

Causes of xy females



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Causes of XY Females A person having 46XY chromosome composition is known as an XY female. At birth the person appears to be a normal male or female but as the person grows up, some of the desired male or female characteristics remain underdeveloped and start giving a look of opposite sex to the person. For example a male start appearing with unusual breast developments while, in females it starts growth of hair on faces, lack of menstrual cycle firming up of muscles etc. Such persons are in fact quite different from transsexuals i. e. those who voluntarily go for sex transformation and change their phenotype from male to female.

Studd (2003) points out that earlier such condition was called Testicular Feminization Syndrome (TFS), but this is now being considered somewhat inappropriate and inaccurate because the testes are not known to produce feminizing factors. This phenomenon is therefore known as Androgen Insensitivity Syndrome (AIS). This insensitivity too is found to occur in varying measures in people. The condition when the Androgens find no response with age, is termed as Complete Androgen Insensitivity Syndrome (CAIS).

The growth as an 'XY female' is considered as abnormal and hence put into the category of patients. Researchers have found that such patients lead a normal life till their abnormal condition is diagnosed by doctors. For example, when a form of male pseudohermaphroditism or testicular feminization syndrome is detected. In this case the androgen receptor sees mutations which in result in X-linked recessive form androgen insensitivity. The childhood of such persons remains quite normal but when the child enters the age of puberty, problem starts. In fact, normally even the parents fail to fathom the distinctive nature of their children and they come to know

about it only when they visit specialist gynecologists in order to treat the sterile behavior.

Abnormality in Androgen Receptor (AR) is stated to be the reason behind CAIS. Testes are formed normally in the fetus when the SRY gene even in cases of CAIS. But as the age grows, the testes start secreting anti-Mullerian Hormones (AMH) leading to the deterioration of Mullerian ducts. This deprives a CAIS woman from a uterus. Though the body also produces testosterone at the opportune moment, but due to the non-cooperation of the Androgen Receptor, the genitalia fail to virilise and undergo undesirable development. Such developments also impact some of the brain cells, thus impacting the psychological behavior of the person as well. This ultimately results in a person who considers herself as a female, but without some of the key characteristics like uterus, pubic and auxiliary hair etc.

Females with Y chromosomes are found to be associated with the risk of gonadoblastomas (tubular adenomas and seminomas) and persons with such symptoms go for removal of undescended testes. In XY females Mullerian and Wolffian duct derivatives are not present or under-grown while the vagina happens to be in a state termed as 'blind vagina'. Such people need to undergo timely psychological consultations in order to get over the complexes of being unequal or inferior as compared to family members or friends around them. Treatment is also possible in cases where the deviation is within controllable limits, but majority of such people have to live with it, because either the treatment happens to be very costly or there's no credible treatment.

References:

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