Magnetic resonance imaging traits may help to differentiate Pelizaeus-Merzbacher and Pelizaeus-Merzbacher-like disease

Características em ressonância magnética podem auxiliar a diferenciar doença de Pelizaeus-Merzbacher e Pelizaeus-Merzbacher-like

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A six-year-old boy with nystagmus and developmental delay from six months of age, was born from healthy first-degree cousins. On neurological examination, he presented with spontaneous and multidirectional nystagmus, gait and upper limb ataxia, and lower limb hyperreflexia/spasticity. Brain magnetic resonance disclosed a hypomyelinating pattern (Figure). Pelizaeus-Merzbacher disease was considered, although there was unusual pontine dysmyelination. Pelizaeus-Merzbacher disease was excluded as the PLP1 gene sequencing was negative. Pelizaeus-Merzbacher-like disease was considered, and a homozygous mutation in GJC2 gene (c.217C > A p.[P73T]) was found. Pontine dysmyelination is not usually described in Pelizaeus-Merzbacher disease and may be a clue to Pelizaeus-Merzbacher-like disease¹².

Figure. Magnetic resonance features of Pelizaeus-Merzbacher-like disease: (A) Diffuse T2 hyperintensity (hollow arrow) and (B) T1 slight hypointensity of cerebral white matter (arrowhead), characterizing hypomyelination; and (C) pontine T2 hyperintensity (white arrow), highly compatible with Pelizaeus-Merzbacher-like disease².

References


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