The care of infant with Prader-Willi-Syndrome

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Overview

• Situation of infant with PWS
• Holistic treatment concept
• Nutrition
• Physiotherapy
• Early intervention
• Endocrine aspects
• Orthopedic problems
• Additional health issues
Situation of infant with PWS

Situation:
- Breech presentation, low birth weight,
- Premature delivery
- Adoption problems after delivery
- Breathing problems
- Feeding difficulties, underweight
- Distinctive muscle hypotonia, Hypokinesia
- Failure to thrive

Support:
- Breathing stimulation and aid (CPAP, BIPAP)
- Secure sleep position, Monitoring if available
- Intensive physiotherapy
- Nutrition training
- Tube feeding
- Assistive Devices
- Supportive measures
- Occupational therapy
Holistic PWS-treatment concept

- Nutritional management
- Therapy with hormones
- Psychosocial support
- Physical activity

PWS
Nutrition starts with physiotherapy

- Muscles are not ill in PWS, but muscle mass is smaller
- Increased muscle tone means increase muscles by physiotherapy
- Special feeding training for example Castillo Morales
- Stimulate the baby and take a special feeding position
- Use special baby bottle teats
- Sometimes tube feeding is necessary
Practical aspects of nutrition

- PWS infant needs 2/3 of energy intake compared with a healthy infant of the same age
- Wake for feedings every 4 hour during the first month
- Give weight chart to the parents for self control
- Target: reach 3.-10. percentile in the weight chart
- Weigh every week and report in growth chart
- Control energy intake in accordance to weight development
- At the end of the first year feeding becomes easier and the infant grows appropriately
From milk to infant food

Breast milk or Baby milk

Vegetable-potatoes-meat-mash

Whole milk-cereal-mash

Cereal-fruit-mash

Snack

Bread-milk-meal

Month of motoric developmental age
Educate family in nutrition

Target: Taking part at the family meals when able to sit at the table, maybe use a special baby seat

• Start with mashed meals when infant can control head in upright posture
• Add enough unsaturated fatty acids (rape oil)
• Be careful if children have dysphagia
• Chewing is a good training for speaking
• Give more and more solid food for learning to chew and moving the tongue (supervise chewing)
• Give piece of bread instead of cookies
• No sweet drinks
• Conditioning taste
Physiotherapy

- Vojta therapy based on neurophysiological principles
- Bobath concept to support movement development
- Speech therapy - Castillo Morales
Early intervention for Infant with PWS

- First: Love your child
- Spend time with the baby
- Turning from left to right when changing nappies
- Playing in prone position when awake
- Baby massage
- Kangaroo – body contact
- Sing, speak to the baby
- Look for eye contact
- Stimulate with music, colors, lights
- Be sensitive to small changes
- Baby swim
Cryptorchidism

- Therapy: Operation at the end of first year
- Potentially preparation with GnRH-nose spray 3 x 2 intakes for 4 weeks or β-hCG i.m. 3 x 1500 IE
- Anaesthesia: www.orphanetanaesthesia.eu

Labial synechia

- Estrogen containing Salve locally approx. 2 weeks, repetition if necessary
- Cave: possible side effect: temporary breast growth
Endocrine aspects II

Thyroid problems
• Infants with PWS have central or peripheral hypothyroidism that’s why check TSH and fT4
• Treatment with L-Thyroxin

Central adrenal insufficiency (CAI)
• Education on rare observation of CAI in PWS
• In urgent cases of clinical symptoms of CAI after check of laboratory parameters 30 mg/m² body surface area hydrocortisone in 3 parts
Treatment with growth hormone

• From age of 6 month until 98% of final height
• No growth hormone test necessary if PWS is genetically confirmed

Target:
• Improvement of body composition
• Increase of final height

Conduction of therapy:
• Dose recommendation max. 0,035 mg/kg body weight/day sc
• Dose management by IgF1, IgFBP3, BIA, longitudinal development/growth speed
• Check for side effects: metabolism, sleep apnea, heart
Prevent orthopedic problems

Prevent Scoliosis:
- No early passive sitting
- Sitting is allowed when child reaches position themselves
- First crawling than sitting
- Build up muscles at the trunk

Prevent Foot problems:
- massage, bandage, tape
- Prevent cranial deformations: change head and toe position in bed every day
Additional health issues

Problems:
- High pain threshold
- Poor thermoregulation
- Teeth erosion-loss of enamel/severe tooth wear
- Decreased vomiting
- Daytime sleepiness

Solutions:
- Attention in case of accidents
- Need intense stimuli
- Caution for hot food but also bathing water and clothing selection
- Start dental care with the first tooth 2x per day, drink water, no sweets
- Watch infant carefully
- Structure the day, plan time for daytime naps

13.03.2021
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The following publications are sponsored by Pfizer:

- PWS Sport book, 2016 (German, Dutch)
- PWS Nutrition book, 2006 (German)
Conclusion

- Early intervention supports infants with PWS and their families in daily life
- Muscular hypotonia is a central problem and has an impact on every aspect of life of an infant with PWS
  - Therefore, physiotherapy is necessary from the first day on to build up muscles
  - Additionally, growth hormone treatment from 6 months on helps to build up muscles
- Energy requirement is lower - up to 2/3 of a healthy infant
  - Individual energy intake has to be adopted to the development according to the weight chart
- Development and therapeutic measures shall be verified every three months
- Orthopedic problems are common