

Clinical and Diagnostic Pearls in the Mucopolysaccharidoses For Primary Care Physicians

Self-Assessment Pre- and Post-test

1. Newborn infants with mucopolysaccharidoses most commonly present with:
 - A. small size for gestational age
 - B. limb anomalies
 - C. congenital heart disease
 - D. facial dysmorphisms
 - E. normal features

2. Definitive diagnosis of a mucopolysaccharidosis is most commonly based on
 - A. plasma amino acid analysis
 - B. urine organic acid analysis
 - C. urine thin layer chromatography
 - D. leukocyte enzyme analysis
 - E. skin fibroblast assay

3. The inheritance pattern of most MPSs is:
 - A. Autosomal recessive
 - B. Autosomal dominant
 - C. X-linked
 - D. Maternal inheritance
 - E. Multifactorial

4. The treatment of somatic features of some MPSs includes:
 - A. dietary restrictions
 - B. frequent orthopedic surgeries
 - C. enzyme replacement therapy
 - D. orthotopic liver transplantation
 - E. splenectomy

5. Patients with severe neurologic deterioration (regression) but little or no signs of hepatosplenomegaly or bone disease are most likely to have:
 - A. MPS I (Hurler syndrome)
 - B. MPS II (Hunter syndrome)
 - C. MPS III (Sanfilippo syndrome)
 - D. MPS IV (Morquio syndrome)
 - E. MPS VII (Sly syndrome)

6. Patients with signs of severe bone disease on radiographs but who have normal intelligence are most likely to have :
 - A. MPS I (Hurler syndrome)
 - B. MPS II (Hunter syndrome)
 - C. MPS III (Sanfilippo syndrome)
 - D. MPS IV (Morquio syndrome)
 - E. MPS VII (Sly syndrome)

7. A 4-year old patient who has short stature with beaked vertebrae and spatulate ribs seen on lateral chest radiographs, developmental regression and coarse facial features should have as the next step in the diagnostic evaluation:
 - A. brain MRI
 - B. plasma amino acids
 - C. urine glycosaminoglycans analysis
 - D. leukocyte iduronidase enzyme assay
 - E. bone marrow biopsy

8. A patient with severe bone disease and neurological regression is found to have iduronate sulfatase enzyme deficiency, diagnostic of MPS II (Hunter syndrome). He has an unaffected 6-year old sister. Genetic counseling for this family would include which of the following important discussions:
 - A. that the parents have a 50% chance with each pregnancy of having a child with the genetic deficiency.
 - B. that the sister may be an unaffected carrier and if so, would have a 50% chance of passing the genetic abnormality on to her sons
 - C. that the paternal uncle of the patient has a 50% risk of being an unaffected carrier
 - D. that the maternal uncle has a 50% risk of being an unaffected carrier
 - E. that the patient's children will have a 50% chance of having the disease

9. Treatment with enzyme replacement therapy have which of the following drawbacks:
 - A. the enzyme treatment needs to be given several times per day
 - B. the enzyme treatment has a high rate of antibody inactivation
 - C. the enzyme treatment does not affect neuronopathic disease
 - D. the enzyme treatment has not been tried in humans
 - E. the enzyme treatment frequently causes nausea and light-headedness

10. Ongoing monitoring for MPS patients should include which of the following routine studies
 - A. echocardiography
 - B. electroencephalography
 - C. brain MRI
 - D. barium swallow
 - E. liver function studies