Condition: XXY (Klinefelter syndrome)

Inheritance:

Chromosomal

Genetic etiology:

47,XXY karyotype due to nondisjunction; some have additional X chromosomes.

Frequency:

1/500 – 1/1,000 male livebirths.

Clinical features:

Individuals with Klinefelter syndrome have a male phenotype, but may fail to develop normal secondary sex characteristics, such as pubic and axillary hair. They tend to have small testes and usually produce few or no sperm. Some degree of breast development may occur. Learning disabilities, especially involving language skills, are commonly seen.

Management:

Treatment with testosterone to promote male secondary sexual development; counseling regarding probable infertility; early intervention to manage 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.