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TYPICAL IMAGING FEATURES IN HUNTER'S SYNDROME

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Background: A 17-year-old boy with known Hunter's syndrome was admitted to the emergency department after an episode of generalized seizures. The episode of seizures was probably related to medication. He had a characteristic coarse facial appearance and macrocrania. There was no mental retardation, dwarfism nor deafness.

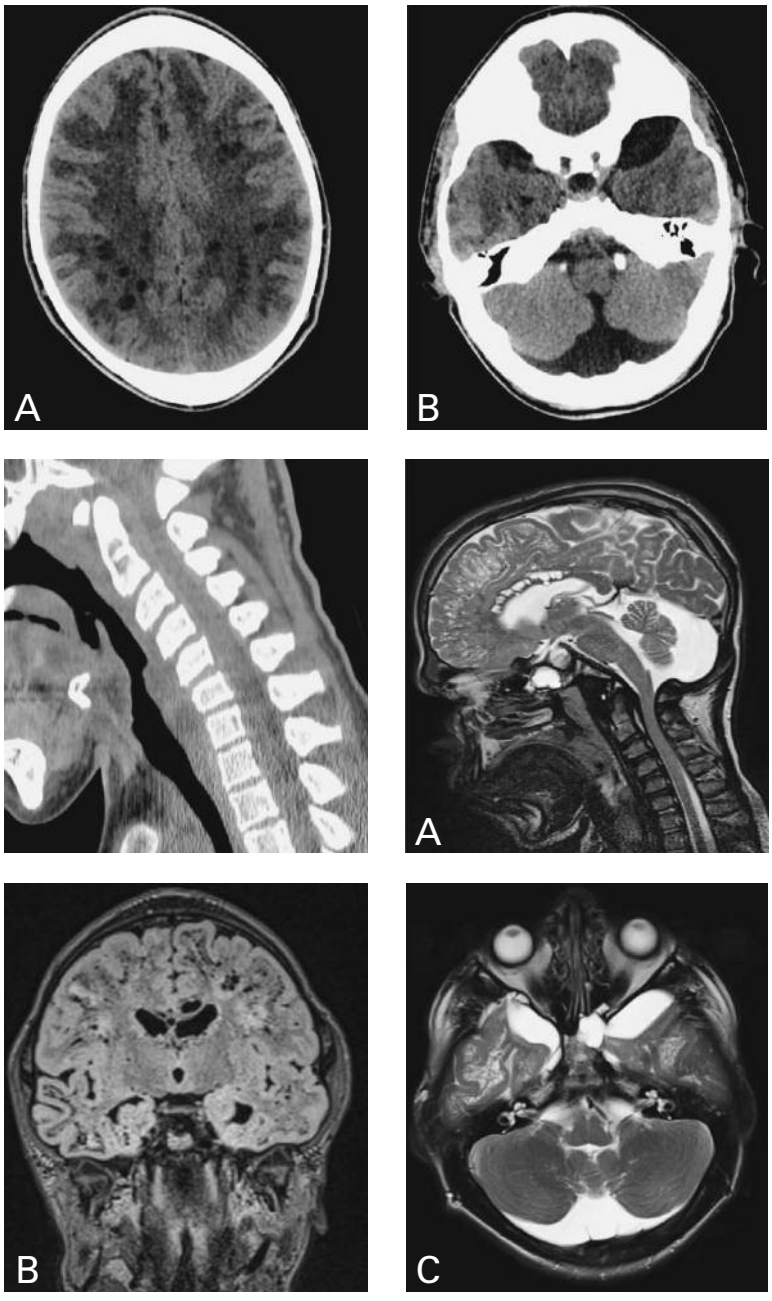

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Fig.		
	1A	1B
	2	3A
	3B	3C

Work-up

CT scan of the brain (Fig. 1) shows on A multiple cyst-like hypodense lesions in the subcortical and deep white matter. There is marked thickening of the calvarium. Section through frontal and temporal lobes (B) shows the same small hypodense lesions in the white matter. Bilateral temporal arachnoid cysts and megacisterna magna can be seen. Note macrocrania and thickening of the skull.

CT scan of the cervical spine, sagittal MPR image (Fig. 2), demonstrates characteristic thickening of the posterior longitudinal ligament in the upper cervical spine.

MRI of the brain (Fig. 3), sagittal T2-weighted MR-image (A), shows cribriform hyperintense lesions are noted in the subcortical and deep white matter, with extensive involvement of the corpus callosum. The lesions show signal intensities equal to cerebrospinal fluid at all sequences and represent dilated perivascular spaces of the white matter. Also note megacisterna magna. On coronal FLAIR-image (B), dilated perivascular spaces are seen throughout the white matter. Note signal intensities of these cyst-like lesions equal to cerebrospinal fluid. On axial T2-weighted image (C), bilateral anterior temporal arachnoid cysts are seen as well as megacisterna magna. Cribriform changes in the temporal white matter are obvious.

Radiological diagnosis

The numerous cyst-like lesions in the subcortical and deep white matter, with CSF density on CT scan and CSF-intensity on MRI, the macrocrania and thickening of the skull and posterior longitudinal ligament in the spine constitute the typical findings of *Hunter's syndrome* (mucopolysaccharidosis type II).

Discussion

The presented case illustrates the clinical and imaging findings typical of Hunter's syndrome (mucopolysaccharidosis II). The mucopolysaccharidoses are a hetero-geneous group of inherited lysosomal storage disorders characterized by failure to degrade glycosaminoglycans or mucopolysaccharides. All mucopolysaccharidoses (MPS) are autosomal recessive except Hunter, which is X-linked. MPS-patients have a variable degree of phenotype presentation (depending on enzyme dysfunction), ranging from mild to severe. Our patient has the mild form of Hunter's syndrome with a typical facial appearance as predominant clinical finding.

The brain findings in patients with MPS are of two types. Dilated perivascular spaces, megacisterna magna, arachnoid cysts and communicating hydrocephalus result from impaired reabsorption of cerebrospinal fluid, presumably due to build-up of glycosaminoglycans in the leptomeninges with arachnoid membrane destruction and a ball-valve effect. Dilated perivascular spaces in patients with MPS were previously falsely believed to be due to macroscopic deposition of mucopolysaccharides. The presence of multiple dilated perivascular spaces in the corpus callosum is a classic feature of MPS. The other spectrum of findings in Hunter's syndrome primarily affects the brain parenchyma and consists of white matter T2 hyperintensities and atrophy. The latter findings are not prominent in our patient.

Other characteristic imaging features of Hunter's syndrome seen in the presented case include macrocrania and thickening of the skull. Thickening of the posterior longitudinal ligament typically occurs in the upper cervical region and is also nicely illustrated in this case.

In conclusion, MPS should be considered when dilated perivascular spaces with predominant involvement of the corpus callosum are observed. Other findings, such as macrocrania and thickened skull should be looked at to raise confidence in suggesting the diagnosis if it is not already known. The severe form of Hunter's syndrome presents in the early years but the mild form might be diagnosed as late as the teenage, with characteristic coarse facial appearance as the only presenting symptom and absence of other typical findings such as deafness and dwarfism.

Bibliography

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