Teaching NeuroImages: Macrocerbellum and optic atrophy in a young boy

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Figure Neuroimaging findings in mucopolysaccharidosis type I

Sagittal T1-weighted (A) and T2-weighted (B) brain MRI disclose marked enlargement of the cerebellum without other posterior fossa anomalies. Axial T2-weighted (C) and T1-weighted (D) brain MRI show a macrocerebellum filling the posterior fossa and mild optic atrophy (arrow).

A 2-year-old boy presented with delayed motor skills and language since birth. Family history disclosed consanguineous parents. Examination showed global muscular hypotonia, optic atrophy, oculomotor apraxia, and normal head circumference. Brain MRI showed optic atrophy and macrocerebellum (figure). Laboratory tests revealed deficient activity of α -L-iduronidase in peripheral blood leukocytes and increased glycosaminoglycan urinary excretion, indicating mucopolysaccharidosis type I.

Macrocerbellum is a rare condition characterized by an abnormally large cerebellum sparing the overall shape and has been described in conditions such as Sotos syndrome, Costello syndrome, Williams syndrome, Alexander disease, fucosidosis, and Lhermitte-Duclos disease,¹ and can rarely be present in mucopolysaccharidosis.²

AUTHOR CONTRIBUTIONS

Dr. Pinto: case report project conception, case report project organization, case report project execution, writing of the first draft. Dr. Souza: case report project conception, case report project organization, case report project execution, writing of the first draft. Dr. Batistella: case report project execution, writing of the first draft. Dr. Oliveira: review and critique.

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