Condition: Mucopolysaccharidosis

Inheritance:

X-linked recessive (Hunter syndrome) or autosomal recessive.

Genetic etiology:

Genetically heterogeneous. MPS I (Hurler syndrome) due to mutation in *IDUA*, which encodes alpha-L-iduronidase. MPSII due to mutation in *IDS*, resulting in deficiency of iduronate-2-sulfatase. Many other forms due to other enzymes involved in lysosomal metabolism of glycosaminoglycans.

Frequency:

Type I approximately 1/100,000; type 2 approximately 1/100,000 – 1/150,000 males.

Clinical features:

Classic Hurler syndrome presents in infancy or early childhood with coarsening of facial features, corneal clouding, dysostosis multiplex (skeletal anomalies), hearing loss, and neurological deterioration. A later onset form due to partial enzyme deficiency is referred to as Scheie syndrome. Type II presents in males with similar features but no corneal clouding Diagnosis is based on accumulation of glycosaminoglycans in urine and detection of deficient enzyme activity in blood leukocytes or fibroblasts.

Management:

Supportive care; bone marrow transplant has been used, and enzyme replacement therapy now is possible.

Genetic counseling:

Based on autosomal recessive inheritance for type I and X-linked recessive for type II; molecular genetic testing available.

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