Condition: Ehlers-Danlos syndrome

Inheritance:

Genetically heterogeneous; most forms autosomal dominant.

Genetic etiology:

Genetically heterogeneous; the classic type of EDS (types I and II) is associated with mutations in the genes encoding peptides involved in forming type V collagen (*COL5A1* and *COL5A2*). Type III is due to mutation in *TNXB* (encoding tenascin-XB); type IV is due to mutation in *COL3A1* (type III collagen).

Frequency:

Classic EDS type I: 1/20,000.

Clinical features:

Ehlers-Danlos syndrome is a broad term that includes multiple distinct disorders of connective tissue characterized by hyperextensibility of skin and hypermobility of joints. In the classic type, skin is highly extensible, smooth, and velvety; there is delayed, abnormal wound healing and generalized tissue fragility; hypermobility of joints; mitral valve prolapsed, and, sometimes, rupture of large arteries. Many other forms of EDS have been described, with various combinations of cutaneous and joint involvement, and different degrees of severity. EDS Type III affects mostly the joints. EDS Type IV leads to rupture of organs (e.g., intestine, uterus) and major blood vessels may occur.

Management:

Supportive management.

Genetic counseling:

Most forms are autosomal dominant; genetic testing is available for some of the genes involved.

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