Condition: Li-Fraumeni syndrome

Inheritance:

Autosomal Dominant

Genetic etiology:

Mutation in the *TP53* tumor suppressor gene.

Frequency:

Rare.

Clinical features:

Cancers involving multiple sites, including soft tissue sarcoma, osteosarcoma, breast cancer, gastrointestinal tract, pancrease, glioma, melanoma, and leukemia.

Management:

Symptomatic treatment and surveillance for development of cancer.

Genetic counseling:

Risk of transmission is 50%, with high penetrance for eventual development of cancer. Genetic testing is available.

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