

HEALTH CARE PROBLEMS PRADER-WILLI SYNDROME

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PRADER-WILLI SYNDROME

- A congenital disorder of chromosome 15
- Many signs and symptoms are seen
- Research is still needed to explain the exact genetic reason for many of the symptoms .
- We know about many special symptoms, how to be aware, treat and manage PWS

MEDICAL PROBLEMS IN PWS

- Special signs and physical reactions with illness
- Special symptoms related to the organs in the body
- Special problems can arise related to overweight
- Special behaviour, characteristic, and problematic
- Symptoms of psychiatric diseases can arise

HYPOTHALAMIC REGULATION PROBLEMS:

- Appetite regulation. Always a desire to eat.
- Temperature: regulation and sensation affected
- Sensation of pain is different
- Hormone regulation is most often disturbed
- Sleep and awake rhythm can be disturbed

PWS : Always a desire to eat

- Children after age 1-2 years and adults :
- Eat when offered food, eat at all meals
- Eat when food is available, seek food
- Also when/if there is a feeling of being “full”
- Often ask about next meal, talk about food
- Think about food, perhaps not telling us
- *A drive to gather calories*
- *A behaviour as if he-she is starving*

Ved tidlig diagnostisk forøges bl.a.
chancerne for at undgå overvægt



Why is weight gain so rapid

- Eat too much... yes
- Caloric need is lower than normal.
- Normal amount of food for a child is too much.
- The *lean body mass* is often small:
- the fat mass is often 50% of the body weight:
- Move less: often seek the chair, hard to move ?
- Adults move less than in other syndromes

Risks when eating in PWS

Eat too fast. some eat with open mouth

Do not swallow before next spoonful is taken

Many do not swallow normally

Risk of choking.

Risk of having too much in the mouth

so he/ she cannot breath

Rule for PWS : never sit alone when eating

Eating and PWS: how to avoid overweight

- Supervision and regulation of food first of all
- No one needs to come into the kitchen !
- Serve the food (like in the restaurant)
- Leave the table and dining room when the meal is eaten, and start another activity. Also at parties
- Avoid: cleaning table and washing plates
- Discussions about food will always end badly

FOOD

- Low fat diet is recommended: volume can be bigger.
- Individualized portions. No special diets,
- Body size and physical activity set the needs
- Secure calcium ,minerals and vitamins incl vit D
- Contact to the dietician is needed
- The food must look good. Taste good.
- Know the amount of calories of the food
- Even breads can be very different in calories
- Sugar free products may contain a lot of fat

ALSO about FOOD

- Do not introduce what cannot be continued
- Food security:
- no doubt
- no hope
- no disappointment

WEIGHT IN PWS

- What is the ideal weight or BMI ? (kg/ m x m)
- Or lean body mass? (the body without the fat)
- When is a person overweight ?
- ***If a persons physical activity is limited because of overweight, the weight is much too high***
- A child should be able to move according to his/her motor abilities

- ***It is also normal for adults to be able to run***

Physical activities that help

- Children/ Adults (PWS) often move too little
- Because of hypotonia, small muscles:
- less drive : this can *worsen the overweight*

- Need activation, motor activities
- **we** have to start activities:

- It is the daily activity that counts !

- Walking: min. ½ hour per day more is better
- Take stairs . Stop the car 5 min walk from home
- Daily exercise program for back and joints
- 5-10 min DAILY:
- ALL can do it. Decide the hour
- Help and supervision are needed
- Because of the poor body image /understanding

SPORT

- Dance
- Swimming
- Horse back riding

- Nordic walking
- Fitness center... use music
- The helper/ caregiver participate !!
- Some enjoy group activities, but not all

TEMPERATURE AND PAIN AND PWS

- High body temperature without disease : can be seen
- Low body temperature: can be seen: ex. with depression.
- **Infections without fever**: not rare in PWS (also severe cases)
- **Sensation**: Do not feel warm or cold as we do.
- Many need help to dress.(which clothes when)
- **Pain**: high pain threshold: often only few complaints:
- examples are: bone fractures and abdominal diseases
- **The doctors need information about PWS. MEDICAL ALERT BOOK**
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TO INTERPRET SYMPTOMS IN PWS

- Is he or she ill or not ill ?:
- We cannot rely on temperature or pain complaints
- We cannot expect the same reactions as seen in others

- We have to consider the general well being.
- listen to the person with PWS
- Any changes? *What has happened ?*

- The appetite , alertness and behaviour: changes ?

HORMONES and PWS

- Growth hormone : deficiency (not all)
- Treatment of children is given. Adults ? (not all need GHT)
- Low levels of sex steroids –teenagers and adults, both sexes:
- Treatment from puberty and thereafter, individualised
- Low levels of thyroid hormones: not rare
- If: treatment is given, rarely side effects
- Cortisol: seems to be normal in most cases, conclusion:
Check/treat with severe infections- treat when needed

HYPOGONADISM,PUBERTY AND SEXUALITY

- Hypogonadism: small genitals, low hormone levels
- Puberty in PWS:
- Often early andrenarche: pubic hair.,
- Often delayed and incomplete puberty. Varies a lot.
- Treatment : based on individual evaluation
- Sexuality many adults have “childish” interests.
Plays with dolls. Great risk of “sex for food”.
- Education about prevention is needed

DIABETES IN ADULTS WITH PWS

- Increased risk, also with normal weight.
- But especially with overweight
- Regular check is needed: morning blood tests
- Treatment:
 - diet, medication some need insulin.
 - Many can be treated with diet alone:
 - Rule: weight loss important to avoid diabetes.

Osteoporoses (fragile bones) in PWS

- Often seen in adults
- DEXA scan will tell
- Osteoporoses : too little calcium in the bones
- Risks:
- Untreated hypogonadism
- Growth hormone deficiency
- Vitamin D deficiency (daily tablets needed)
- Too little movement

SLEEP AND SEIZURES

- Sleep apnoea: some need treatment : CPAP
- Daytime sleeping? How is the night sleep?
- Falling asleep when sitting : is not epilepsy

- Epilepsy
- can be seen in PWS, but not often.
- can be treated
- Severe long convulsions never seen in PWS

BODY COMPOSITION and MEDICATION IN PWS. : HOW MUCH TO GIVE

- The *lean body mass* is most often small:
- the fat mass is often 50% of the body weight:
- The medication is *distributed in a very small body*

- Side effects can be seen if the dose is too high
- Often best to start with low or half dose concerning :
 - *Antihistamines
 - * Medication for epilepsy
 - *Medication for depression and anxiety

ANÆSTHESIA AND PWS

- To day: most often without problems
- Problems: especially because of overweight
- Apnoea during awakening can be seen
- Awakening can be delayed, also in lean persons
- A full stomach before anaesthesia gives problems:
 - food runs back into the lungs.
- *The doctors must know about PWS and anaesthesia*

ORGAN RELATED SYMPTOMS IN PWS

- **Eyes**
- **Hearing**
- **Mouth and teeth**
- **Heart and lungs**
- **Stomach, intestines and bladder**
- **Legs and feet**
- **Spine**
- **Skin**

EYES IN PWS

- Strabismus,,: squint: many are operated
- Short-sightedness
- Long-sightedness
- Need of glasses ?
- Change of vision over years:
- Ophthalmologist every 2-3 years for all
- Remember especially to check after age 40

HEARING IN PWS

- Hearing in PWS : we have very little literature
- Supposed to be normal ??
- When did you check ?
-
- When to test:
- *Especially when speech is delayed:*
- *But also if never before been checked ?*
- In the elderly check every 2-3 years (above 40)

HEART AND LUNGS

- HEART: Congenital heart diseases are rare in PWS
- Problems arise after years with severe overweight
- LUNG: Ventilation becomes restricted with severe overweight or with severe scoliosis and kyphoses.

Persons with PWS may have allergy and asthma like many others: if so, they will need treatment for that

Lung infections

- A severe risk in PWS. Can be fatal

Small children:

not able to cough

perhaps no fever

Adults

Symptoms can be overlooked:

Perhaps no fever, no complaints, breathing changed ?

Only symptom registered: do not wish to eat

MOUTH AND TEETH

- Little saliva:
- Dry mouth
- Teeth need cleaning after meals

- **Reflux** and Regurgitation
- The acid from stomach reflux , harms the teeth.
- Coca cola destroys the teeth

- Dentist at least every 6 month

THE STOMACH AND PWS

- The stomach easily becomes distended when full
- Gastroparesis: when the stomach does not empty
- Vomiting is rare in PWS: difficult to “empty”
- Some may eat hair: makes a “ball” in the stomach

- A big risk for gastric rupture: after binge eating
- The pain is often little and complaints are few !!
- **But** THE PERSON WITH PWS DO NOT WANT TO EAT

GASTROPARESIS

- We do not know all about all cases:
- Can occur:
- After binge eating, with gastroenteritis
- constipation
- can arise with other diseases as well ?
- Just when he or she is ill ?

- *Often a very bad smell*
- All Danish cases seen the last years had severe constipation.

Treatment of distended abdomen

- Ultrasound scan
- CT scan or X ray picture (free air?)
- Check urine

- Tube in the stomach to get "the air out"
- Treat constipation
- Treat infections
- Operation ?








INTESTINES and PWS

- TRANSIT TIME: Delayed: 24% of adults: 2- 3 days
- CONSTPATION: 40%, often not recognized.
- Complaints might be few . most often easy to treat

- Can be the reason for both rectal and urinary incontinence and rectal picking.

- RECTAL PICKING ?: Limit the time alone in toilet

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

BLADDER & URINARY SYSTEM

- Most children achieve “control”, but often delayed
- Some do not empty the full bladder when at toilet
- Enuresis (bed wetting): be aware of evening drinking!
- Incontinence: with overweight? with age? Do not feel ?
- Urinary infections: with poor hygiene and overweight

- Treatment:
- Toilet visits at fixed hours.
- Empty the bladder. Learn to use the time for it !

WATER INTOXICATION IN PWS

- Can happen with medical treatment for enuresis.
- If too much water is drunken before going to bed.
- The water stay in the body and disturb the salt balance
- Can lead to seizures

THE SPINE AND PWS

- KYPHOSES : C form: over bent
- Often seen, caused by muscle hypotonia
- Worsens with age

Daily exercises prevent permanent fixed “bending”

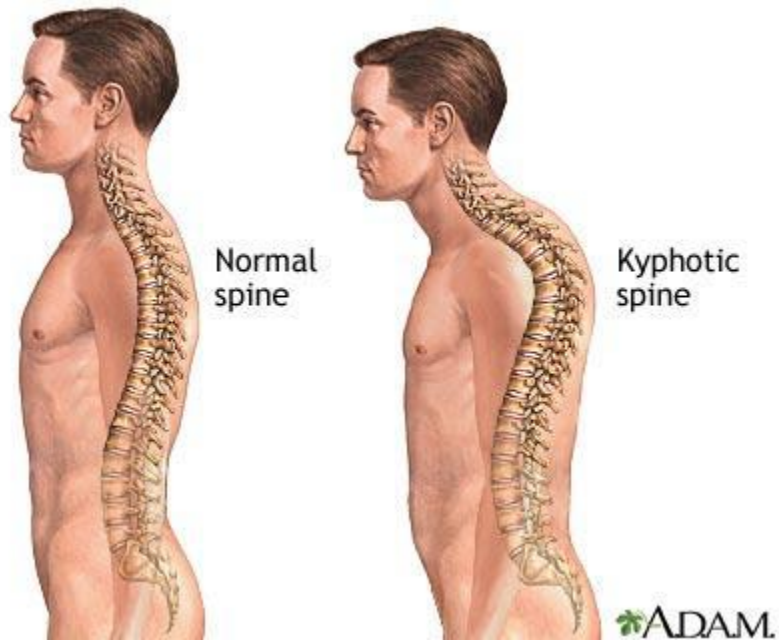
Severe back problems can compromise breathing

- SCOLIOSES: S form . Asymmetry of back.
- Often seen. Careful examinations needed
- Brace or operation ? Progress stops after puberty

Kyphoses and scolioses in PWS

- Kyphoses

- Scolioses



FEET AND LEGS IN PWS

- FEET: Many need shoes with extra support to the feet:
- special soles and “bracing “ shoes.
- JOINTS: Hips, knees, feet: overweight is harmful
- BROKEN LEGS: not rare, can be overlooked (no pain)
- OSTEOPOROSSES: untreated hormone deficiency:
- risk for fractures (both sexes)
- OEDEMA:
- Caused by overweight and too little physical activity.
- Poor circulation and wounds : give *skin infections*

THE SKIN AND NAILS IN PWS

- The skin can be very sensitive to the sun
- Sun exposure: Remember a shirt, and a hat,
- Vitamin- D from the sun : yes, supplement needed
- Nails:
- biting nails ? need for help to cut the nails ?

SKIN PICKING IN PWS

- Can be a very big problem in PWS
- The reason is unknown. Often a habit
- But: Can be associated to stress and boring.

- The persons seems to become absorbed in the picking
- Sometimes even not aware they are picking ?
- Treatment:
- keep the skin smooth, plaster on wounds,
- Occupation of hands and rewards for healed skin .
- Be there, and change activity

Hygiene in PWS

- Often a very big problem. A tabu. Ask parents
- Especially if no rules has been set early.

Many are not able to wash themselves correctly

Do not understand why we are changing clothes

many create own rules, create many conflicts

Best from early age to have rules for:

Bath, soap, water use, clothes,

Tiding , cleaning your room. etc

Behaviour and Psychiatry

- Behavioural problems and psychiatric symptoms are often misunderstood
- and then not correctly treated .

- Behaviour is often seen as psychiatric symptoms
- Or:
- Psychiatric symptoms are seen as behavioural problems

BEHAVIOUR

- Behavioural problems arise because of situations in the environment that are too difficult for the person with PWS.
- Medication can sometimes help to relax and cope.
- As a rule:
- behavioural problems can not be solved by medication

PSYCHIARY

- Psychiatric diseases can arise if the environment is too stressing for the person,
- Symptoms can be very “painful”
- People with PWS are at greater risk for psychiatric diseases than others
- The symptoms can arise gradually or “out of the blue” (or first symptoms were not seen)
- Medication can help a lot. To continue

Psychiatric diseases in PWS

- Depression:
- Sadness, low self esteem, slow, no interest, low temperature (exclude low thyroids!)

- Psychoses:
- Hallucinations
- See things, hear voices, fearful thoughts
!! distinguish from childish fantasy (harmless)

MENTAL HEALTH IN ADULTS (PWS)

- Changes ?
- Dementia is not common (loss of abilities)
- Depression is common (low activity, no interest)
- Be aware of the difference

- Remember to write a status at least once a year

- What is important?
- To be happy and healthy and also with high self esteem

Emotions in PWS

- Love sameness
- Relax with well known daily routines
- Love praise
- Wants to please, Wants to be good ,
- Wants to be loved. Love to be loved
- Developmental age where this is typical: 2-3
- Good to know when we set our expectations

Why is it then so difficult ?

- The person with PWS is not just a bag of symptoms. There is person behind
- The one with PWS often does not know or understand very much about the disease and cannot manage without help
- If we shall succeed with help and personal assistance we must to try to understand why and how persons with PWS think and react

CARE and PWS

- Know the persons developmental age :
-
- Cognitive.. What can be understood
- Emotional....What is emotionally accepted
- Social..... How much can he/she interact

- Persons with PWS share a lot of symptoms, but their need of support varies a lot.

SUMMARY of messages

- Acute situations:
- different temperature and pain reactions , the eating
- Every day care, not weight related symptoms in PWS: eye and vision, dental care, a sensitive skin, scoliose and kyphose, intestine and bladder function, sleepines and hyperphagia. Hormone regulation
- Related to overweight: heart and lung function, oedema, incontinence, leg ulcers, reduced motor function, sleepiness, diabetes, side effects of medications. diabetes

REGULAR MEDICAL CHECK EVERY ½-1 year.

- * weight and blood pressure.
- *lung and heart and circulation, edema?
- *gastroparesis? *constipation ? urinary incontinence ?
- *skin, teeth, joints, back

Any medication ? And still needed ?

Is the food OK ?

Sexuality?

blood tests: hormones, blood sugar, vitamin D,

Physiotherapist : regular check and advices is given?

dietician and dentist every ½ year. DEXA scan every 2. year

Check vision and hearing every 2-3 year.

Check IPWSO guidelines: www.ipwso.org