

Pseudohermaphroditism

Hermaphroditism

Hermaphroditism is a type of abnormal differentiation and determination of sex. The main basis of this disorder is that the male and female reproductive systems are formed indifferently until a certain period of prenatal development.

True hermaphroditism (*hermaphroditism verus*) is very rare. It arises as a result of genetic mosaicism and in the affected individual is manifested by the presence of ovarian and testicular tissue at the same time. In certain cases, hermaphroditism can be caused by a single gene disorder or non-genetic influences, such as defects in the expression of SRY gene or genes encoding sex hormone receptors or altered levels of these hormones.

Pseudohermaphroditism

In the gonads of pseudohermaphrodites, there is tissue belonging to only one sex. **Female pseudohermaphroditism** is characterized by a 46,XX karyotype, the presence of ovaries and Barr's body, but male external genitalia. **Male pseudohermaphroditism** is characterized by a 46,XY karyotype, the presence of testes and feminization of the external genitalia.

Female pseudohermaphroditism

A common cause of female pseudohermaphroditism is adrenogenital syndrome.

Affected girls have normally developed ovaries, fallopian tubes and uterus, but due to the high production of androgens, they experience masculinization of the external genitalia, enlargement of the clitoris and fusion of the labia majora to form a scrotum-like structure. Vestibulum vaginae may persist in some cases. In the mildest form, the masculinization of the external genitalia is manifested only by an enlarged clitoris.

Male pseudohermaphroditism

It can arise from several reasons:

1. abnormal formation of testicles during embryonic development;
2. endocrine testicular disorder;
3. target cell defects (lack of receptors) for androgens.

An example of male pseudohermaphroditism is testicular feminization syndrome (X-linked androgen resistance syndrome). The inability of cells to respond to the action of androgens does not allow the differentiation of the male genital organs, and the preserved function of AMH (anti-Müllerian Hormone) also stops the development of the fallopian tubes and uterus. As a result of these processes, a short and blind-ending vagina is formed. Small, underdeveloped testicles are stored in the groin or labia and do not produce sperm.

Links

References

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