



## Images in Clinical Medicine

## Mucopolysaccharidosis

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**Fig. 1.** Chest radiography shows widened ribs and coarse trabeculation and heterogeneous bone density. The clavicles are short and the vertebral bodies are flat.

A 24-year-old woman with asthma had chest radiography (Fig. 1) in the outpatient department. The examination revealed widened ribs and coarse trabeculation and heterogeneous bone density. Shortening of the clavicles and flattening of the vertebral bodies were also noted. A review of the patient's chart revealed a prior diagnosis of mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome).

The mucopolysaccharidoses are a heterogeneous group of lysosomal storage diseases caused by deficiency of enzymes that degrade glycosaminoglycans. The diseases are autosomal recessive conditions and cause accumulation of abnormal metabolite substrates that are deposited in the bone marrow and multiple viscera.

Radiographic findings vary. The key findings include a dorso-lumbar gibbus with anteriorly beaked vertebral bodies; wide, oar-shaped ribs; short, thick clavicles; small, tapered ilia; a steep acetabular roof; and coxa valga.

The differential diagnosis includes Legg-Calve-Perthes disease, spondyloepiphyseal dysplasia, multiple epiphyseal dysplasia, and other metabolic diseases, such as Gaucher disease and Niemann-Pick disease.

## Further reading

- [1] Glass RBJ, Norton KI, Mitre SA, Kang E. Pediatric ribs: a spectrum of abnormalities. *RadioGraphics* 2002;22:87–104.
- [2] Donnelly AF. *Diagnosis imaging: pediatrics*. 1st ed. 6. Utah: Amirsys; 2005. p. 118–21.
- [3] Resnick D, Kransdorf MJ. *Bone and joint imaging*. 3rd ed. Philadelphia: Elsevier and Saunders; 2005. p. 1318–21.

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