Condition: Congenital adrenal hyperplasia

Inheritance:
Autosomal recessive.

Genetic etiology:
Most common form due to mutation in *CYP21A2* gene which codes for 21-hydroxylase. Mutations in genes for other enzymes in the adrenal steroid biogenesis pathway lead to other forms of CAH.

Frequency:
1/15,000.

Clinical features:
The classic form of congenital adrenal hyperplasia is due to mutation in the gene encoding 21-hydroxylase. This leads to decreased cortisol production and increased production of adrenal androgens. This leads to virilization at birth, in childhood, and to precocious puberty. In females, this may present at birth with ambiguous genitalia. Approximately 25% have only virilization, whereas the others have, in addition, salt wasting due to aldosterone deficiency.

Management:
Treatment with glucocorticoids, and, in cases of salt wasting, with mineralocorticoid. Virilized females will require surgical treatment of abnormal genitalia. Women at risk of having an affected fetus are treated with dexamethasone until it can be determined if the fetus is female and affected; if not, the dexamethasone treatment is discontinued. Such treatment can avoid virilization of affected females.

Genetic counseling:
Based on autosomal recessive transmission; molecular genetic testing is available.

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