Condition: 45,X (Turner syndrome)

Inheritance:
Chromosomal

Genetic etiology:
45,X karyotype due to nondisjunction or loss of a structurally abnormal X or Y chromosome. Many are mosaics, with a cell line containing 46 chromosomes with a structurally abnormal X or Y.

Frequency:
Approximately 1/3,000 female livebirths.

Clinical features:
Individuals with Turner syndrome have a female phenotype, but may fail to develop normal secondary sex characteristics, such as pubic and axillary hair. They tend to have short stature, primary amenorrhea and usually are infertile. Coarctation of the aorta may occur, and some have renal anomalies. Lymphedema is common at birth, and redundant nuchal skin may lead to "webbing" of the neck. Learning disabilities, especially visual-spatial perceptual problems, are common.

Management:
Treatment with hormones to promote secondary sexual development; surgical correction of congenital heart defects; anticipatory guidance and early intervention to overcome learning disabilities.

Genetic counseling:
Recurrence is rare and can be detected by prenatal chromosomal analysis.

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