Condition: 47,XXX

Inheritance:
Chromosomal

Genetic etiology:
47,XXX karyotype due to nondisjunction; some have more than 3 X chromosomes.

Frequency:
Approximately 1/1,000 liveborn females.

Clinical features:
Individuals with XXX syndrome have a female phenotype and tend to be fertile. They tend to have cognitive impairment but usually do not have major congenital anomalies.

Management:
Anticipatory guidance and support for developmental impairment.

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.