Intracranial findings of Hunter syndrome

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A 13-year-old boy presented with mental retardation, progressive hearing loss and coarsened facial features. Brain MRI demonstrated punctate and oblong CSF-equivalent cystic areas within the corpus callosum, basal ganglia and periventricular white matter (Figs. 1 and 2), with surrounding hyperintensity on T2-W and FLAIR images (Fig. 1). There is mild lateral ventriculomegaly (Figs. 1 and 2). Both the boy and his mother were mutation carriers for mucopolysaccharidosis type II (MPS II, Hunter syndrome).

Fig. 1 Axial FLAIR image

Fig. 2 Coronal T2-W image

Hunter syndrome is an X-linked recessive lysosomal storage disorder [1]. Additional clinical associations include skeletal dysplasia, hepatosplenomegaly, recurrent respiratory infections and thickening of heart valves [1, 2]. Characteristic intracranial findings include innumerable cysts within the periventricular white matter, corpus callosum and basal ganglia with a “cribriform,” “sieve-like,” or “honeycomb” appearance, from glycolipid and glycosaminoglycan buildup in Virchow-Robin perivascular spaces. Additional findings include white matter hyperintensity on T2-W and FLAIR images, hydrocephalus, and dural thickening [2].

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