MRI diagnosis in Metachromatic leukodystrophy: A case report.

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Abstract:

Leukodystrophies encompass a wide spectrum of inherited neurodegenerative disorders affecting white matter of central nervous system. There are distinctive clinical, biochemical, pathologic, and radiologic features of each leukodystrophies. Magnetic resonance imaging is primary imaging modality in the identification of underlying white matter abnormalities, to monitor the progression and the response to therapy. Metachromatic leukodystrophy (MLD) is a rare group of inherited, lysosomal storage disease characterized by intra-lysosomal accumulation of sphingolipid sulfatides due to reduction or complete deficiency of Arylsulfatase-A enzyme necessary for normal myelin sheath formation. Diagnosis is usually suspected on MRI of brain and confirmed by enzyme assays. Hematopoietic stem cell transplantation in infancy has shown to delay the progression of disease, making early diagnosis very imperative. We present a case of 5 years old male child of metachromatic leukodystrophy, presented with regression of milestone and progressive spasticity, who underwent MRI brain and diagnosis was confirmed by enzyme assay.

Key Words: Arylsulfatase A deficiency, Magnetic resonance imaging (MRI), Metachromatic leukodystrophy (MLD).

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