Androgen Insensitivity Syndrome

Subjects: Genetics & Heredity
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Androgen insensitivity syndrome is a condition that affects sexual development before birth and during puberty. People with this condition are genetically male, with one X chromosome and one Y chromosome in each cell. Because their bodies are unable to respond to certain male sex hormones (called androgens), they may have mostly female external sex characteristics or signs of both male and female sexual development.

Keywords: genetic conditions

1. Introduction

Complete androgen insensitivity syndrome occurs when the body cannot use androgens at all. People with this form of the condition have the external sex characteristics of females, but do not have a uterus and therefore do not menstruate and are unable to conceive a child (infertile). They are typically raised as females and have a female gender identity. Affected individuals have male internal sex organs (testes) that are undescended, which means they are abnormally located in the pelvis or abdomen. Undescended testes have a small chance of becoming cancerous later in life if they are not surgically removed. People with complete androgen insensitivity syndrome also have sparse or absent hair in the pubic area and under the arms.

The partial and mild forms of androgen insensitivity syndrome result when the body's tissues are partially sensitive to the effects of androgens. People with partial androgen insensitivity (also called Reifenstein syndrome) can have genitalia that look typically female, genitalia that have both male and female characteristics, or genitalia that look typically male. They may be raised as males or as females and may have a male or a female gender identity. People with mild androgen insensitivity are born with male sex characteristics, but they are often infertile and tend to experience breast enlargement at puberty.

2. Frequency

Complete androgen insensitivity syndrome affects 2 to 5 per 100,000 people who are genetically male. Partial androgen insensitivity is thought to be at least as common as complete androgen insensitivity. Mild androgen insensitivity is much less common.

3. Causes

Mutations in the *AR* gene cause androgen insensitivity syndrome. This gene provides instructions for making a protein called an androgen receptor. Androgen receptors allow cells to respond to androgens, which are hormones (such as testosterone) that direct male sexual development. Androgens and androgen receptors also have other important functions in both males and females, such as regulating hair growth and sex drive. Mutations in the *AR* gene prevent androgen receptors from working properly, which makes cells less responsive to androgens or prevents cells from using these hormones at all. Depending on the level of androgen insensitivity, an affected person's sex characteristics can vary from mostly female to mostly male.

3.1. The gene associated with Androgen insensitivity syndrome

- AR

4. Inheritance

This condition is inherited in an X-linked recessive pattern. A condition is considered X-linked if the mutated gene that causes the disorder is located on the X chromosome, one of the two sex chromosomes in each cell. In genetic males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In genetic females (who have two X chromosomes), a mutation must be present in both copies of the gene to cause the disorder. Males are affected by X-linked recessive disorders much more frequently than females.

About two-thirds of all cases of androgen insensitivity syndrome are inherited from mothers who carry an altered copy of the *AR* gene on one of their two X chromosomes. The remaining cases result from a new mutation that can occur in the mother's egg cell before the child is conceived or during early fetal development.

5. Other Names for This Condition

- AIS
- androgen receptor deficiency
- androgen resistance syndrome
- AR deficiency
- DHTR deficiency
- dihydrotestosterone receptor deficiency

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