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.

Macrocerebellum 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, and can rarely be present in mucopolysaccharidosis.

**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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**REFERENCES**

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