

Ontogenesis of sex in mammals

Ontogenetic notes

- **Gonosomal complement**
 - female:XX,
 - male: XY.
- The Y chromosome (short arms) contains a highly conserved **SRY (sex determining region of Y)** region, which contains the '**testis specific protein Y-linked (TSPY) gene for male gonadal differentiation**'.
- Gene TSPY is peculiar in that it contains no introns and ends with a polyadenyl-like sequence (hypothesized to have originated by reverse transcription from mRNA, but this must have happened a very long time ago - 166 million years).
- The SRY region is homologous to genes for the formation of non-histone proteins on the X chromosome.
- **The product of the TSPY gene binds to the promoter:**
 1. the gene for **cytochrome-P450-aromatase**, which converts testosterone into the female hormone estradiol and inhibits its transcription,
 2. the **Müllerian inhibitory substance** gene, which is responsible for testicular differentiation and female organ regression and activates its transcription.
- *By week 6, human gonadal development is indifferent,*
- the bases of the gonads develop in the **plica genitalis**,
- **male:**
 1. the testes develop from the coelomic epithelium,
 2. **Sertoli's bb'** - from the ductal cells of the mesonephros, producing MIF (Müllerian inhibition factor),
 3. **Leydig's bb'** - from the mesenchyme, under the action of hCG they start to produce testosterone, which stimulates the development of Wolffian ducts and male external genitalia and the descent of the testes,
 4. sperm development is regulated by genes on the long arms of the Y chromosome,
- **female:**
 1. in the absence of SRY, the fallopian tubes, uterus and part of the vagina develop from the Müllerian ducts,
 2. female genitalia develop under the influence of estradiol,
 3. the coelomic epithelium gives rise to the ovarian cortex,
 4. oogonia enter mitotic division 3 to 6 months prenatally, forming oocytes,
 5. indeferent bb change to follicular (ca. 4th month), surround the oocyte and give rise to primordial follicles → oocytes of primordial follicles enter reductive division and remain in the dictyotenic stage (diplotene prophase of meiosis I) until follicle maturation at puberty (and beyond),
- **development of sexual dimorphism is completed at 12 to 14 weeks' pregnancy.**

Disorders of sexual development

Gonadal dysgenesis

- Turner syndrome - karyotype 45,X.

True hermaphroditism

- Basis of both ovary and testes,
- chimera 46,XX/46,XY (fusion of two zygotes).

Male pseudohermaphroditism

- Testes and female or bilateral genitalia,
- testicular **feminization'** - insensitivity to testosterone, no receptors for it are expressed, karyotype 46,XY,
- incomplete insensitivity to testosterone,
- mutations geneu for 5 α -reductase,
- mosaicism 45,X/46,XY.

Female pseudohermaphroditism

- ovaries, '**masculinization** of the external genitalia
- overproduction of testosterone in the adrenal glands
- 21-hydroxylase block (adrenogenital syndrome, AR, gene located in the HLA class III region)

Links

Related Articles

- Chromosomal sex determination

References

- KAPRAS, Jan – KOHUTOVÁ, Milada. *Kapitoly z lékařské biologie a genetiky III.*. 1. edition. Karolinum, 2009. pp. 101. ISBN 978-80-246-0001-7.