Condition: Fabry disease

Inheritance:
X-linked recessive

Genetic etiology:
Mutation of *GLA* gene, encoding alpha galactosidase A.

Frequency:
1/50,000 males.

Clinical features:
Fabry disease is a lysosomal disorder due to failure to break down globotriaosylceramide in vascular endothelial cells. Clinical presentation is with pain in extremities, angiokeratomas on the skin, hypohidrosis, opacities of the lens and cornea, and renal and cardiac dysfunction progressing to end-stage renal failure and cardiomyopathy. Some manifestations may occur in carrier females.

Management:
Supportive care; enzyme replacement by intravenous infusion.

Genetic counseling:
Based on X-linked recessive inheritance; molecular genetic testing is available.

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