Condition: Hemophilias

Inheritance:

Genetically heterogeneous; most common X-linked recessive.

Genetic etiology:

Hemophilias result from mutation in various genes that encode the proteins involved in the blood clotting cascade. The most common are hemophilia A and B, due respectively to deficiency of factors VIII or IX. Both of these are X-linked recessives. The most common F8 gene mutation is an inversion that splits the gene.

Frequency:

Hemophilia A: 1/4,000 at birth; hemophilia B: 1/20,000 at birth.

Clinical features:

Hemophilia is characterized by deficient activity of the blood clotting system. Most commonly, bleeding episodes occur into joints and soft tissues after minor trauma. Bleeding can also occur after surgical procedures, including circumcision or tooth extraction.

Management:

Infusion of factor VIII or IX during episodes of bleeding; supportive care.

Genetic counseling:

Hemophilia A and B are transmitted as X-linked recessive traits. Genetic testing is available.

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