular blood tests for hypothyroidism, measuring hormones in blood

- Blood test will show if hormone treatment is needed.

- Treatment is easy:
  - a daily tablet, taken ½ hour before breakfast

- Regular blood test is needed to regulate the tablet doses
ADRENAL INSUFFICIENCY

- Adrenal insufficiency in PWS seems to be rare.

- Adrenal hormones are important for the body to react normally when we are ill, having severe infections

- It is recommended to measure adrenal hormone cortisol with blood tests in acute situations with severe physical illness, to know if supplement is needed.
Autonomic dysfunction

• Our inner nervous system. We cannot control-
• Works by itself: Sympatic and parasympatic nerve system.

• Stimulates and regulates.:  
• Bowel and urinary system  
• Blood pressure  
• Sweat, blood circulation in organs and skin  
• Sleep  
  Temperature with diseases: fever or not  
• Nervous Vagus is important for the autonomic nerve system
More Symptoms to be aware of in PWS

IMPORTANT for all ages also when aging
Chewing and swallowing and teeth

- Mouth pharynx and upper esophagus: muscles are weak:
- Some are not chewing the food sufficiently: risk of choking
- Food is often not swallowed normally: Saliva is sticky
- After a meal some has food in mouth, pharynx and upper esophagus
- Big pieces: risk of choking
- Advice: drink ½ glass of water when the meal is finished:
- Teeth: gastro-intestinal reflux is common in PWS: Teeth can be acid-destroyed
Constipation

- One of the most common problems in adults with PWS

- Because of slow intestinal passage, which is the case for many adults, the result can be severe constipation, sometimes so extreme, so it causes vomiting.

- The cause is slow movement of muscles in the intestines.
  - Laxantia can help, and the dietician can prescribe the best diet.

- Constipation can affect mood, alertness and general wellbeing
# Bristol Stool Chart

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

- Sleep investigations have shown that sleep pattern can differ from normal (the different sleep phases, deep sleep and dream phases)
- Sleep apnea (breath holding during sleep) can be central or obstructive:
  - Central sleep apnea: abnormal regulation from the brain
  - Obstructive sleep apnea: caused by enlarged tonsils or adenoids.
- If snoring or sleep apnea is noted: sleep investigation is recommended and
- Evaluation by an ear-nose-throat doctor
- Some will need CPAP treatment.
- Narcolepsy and kataplexia: are rare (sudden sleep attacks)
Edema and skin infections in PWS

- Leg edema is often seen, also with normal weight:
- Caused by insufficient lymph drainage, poor circulation:
- Treatment: supporting stockings, walking, avoid long hours of sitting
- Diuretics are only indicated for leg edema caused by heart or lung problems
- Risk of skin infection with swollen legs, and potentially life threatening

- With severe overweight edema can be seen in the whole body and lungs
- Diuretics given if the heart and lung problems
- Treatment: weight loss and walking, moving
Not to forget the back and the muscles

• Many with PWS have scoliosis and/or kyphoses.

• Children are often treated with bracing or operation in teenage years.

• Many of the adults today are not treated as children and can have back problems that causes, with age, more and more stiffness and pain:

• Treatment:
  • contact to physiotherapist that prescribe exercises to done daily at home, supervised by staff!
Kyphoses und Scoliose

- Kyphoses
- Scolioses
Psychiatric diseases

• Can arise in all
• Can arise in all ages

• Be aware of changes in mood and behavior
• Depression can be seen: symptoms: loss of energy
• Dementia: symptoms: loss of skills..... Important difference to depression
• Psychoses can arise
• Psychiatric diseases can be treated with success, must be diagnosed
• Medication must be started with low dose, to avoid side effects
What to do:
Yearly medical visits and blood tests

- Secure regular medical evaluation and treatment when needed.
- Yearly evaluation of the diet, must be individualized
- **Blood tests yearly for:**
  - Blood sugar level (diabetes?)
  - Hormones: testosterone (men) estrogen (women), thyroids (both)
  - Vitamin D, calcium
- **Other investigations**
  - Sleep studies
  - Growth hormone needed?
  - DEXA scan if obese in some cases
And not to forget

- Dentist every ½ year, or more often if needed

- Vision:
  - need of glasses, test every 2-3 year after age 40

- Hearing:
  - test hearing after age 40 and then every 2-3 year, especially if there is a family history of hearing problems
Be prepared:

- Have the one page with you, in all languages:
  Important Medical Facts about Prader-Willi Syndrome - IPWSO

- Insist for medical evaluation:
  - When no wish to eat, vomiting
  - Being ill, is behaving differently.
  - Swollen bones, bleeding: fractures: X ray?
Always remember

• Write a yearly report: print photos

• What has happened during the year.
• How was the physical and mental function
• Family contact?
• Any loss of skills, any diseases

• What did you enjoy? What do you want to do?