Prader-Willi Syndrome
Adolecent and Adult Endocrinology

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Disclosures

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Content

- Growth hormone (GH) deficiency
- Central adrenal insufficiency
- Central hypothyroidism
- Hypogonadism
Hormone deficiencies are common in PWS

- PWS is a disease with hypothalamic dysfunction
- The hypothalamus regulates appetite, metabolism, body temperature, mood, sleep and secretion of hormones from the pituitary

- Hormone deficiencies are common in PWS
  - Growth hormone deficiency
  - Central adrenal insufficiency
  - Central hypothyroidism
  - Hypogonadism
Growth hormone (GH) effects in the body

- Increases skeletal growth
- Increases muscle mass
- Decreases fat mass
- Increases metabolism
- Benefical for blood lipids
- Increases blood glucose
- Improves Quality of Life
Current registered indications for use of GH in PWS

• PWS is a registered indication for GH treatment in children with PWS

• PWS is registered for treatment in the transition period or for adults with PWS only in a few countries

• In most countries the diagnosis of GH deficiency in adults has to be established according to guidelines
Effects of GH treatment in children

GH treatment in children improves:
• height
• body composition
• motor skills
• cognitive function
• behaviour
Effects of GH treatment in adults

- All studies show a decrease in fat mass and increase in lean body mass

- Improvement in measures of physical performance during GH therapy was reported in all studies

- Limited number of studies did not show any effect on bone mineral density

- Improvements in measures of neuropsychological function as well as QoL have been repeatedly reported
Few and expected side effects of GH treatment

- A small increase in fasting glucose and trends towards higher insulin and insulin resistance but not for diabetes

- Transient and dose dependent side effects:
Central Adrenal Insufficiency?

Conflicting results, central adrenal insufficiency might be present in a few Evaluation and Hydrocortisone treatment when clinically indicated
Central hypothyroidism seems to be less frequent in adults but should be evaluated on a regular base at any age.
Low level of sex hormones
Hypogonadism

Primary gonadal deficiency or hypothalamic dysfunction

Males
- Incomplete puberty
- Lack of male body habitus
- No cases of paternity reported

Females
- Incomplete puberty
- Primary amenorrhea or oligomenorrhea
- Four documented cases of fertility worldwide
Treatment of Hypogonadism

- Sex hormones are used for induction of puberty in both genders
- Only one study with sex hormone treatment in adult males confirming the clinical experience secondary sex characteristics respond well to treatment with sex steroids
- Sex hormone treatment is not feasible for all

- Many adolescents and adults have a desire for romance, fantasies of marriage and a wish for children
- Education in relationship, sexuality and contraception is needed
Conclusions

• PWS is associated with documented hormone deficiencies which should be monitored and treated when indicated

• Hormone therapy does not change the intrinsic abnormalities of PWS but offers an opportunity to reduce some of the adverse consequences of PWS

• Treatment of hormonal deficiencies can be a challenge due to the somatic and behavioural phenotype
Thank you

Professor Andrea Prader