Condition: Y chromosome infertility

Inheritance:
Y-linked.

Genetic etiology:
Deletion of one or more genetic regions on the Y chromosome, designed *AZFa, AZFb, AZFc.* Genes include *DAZ1, DDX3Y, RBMY1A1,* and *USP9Y.*

Frequency:
1/2,000 – 1/3,000.

Clinical features:
Affected males have azoospermia or oliogospermia. Some may have small testes or cryptorchism.

Management:
In vitro fertilization may be possible using intracytoplasmic sperm injection (ICSI) using sperm extracted from the testes.

Genetic counseling:
Transmission is via the Y chromosome, which is important to recognize if a male is conceived from an affected father by ICSI. Genetic testing is available.

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