

Prader-Willi Syndrome

Adolescent and Adult Endocrinology

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Disclosures

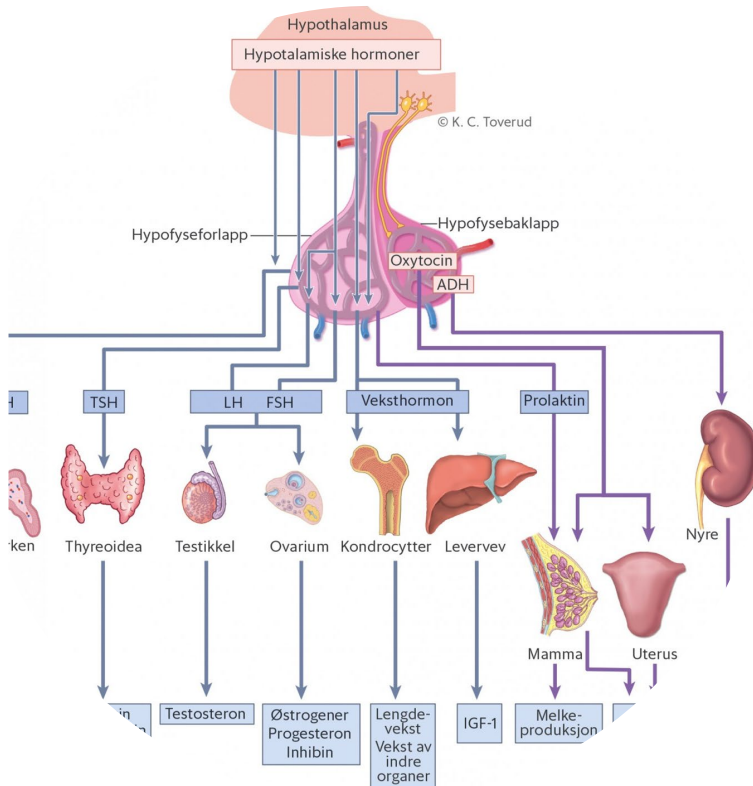
Member of Nordinet IOS, Patro and Swedish KIMS board (Pfizer)
Patro investigator

Have received lecture fees from NovoNordisk, Sandoz and Pfizer

Content

- Growth hormone (GH) deficiency
- Central adrenal insufficiency
- Central hypothyroidism
- Hypogonadism

Hormone deficiencies are common in PWS

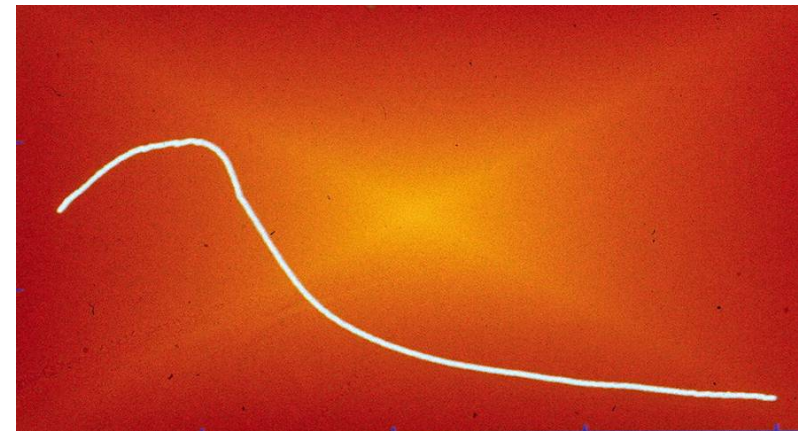


- PWS is a disease with hypothalamic dysfunction
- The hypothalamus regulates appetite, metabolism, body temperature, mood, sleep and secretion of hormones from the pituitary
- Hormone deficiencies are common in PWS
 - Growth hormone deficiency
 - Central adrenal insufficiency
 - Central hypothyroidism
 - Hypogonadism

Growth hormone (GH) effects in the body

- Increases skeletal growth
- Increases muscle mass
- Decreases fat mass
- Increases metabolism
- Beneficial for blood lipids
- Increases blood glucose
- Improves Quality of Life

Age and GH secretion



Age

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**

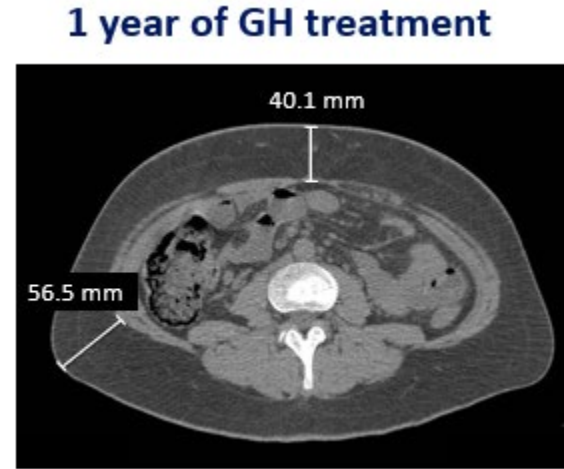
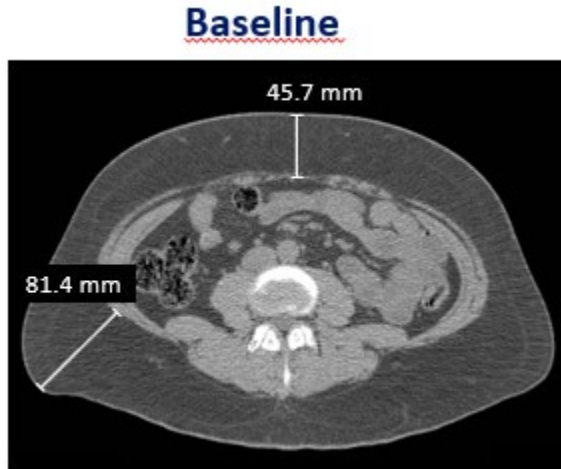
Effects of GH treatment in children

GH treatment in children improves:

- height
- body composition
- motor skills
- cognitive function
- behaviour

Effects of GH treatment in adults

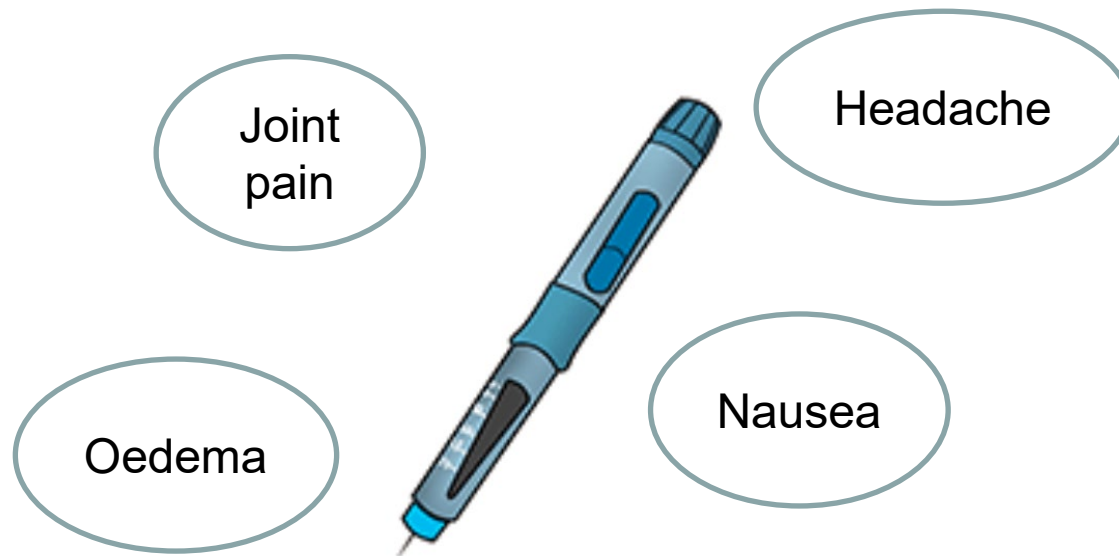
- All studies show a decrease in fat mass and increase in lean body mass



- 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

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