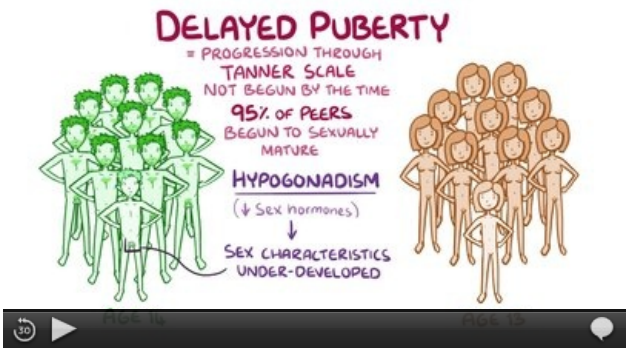
Hypogonadism

Definition

Delayed puberty (*pubertas tarda*) is an absence of breast development in girls after 13 years of age or prepubertal testicular size in boys after 14 years of age.

Classification

1. **functional hypogonadotropic hypogonadism** – a transient disorder in which there is a delay in sexual development due to an imbalance of energy intake and expenditure (excessive exercise, malnutrition, inflammatory bowel disease (IBD), chronic lung disease, endocrinopathy); clinic: *puberty delay, growth retardation and weight failure;*
2. **permanent hypogonadotropic hypogonadism** – arises from a number of congenital and acquired causes; acquired causes are, for example, inflammations, tumors and injuries of the CNS and hypothalamus or their iatrogenic damage; congenital causes are eg Kallman's syndrome (a disorder of formation and migration of neurons forming GnRH and agenesis of bulbus olfactorius - *hypogonadism + anosmia*), Prader-Willi syndrome (defect of chromosome 15 with hypothalamic involvement - (*hyperphagia, hypogonadism, growth retardation*), Laurence-Moon-Biedl syndrome (*hypogonadism, PMR, polydactyly, obesity, short stature*) gonadoliberin or LH / FSH receptor mutations;
3. **hypergonadotropic hypogonadism** – the causes are genetic syndromes – Klinefelter's syndrome (extra X chromosome in men – gynecomastia, eunuchoid habitus, hypogonadism), Turner syndrome (structural or numerical abnormality of the X chromosome in women – short stature, gonadal dysgenesis, congenital defects), congenital errors of metabolism – galactosemia, mucopolysaccharidosis, cryptorchidism (testicular descent disorder), testicular dysgenesis/agenesis, testicular injury/torsion, infections (parotitis), autoimmune disorders (AI oophoritis/orchitis), iatrogenic testicular involvement in cancer treatment;
4. other causes – androgen production defect, androgen resistance.



English summary video - definition, pathogenesis, symptoms, complication, treatment.

Diagnosis

A comprehensive examination of the child; anamnesis + physical examination; laboratory: LH / FSH, testosterone, estradiol, prolactin, TSH/fT4, iontogram, urea, creatinine, liver enzymes, IGF-I, growth hormone; further examination: exclusion of AI disease; genetic testing; imaging methods (USG - ovaries in girls, brain MRI in central hypogonadism); stimulation tests (GnRH stimulation test – demonstration of central hypogonadism, hCG stimulation test –stimulation; gonads and determination of hormone levels at low gonadotropin levels).

Treatment

In functional disorders, treatment of the underlying disease + induction of puberty by testosterone in boys, transient administration of hCG, in girls transient administration of estrogens; permanent hypogonadism - sex hormone replacement therapy (we start from small doses which we gradually increase). In girls, we start estrogen therapy a and after 2 years we add progestogens to induce an artificial menstrual cycle. We then proceed with cyclic administration of both hormones to achieve feminization. We administer testosterone to the boys.

References

Literature

- LEBL, Jan. *Klinická pediatrie.* 1. edition. Praha : Galén, 2012. ISBN 978-80-7262-772-1.