Teaching NeuroImages: Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome) in a previously undiagnosed infant

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Figure Imaging features of Maroteaux-Lamy syndrome

On plain films (A) and sagittal T2 (B), the L1 vertebra is hypoplastic, causing kyphosis. Sagittal FIESTA (C) shows CSF attenuation (white arrows), indicating craniovertebral stenosis. The odontoid process is noted to be nondysplastic (dots). Axial T1 (D) at C2 demonstrates dural thickening (arrows), due to glycosaminoglycans deposition.

A 10-month-old boy presented with mild developmental delay and thoracolumbar kyphosis. This was partially correctable, with no neurologic deficit. Neuroimaging (figure) showed characteristic features of type VI mucopolysaccharidosis.¹ Biochemical analysis provided definitive diagnosis (arylsulfatase-B enzyme activity <10% of normal).²

Maroteaux-Lamy syndrome is a storage disease, where glycosaminoglycans accumulate within the lysosomes. Dural deposition leads to craniovertebral stenosis. While dural thickening is seen in granulomatous diseases and pachymeningeal metastasis, these are rare in infancy. The spinal findings of mucopolysaccharidosis VI resemble those of mucopolysaccharidosis IV, but in the latter, odontoid hypoplasia with instability allows a distinction to be made.

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