Radiographic expression of the mucopolysaccharidoses.

Dysostosis Multiplex.

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Introduction

The mucopolysaccharidoses (MPS) have in common a quite specific radiological expression, which is termed “dysostosis multiplex.” This poster presentation covers the entire skeletal system to show the spectrum of dysostosis multiplex changes suggesting the presence of MPS disease in the individual. Also, several previously unreported dysostosis multiplex findings are shown.

Background

All types of MPS disease exhibit dysostosis multiplex to a greater or lesser degree. The role of the radiologist and other clinicians is to recognize any of these diverse changes on radiographs so that the patient can be tested to ascertain which MPS disease is present for treatment, genetic counseling, and management. It is suspected that perhaps certain mildly afflicted individuals go through life asymptomatic but undiagnosed. The diagnosis is usually suggested by a combination of “clinical” and radiographic features. The radiological manifestations play an extremely important role.

Dysostosis multiplex changes are widespread in the skeletal system and involve many different bones. At times even the “routine” chest film will reveal changes suggesting the diagnosis of possible MPS disease.

The following mucopolysaccharidoses manifest dysostosis multiplex:

- MPS I-H (Hunter)
- MPS I-S (Scheie)
- MPS II (Hunter)
- MPS III A, B, C (Sanfilippo)
- MPS IV A, B (Morquio)
- MPS V (Maroteaux-Lamy)
- MPS VI (Maroteaux-Lamy)
- MPS VII (Sly)

Dysostosis multiplex is also a feature of mucolipidoses and other storage diseases, including:

- Mucolipidoses (ML):
  - ML I (I-cell disease/Leroy)
  - ML II (Pseudo-Hunter Polydystrophy)
- Other storage diseases:
  - Multiple Sulfatase Deficiency
  - Carbohydrate-Deficient Glycoprotein Syndrome
  - GM1 Gangliosidosis
  - Geleophysic Dysplasia

The following patient images show how dysostosis multiplex can affect the outward appearance of the patient.

Radiological Findings - Dysostosis Multiplex

Familiarity with the radiological findings of dysostosis multiplex will allow radiologists and other clinicians to identify patients who may have mucopolysaccharidoses and other storage diseases so that these patients can be tested and referred for treatment and counseling.

Figures 1 through 15 illustrate the radiographic appearance of dysostosis multiplex in multiple areas of the skeletal system in patients affected with mucopolysaccharidoses.

- **Skull**
  - Abnormal J-shaped sella
  - Thickened diploic space

- **Thorax**
  - Short, thick clavicles
  - Paddle [oar-shaped] ribs

- **Spine**
  - Superiorly notched [inferiorly beaked] vertebral bodies
  - Middle beaked in MPS IV (Morquio)
  - Posterior scalloping

- **Pelvis**
  - Rounded iliac wings
  - Iliac, tapered inferiorly

- **Long Bones**
  - Mildly hypoplastic epiphyses [often generalized]
  - Long/narrow femoral neck
  - Hypoplastic/fragmented CFU
  - Thick [short] diaphyses
  - Proximal humeral notching

- **Hands**
  - Metacarpals: Proximal pointing
  - Metacarpals: Thick, short, with thin cortices
  - Carpals: Irregular, hypoplastic
  - Tarsal bones: Irregular contours

Enzyme Replacement Therapy (ERT) for MPS VI

A new era in the treatment of the mucopolysaccharidoses (MPS) has occurred with the advent of enzyme replacement therapy (ERT). A case in point is the new treatment Naglazyme® (galsulfase), introduced in 2005 by BioMarin Pharmaceutical Inc., specifically for the treatment of MPS VI. It is apparent that the earlier this therapy is initiated the better the expected results. Therefore it becomes imperative that the clinician (geneticist, pediatrician, radiologist, etc.) determine the diagnosis as early as possible.