Condition: Retinoblastoma

Inheritance:
Autosomal Dominant**.**

Genetic etiology:
Rb gene mutation.

Frequency:
Approximately 1/15,000.

Clinical features:
Unilateral or bilateral retinoblastoma occurring during childhood; osteosarcoma beginning during adolescent years.

Management:
Symptomatic management and monitoring for tumors.

Genetic counseling:
Affected individuals at 50% risk of transmitting a gene mutation; both familial and de novo cases occur. Genetic testing is available.

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