Condition: Wilson disease

Inheritance:
Autosomal recessive.

Genetic etiology:
Mutation in gene ATP7B, involved in complexing copper with the protein ceruloplasmin, involved in excretion of copper from liver cells into bile. Pathology results from failure to normally excrete copper from liver cells into bile. There is low serum copper and ceruloplasmin, increased urinary excretion of copper, and deposition of copper into tissues.

Frequency:
1/30,000.

Clinical features:
Major features are liver dysfunction, neurological and psychiatric problems, and Kayser-Fleischer rings in Decemet's membrane of the cornea. Liver dysfunction includes episodes of jaundice, acute or chronic hepatitis-like disease, and chronic liver failure. Neurologic symptoms include tremors, motor incoordination, and spastic dystonia. Psychiatric symptoms include depression, neurotic behavior, and loss of intellectual function.

Management:
Use of copper chelating agents, most commonly penicillamine, and ingestion of zinc, which interferes with copper absorption; restriction of intake of foods rich in copper; liver transplantation for those with irreversible liver failure.

Genetic counseling:
Based on autosomal recessive inheritance; genetic testing is available.

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