

Testicular feminization

Testicular feminization or syndrome of complete androgen resistance (**complete androgen insensitivity syndrome – CAIS**), is characterized by karyotype **46,XY** and **female external genitalia** (frequency 1/20000 to 60000 newborn boys). However, there is also a syndrome of incomplete androgen resistance (**partial - PAIS**) or resistance with normal male genitalia (**mild - MAIS**). Most cases are hereditary (70 %)^[1].

Causes

The main cause of the syndrome is **androgen receptor defect**. Gene (**AR**), encoding this receptor is located at **X chromosome (Xq12)**.

According to the defect, we can divide patients into:

- **receptor-negative** – they typically have point mutations or substitutions in the exons of a given gene encoding a receptor-binding domain androgens;
- **receptor-positive** – in which an exon point mutation allows normal androgen-receptor junction, but this complex is unable to bind to DNA and trigger transcription.

Diagnostics and symptoms

Congenital inguinal hernias may be one of the first symptoms. 1-2% of all women with this hernia suffer from androgen resistance. Affected women tend to be **taller, slender, with long lower limbs**. IQ is normal. The suspicion of the syndrome arises in case of primary amenorrhoea and very little to no pubic hair. The ultrasonograph will show the absence of organs from the Müllerian leads, the gynecological examination will show the blind end of the vagina with the absence of a neck. The uterus as well as other structures from Wolff's ducts or prostate do not develop.

Treatment

The female role of patients is usually strengthened. Patients are often castrated before or after puberty because the **risk of gonadal neoplasia** increases with age. In case of castration before puberty, substitution with estrogens is necessary. For milder androgen resistances, the opposite choice is also possible.

Links

Related articles

- Endocrine diseases of the gonads
- Disorders of sexual differentiation and sexual maturation

External links

- English Wikipedia (https://en.wikipedia.org/wiki/Complete_androgen_insensitivity_syndrome)

References

- GREENSPAN, Francis – BAXTER, John. *Základní a klinická endokrinologie*. - edition. H & H, 2003. pp. 843. ISBN 9788086022567.
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References

1. | KOLEKTIV, Roztočil. *Moderní gynekologie*. - edition. Grada Publishing a.s., 2011. 508 s. ISBN 9788024728322.]