Condition: Osteogenesis imperfecta

Inheritance:
Autosomal dominant.

Genetic etiology:
Mutations in genes encoding components of type I collagen (*COL1A1* and *COL1A2*).

Frequency:
Approximately 1/15,000.

Clinical features:
The basic phenotype of osteogenesis imperfect is the occurrence of brittle bones with high tendency to fracture. In some instances there is associated short stature. Other features include bluish sclerae, hearing loss, and dentiogenesis imperfecta (weakening of the teeth). There is a wide range of variable expression, including neonatal onset with lethality early in life at one extreme, and relatively mild expression at the other.

Management:
Supportive therapy, in particular management of fractures; bisphosphonate therapy to prevent fractures is under investigation.

Genetic counseling:
Based on autosomal dominant transmission; both biochemical and molecular genetic testing are available.

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