Condition: Hereditary angioedema

Inheritance:
Autosomal dominant.

Genetic etiology:
Mutation in *C1NH* gene, encoding complement C1 inhibitor gene.

Frequency:
1/50,000.

Clinical features:
Affected individuals have episodes of edema, particularly involving the extremities, nasopharynx and larynx, along with symptoms of abdominal pain, diarrhea, and vomiting. It is associated with C1 esterase inhibitor deficiency, leading to activation of the complement pathway.

Management:
Prophylaxis of attacks by treatment with attenuated androgens (e.g., Danazol), or antifibronolytic agents (e.g., epsilon-aminocaproic acid). Infusion of C1 inhibitor concentrate can be administered for prophylaxis at times of high risk, as before surgical procedures. Acute attacks may be treated with attenuated androgens or infusion of C1 inhibitor.

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

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