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Testicular feminization syndrome

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Abstract: aim of this study is to describe the testicular feminization syndrome {androgen insensitivity syndrome}

Introduction : testicular feminization is the syndrome when a male , genetically XY, because of various abnormalities of the X chromosome , is resistant to the action of the androgen hormones , which in turn stops the forming of the male genitalia and gives a female phenotype . The androgen insensitivity syndrome occurs in one out of 65000 births and can be incomplete {various sexual ambiguities} or complete {person appears to be a woman} the syndrome is X-linked recessive condition. The taint being transmitted maternally

Discussion: the syndrome results because, with the exception of the urogenital sinus {which may be oversensitive} , the target organs of the hormone such as breasts , hair follicles vocal cords and phallus are inherently insensitive to androgens . the androgen receptors may be completely absent or they may be present in normal numbers but insensitive to the androgen due to a mutation of these receptors , is a type of nuclear receptor that is activated by binding to either testosterone or dihydrotestosterone in the cytoplasm and translocate into the nucleus ehere it bind to DNA , provided androgen response elements and coactivators and coactivator are present this combination function as transcription to mediate the effects if androgen including development and maintaince of male sexual phenotype and generalized anabolic effects over 400 androgen receptors mutation have been reported the failure of virilization is either :-

1) complete androgen in sensevity syndrome [CAIS] the patient is an apparent female, with well developed breast and a normal vulva who presents with primary amenorrhea. The tubes and uterus are absent but urogenital science component of vagina is invariable present. Gonads are always testes and are found intra-abdominally or in hernia sacs. Plasma level of testosterone and other androgens are high due to increased LH

2) partial androgen insensitivity syndrome [PAIS] or reifenstein syndrome. The patient have reduced binding affinity of testosterone to receptors or maybe defect in transcription. The endocrine profile are similar to complete form, some men may have enlarged phallus and blind vaginal pouch of birth. There may be cryptochidism and gynecomastia, the testes are azoospermia.

3) In this form is 5- alpha reductase deficiency. Is a failure of conversion of testosterone to dihydrotestosterone at the target tissue. The presentation are as describes for [PAIS].

There is clinical recognizable syndrome found in patients who are essentially normal-appearing women, but who are undescended testes in place of ovaries. The most significant features are:-

1) Female habitus, breast development, and other secondary sex characteristics.
2) Scanty or absent axillary or pubic hair in most eases.
3) Female external genitals, with a tendency to underdevelopment of labia, and a blind-ending vagina.
4) Absence of internal genitals except for rudimentary anlage and for gonads which may be located intra-abdominally or along the course of the inguinal canal.

5) Gonads histologically consistent with undescended testes.

6) Urinary excretion studies have suggested such testes produce estrogen and androgen. Elevated gonadotropins have also found.

Conclusion:

Testicular feminization syndrome represents well-defined form of pseudohermophroditism. The patient should undergo or chidectomy because of the aggregate risk for malignant transformation androgen levels fall and management of [PAIS] depends on degree of ambiguity of genitalia. Some respond to high of TH genadectomy and hormone replacement therapy for those assigned the female sex.

Reference:

1. Ozülker T, Ozpaçaci T, Ozülker F, Ozekici U, Bilgiç R, Mert M (January 2010). "Incidental detection of Sertoli-Leydig cell tumor by FDG PET/CT imaging in a patient with androgen insensitivity syndrome". Ann Nucl Med. 24 (1): 35–9. doi:10.1007/s12149-009-0321-x. PMID 19957213.
2. Audi L, Fernández-Cancio M, Carrascosa A, et al. (April 2010). "Novel (60%) and recurrent (40%) androgen receptor gene mutations in a series of 59 patients with a 46,XY disorder of sex development". J. Clin. Endocrinol. Metab. 95 (4): 1876–88.
3. Sparrow R (2013). "Gender eugenics? The ethics of PGD for intersex conditions". Am J Bioeth. 13 (10): 29–38. doi:10.1080/15265161.2013.828115. PMID 24024804.
4. Behrmann J, Ravitsky V (2013). "Queer liberation, not elimination: why selecting against intersex is not "straight" forward". Am J Bioeth. 13 (10): 39–41. doi:10.1080/15265161.2013.828131. PMID 24024805.
5. Nisker J (2013). "Informed choice and PGD to prevent "intersex conditions"". Am J Bioeth. 13 (10): 47–9. doi:10.1080/15265161.2013.828125. PMID 24024809.
6. "Submission on the ethics of genetic selection against intersex traits". Organisation Intersex International Australia. Retrieved 28 September 2014.