

AGEING IN PRADER-WILLI SYNDROME

What do we know. What can we do

to avoid too early aging

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My wish, hope and goal for to day

- To tell that early physical ageing is not a rule for all with PWS
 - To give information about medical facts and possible treatments (PWS.)
 - To avoid too early ageing ,because of lack of knowledge about medical needs
 - To secure a focus always on individual needs, also with changes and ageing
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- We still need to learn more, more research is needed about PWS and ageing
 - We must avoid false expectations, that can arise without documentation
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- And you to day are the experts, so we look forward to learn from each other

What we know to day about PWS

- It is possible to have a long ,happy and healthy life with PWS
- More and more adults are living healthy , happy with normal weight

Special support is always needed, and individual realistic expectations

- Family, caregivers and health professionals must know PWS:
- And know about diseases in PWS and react when specific help is needed.

Adults to day with PWS becomming older

- They are different
 - They have very different backgrounds
 - Some have been healthy without diseases, for many years
 - Some have had several illnesses and perhaps still have.
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- Upbringing, family support before and now , varies a lot
 - Childhood possibilities, school, social contacts and acceptance vary
 - Actual situation: illnesses, living situation, occupation ?
 - All this will influence on the ageing process

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 (wrong!)
- We need published data on healthy ageing in PWS.
- But we know adults with PWS who are healthy and now “older”
- We have case reports, about people with diseases and dementia
- We need more information about the medical conditions of those who died



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

Diseases in PWS- It can be difficult

- Symptoms can be different and unexpected in PWS compared to others
- Less pain with fractures, and abdominal diseases, no fever with infections
- People with PWS can not always explain their symptoms when if they are ill

A great risk that diseases are overlooked and diagnosed late, or even too late

Many know cases with too late diagnoses, too late medical intervention

What we know about PWS

- 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)

The journal of Clinical Endocrinology & Metabolism, 2020, Vol. 105, No. 12, e4671–e4687(Pellikan et al)

- 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

CAUSES OF DEATHS IN 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.
- But often no information about the actual health status of those above who died
- Sudden unexpected death, and often no reason found and described

More research is needed

- Only one brain study:
- A MRI study finds that the brain ages in adults with PWS is advanced for age, but the participants in the study had different background and medical status.
- So is ageing different ? , also when correct medical treatment is given ?
- It makes research difficult when adults with PWS have different backgrounds
- Daily life:
- Individual evaluation and correct treatment in acute situations and daily
- To avoid too early ageing because of diseases

So what to be aware of: Weight....

- Weight : Overweight can result in diseases in PWS:
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- 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 (follow screenings for 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
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- 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 hypogonadism

- 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
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- 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

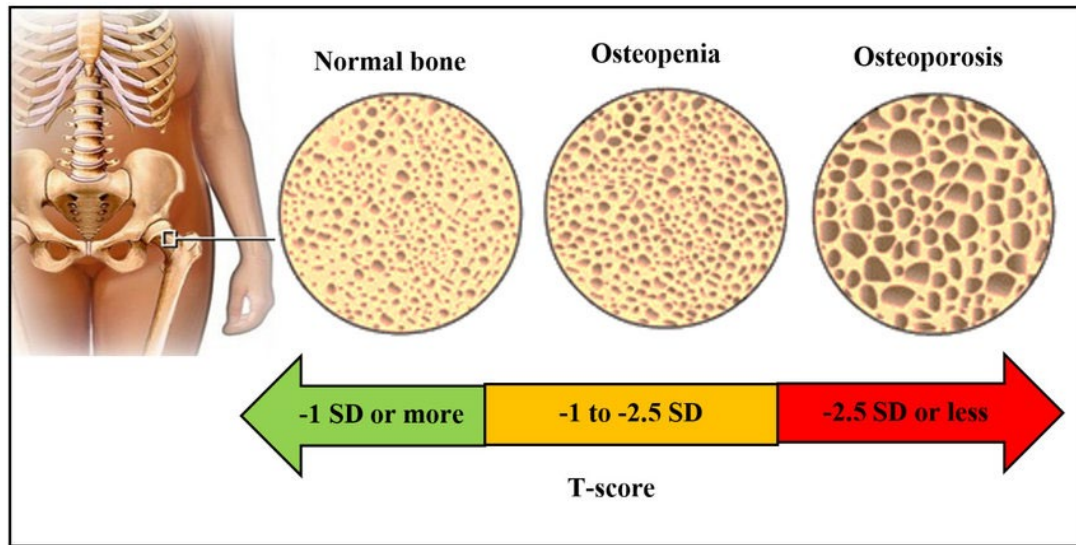
Hypothyroidism diagnoses and treatment

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

Bones and osteoporoses.



- Osteoporoses often seen with age in PWS
- Causes: hypogonadism, reduced activity, (a family thing)
- Both men and women in PWS
- Risk. :
- Bone fractures. Vertebra. hips
- DEXA scan recommended for all to evaluate bone structure

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
- 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

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

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(sudden sleep attacks)

Sleep disorders in 60 adults with PWS

Sleep. 2017 Ghergan et al

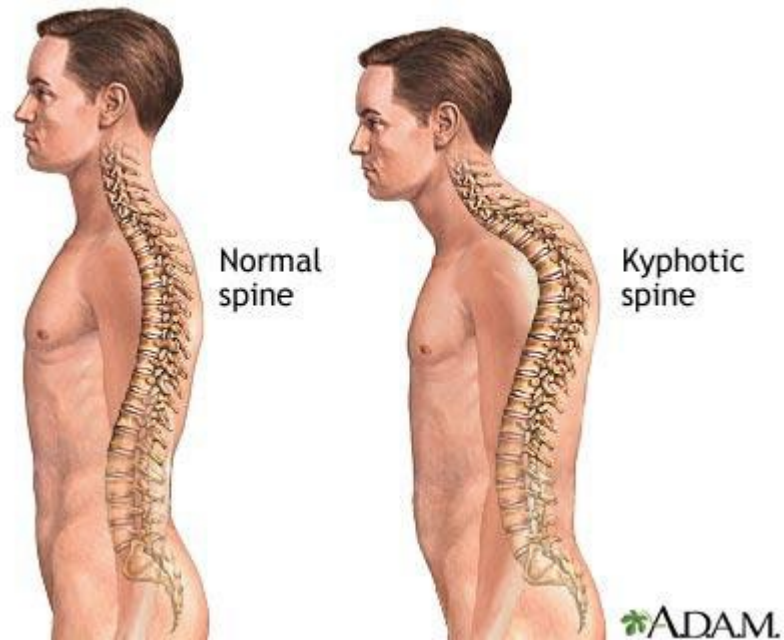
- Many adults are reported to be more sleepy than others
- A detailed investigation about sleep and sleep disorders:
- 60 adults with PWS , men and women 15-35 years old
- Most were overweight and obese
- 67% excessive sleepiness: some with narcolepsy (sudden sleep)
- 15% had sleep breathing disorders
- 14 had obstructive sleep apnea
- Some will benefit from CPAP or special medication for Narcolepsy

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 to day are not treated as children and can have back problems that causes, with age, more and more stiffnes and pain:
- Treatment:
- contact to physiotherapist that prescribe excercises to be done daily at home, superwised by staff !

Kyphoses und Scoliose

- Kyphoses



- Scolioses



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
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- With severe overweight edema can be seen in the whole body and lungs
 - Diuretics given if there are heart and lung problems
 - Treatment: weight loss and walking, moving

Behaviour , difficulties

- Arise with changes : family relations or loss , changing of staff , and more
- Misunderstandings
- Too high expectations from family or staff.
- Grief can come late
- Difficult behaviour demands personal support, often extra support
- Difficult behaviour is not a wish from the one with PWS.
- Difficult behaviour is painful for the one with PWS
- Difficult behaviour can not be cured with medication alone
- Analyse: when and why it happened, which changes were there

Psychiatric diseases

- Can arise in all. Can arise in all ages
- Be aware of changes in mood and behavior
- Can be because of physical illness or because of psychiatric diseases
- 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 medication, must be diagnosed
- Medication must be started with low dose, to avoid side effects

DEMENTIA?

Definition: Changes that are persistent and interfere with activities of daily living and social interaction in:

- Memory- impaired or loss
- Cognition- loss or reduced abilities in doing tasks that they are normally capable of doing.
- Personality- change from the usual

DEPRESSION?

Definition: A mood disorder that causes persistent feelings of sadness or loss of interest

- Memory- may be impaired
- Cognition- slow thinking but typically are still able to perform tasks correctly
- Personality- may see low energy, sadness or fluctuation in mood – anger, anxiety

PWS Aging Signs Checklist for Residential Care Providers

Monthly / Quarterly Observation Tool

Cognitive & Mental Health Changes

Increased confusion or forgetfulness		Neuropsych eval	
Decline in problem-solving or planning		Track cognitive tasks	
Changes in language or word finding		Speech therapy referral	
New or worsening anxiety or fears		Psychiatry consult	
Withdrawal from preferred activities		Behavioral health	
New or worsening OCD behaviors		Medication/CBT review	

Mobility & Physical Function

Decrease in walking speed or endurance		PT referral	
Increased falls or unsteadiness		Fall risk assessment	
Loss of muscle tone/strength		Review GH, PT plan	
Difficulty with stairs, rising from chairs		Functional mobility plan	
Worsening scoliosis/postural changes		Ortho review	

Cardiovascular and Metabolic Health

Fatigue or shortness of breath		Cardiology workup	
Swelling in legs or ankles		Check cardiac/kidney	
New/worsening hypertension		Medication review	
Changes in cholesterol/glucose		Lab monitoring	
Unexplained weight loss		Nutritionist referral	

Bone Health & Endocrine Signs

Frequent fractures or bone pain		DEXA scan	
Shortened height/spine changes		Endocrine consult	
Loss of GH therapy benefits		GH dosing review	
Signs of menopause/andropause		Hormone panel	
GI slowdown or constipation		GI consult/diet review	

Sleep & Breathing

Increased daytime sleepiness		Sleep study	
Snoring or apnea symptoms		BiPAP/CPAP review	
Resistance to sleep equipment		Re-education/device check	
Frequent waking during the night		Sleep hygiene adjustment	
Morning grogginess		Adjust sleep schedule	

Sensory & Neurological Health

Vision changes		Eye exam	
Hearing issues		Audiology referral	
New tremors/coordination issues		Neuro consult	
Headaches, dizziness, seizures		Urgent medical eval	

Social, Emotional, and Behavioral

Reduced social interaction		Increase engagement	
More rigid/resistant to change		Behavioral support	
Regression in self-care		OT reassessment	
Increased emotional outbursts		Medication/therapy review	
Paranoia or psychosis symptoms		Urgent psych referral	

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
 - DEXA scan if obese in some cases

And not to forget

- Dentist every $\frac{1}{2}$ 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

In difficult situations

- Insist for medical evaluation :
- When no wish to eat, vomiting
- Being ill, is behavior suddenly different. ?
- Swollen bones: fractures ? : insist for X ray
- Have the one page with you, in all languages:

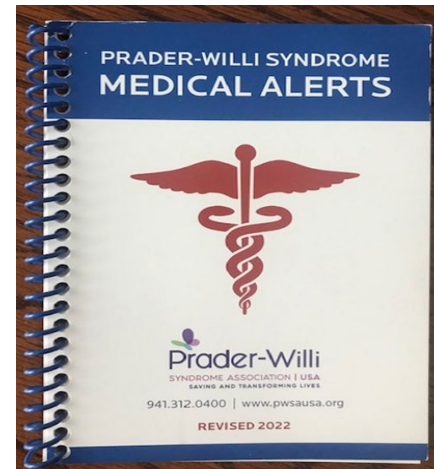
[Important Medical Facts about Prader-Willi Syndrome - IPWSO](#)

Have written information ready

www.ipwso.org

- The one page for acute situations
- [Important Medical Facts about Prader-Willi Syndrome – IPWSO](#)

- Alerts booklets: in many languages, bring when you travel
- [Medical Alerts Booklet – IPWSO](#)



- www.ipwso.org:
- Here you can have the documents translated into to your own language

Always remember

- Write a yearly report: the year book with printed 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 ?





