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Optic Canal Narrowing in Hunter Syndrome (Mucopolysaccharidosis II)

I read with interest the article "Brain and Spinal MR Imaging Findings in Mucopolysaccharidoses: A Review"[1] and would like to illustrate optic canal narrowing in a patient with Hunter syndrome (mucopolysaccharidosis [MPS]-II).

A 6-year-old boy presented with significant loss of vision. He had been evaluated (at 3 years of age) for mucopolysaccharidosis; clinical phenotype, heparin sulfate in urine electrophoresis, and plasma enzyme levels (iduronate 2-sulfatase level: 4.2 nmol/h by fluorimetric assay by using 4-methyl umbelliferone; normal range: 167–475 nmol/h; control: 194.3) were suggestive of Hunter syndrome.

MR imaging of the brain showed periventricular white matter hyperintensities (Fig 1*A–C*), enlarged Virchow-Robin spaces (Fig 1*A*, *-B*), ventriculomegaly, and atrophy (Fig 1*A–C*, *-E*, *-F*), known features of mucopolysaccharidosis. The bilateral optic canals were symmetrically narrowed (Fig 1*D–F*), and hyperintensities were noted around both optic nerves (Fig 1*C–F*).

Hunter syndrome, or mucopolysaccharidosis II, is an X-linked progressive lysosomal storage disease due to deficiency of the lysosomal enzyme iduronate-2-sulfatase. Known ocular manifestations of Hunter syndrome include exophthalmos, hypertelorism, pigmentary retinopathy, uveal effusion, optic nerve head swelling (disc edema,[2] possibly due to compression by scleral deposits), and optic atrophy.

Optic canal narrowing is a known feature of osteopetrosis,[3] and fibrous dysplasia and has been described in familial hypophosphatemic rickets, carbonic anhydrase type 2 deficiency (which includes osteopetrosis), and MPS-4.[4] It has not been previously reported in Hunter syndrome. Perhaps optic canal narrowing is underdiagnosed in Hunter syndrome because intellectually impaired children may not be able to describe visual loss.

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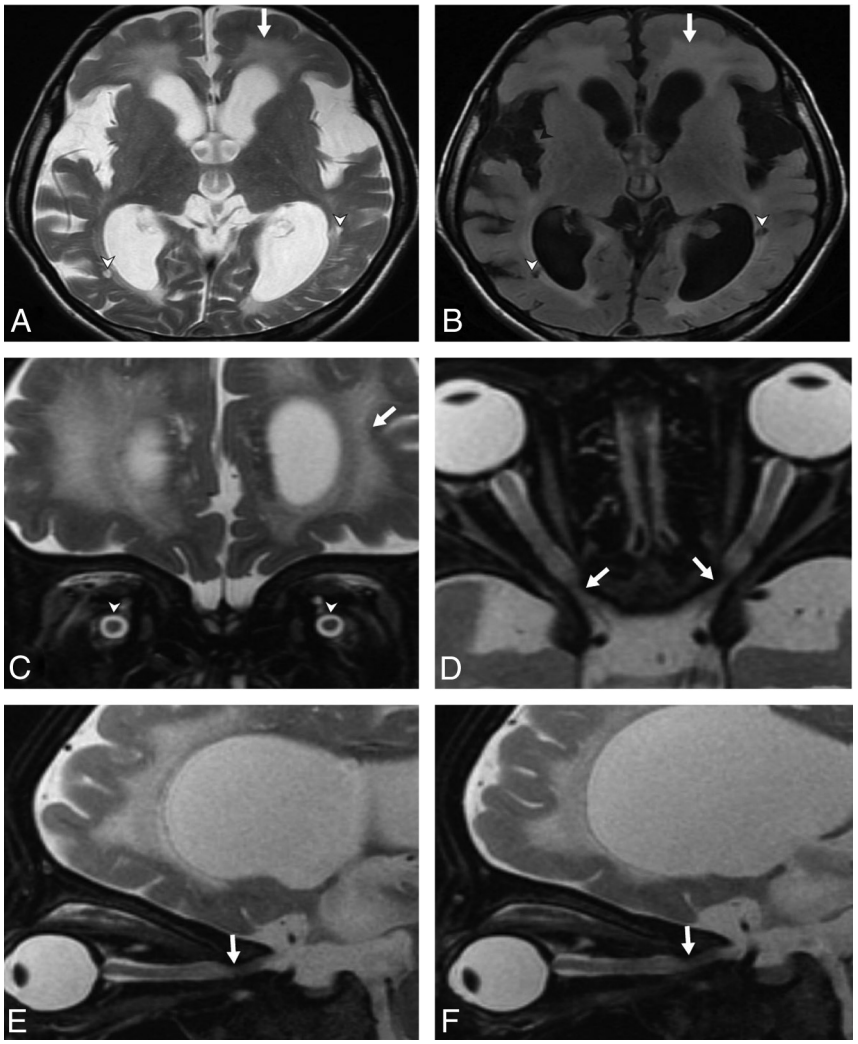

FIG 1. Axial T2 (*A*) and axial T2 FLAIR (*B*) images show periventricular white matter hyperintensities (*arrows*), prominent Virchow-Robin spaces (*arrowheads*), ventriculomegaly, and brain atrophy. *C*, Coronal T2 image shows hyperintensities in periventricular white matter (*arrows*) and perioptic spaces (*arrowheads*), ventriculomegaly, and brain atrophy. Axial T2 (*D*) and right (*E*) and left (*F*) sagittal T2 images show narrowing of the optic canals (*arrows*), hyperintensities in perioptic spaces, and brain atrophy.