



# POSNA

## The Core Curriculum

### Mucopolysaccharidoses

#### Objectives

1. Describe the common feature of all the mucopolysaccharidoses
2. Describe the genetic transmission of the mucopolysaccharidoses
3. Describe the organ systems involved in the different phenotypic expressions of the mucopolysaccharidoses

#### Discussion points

1. What is a mucopolysaccharide?
2. Would it be logical to call this family of disorders lysosomal storage disease? Why are the lysosomes enlarged?
3. What relationship does infantile kyphosis have with the mucopolysaccharidoses?
4. How is the diagnosis of a mucopolysaccharidosis presently established?

#### Discussion

The mucopolysaccharidoses are a family of rare genetic disorders all characterized by a defect in degradation of proteoglycans. A recent study in Northern Ireland estimated the overall incidence of mucopolysaccharidoses as 1:25,000 live births. The term mucopolysaccharide would be obsolete were it not fixed to these disorders, as the macromolecules formerly known as mucopolysaccharides or protein-polysaccharides are now called proteoglycans. They consist of a protein core with covalently bound polysaccharide (glysoaminoglycan) chains. Examples of glysoaminoglycans are chondroitin sulfates 4 and 6, keratan sulfate, heparin sulfate, and dermatan sulfate. Proteoglycans are normally degraded in the cell by lysosomes, an enzyme dependant process. Absence or deficiency of any necessary enzyme will result in an accumulation of proteoglycans in the intracellular lysosomes. Urinary excretion of excess amounts of the undegraded glysoaminoglycan is diagnostic. The resulting phenotype depends on the specific enzyme deficiency, with varying effects on the skeletal, ocular, visceral organs, or central nervous system. Two of the more severe forms of the mucopolysaccharidoses, Hunter and Hurler syndromes, may be suspected by noting infantile kyphosis. Morquio syndrome also has significant associated orthopaedic problems, consisting of short stature, kyphosis, and genu valgum. Stiff joints and short stature are features of Maroteaux-Lamy syndrome. All the mucopolysaccharidoses are transmitted by recessive inheritance, X-linked for Hunter and autosomal for the rest.

## References

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