Condition: Androgen insensitivity syndrome

Inheritance:
X-linked recessive.

Genetic etiology:
Mutation in *AR* gene, which encodes the androgen receptor.

Frequency:
1/20,000 – 1/50,000.

Clinical features:
Individuals with androgen insensitivity syndrome have complete or partial insensitivity to the effects of androgens due to dysfunction of the hormone receptor. Those with complete insensitivity have normal female external genitalia, but have undescended testes and lack uterus and Fallopian tubes due to action of Mullerian-inhibiting substance. Those with partial insensitivity have some degree of masculinization of the external genitalia, and therefore may present with ambiguous genitalia.

Management:
Individuals with complete androgen insensitivity have a female phenotype; gonadectomy is performed after puberty to prevent gonadoblastoma. Vaginal dilation may be required. For those with partial androgen insensitivity, sex assignment and management is a complex issue and requires a multidisciplinary approach, including surgical, endocrinological, genetic, and psychological support.

Genetic counseling:
Based on X-linked inheritance; genetic testing is available. Some carriers may have manifestations based on nonrandom X chromosome inactivation.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.