Condition: Familial hypercholesterolemia

Inheritance:

Autosomal dominant, with more severe disease in homozygotes.

Genetic etiology:

Mutation in *LDLR* gene, encoding the low density lipoprotein receptor.

Frequency:

Heterozygotes approximately 1/500; homozygotes approximately 1/1 million.

Clinical features:

The disorder is associated with partial (heterozygotes) or complete (homozygotes) dysfunction of the LDL receptor, leading to hypercholesterolemia and premature atherosclerosis. Age of onset is earlier and severity is greater in homozygotes.

Management:

Supportive care; dietary restriction; statin drugs may be helpful, but have limited effectiveness in setting of very high cholesterol.

Genetic counseling:

Carrier status transmitted as autosomal dominant trait; molecular genetic testing available.

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