

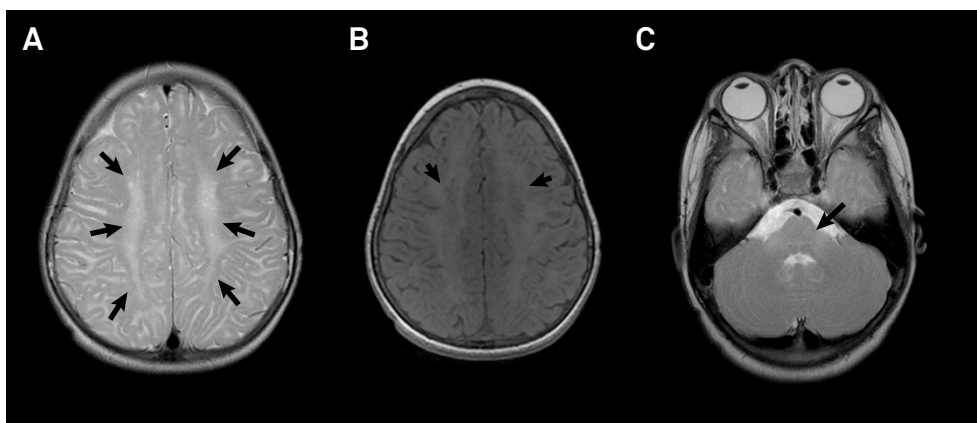
# Magnetic resonance imaging traits may help to differentiate Pelizaeus-Merzbacher and Pelizaeus-Merzbacher-like disease

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

Matheus Augusto Araújo CASTRO<sup>1</sup>, Pedro Henrique Almeida FRAIMAN<sup>1</sup>, Clecio de Oliveira GODEIRO-JUNIOR<sup>1</sup>

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 disease<sup>1,2</sup>.






**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 disease<sup>2</sup>.

## References

1. Akinaga K, Lee TY, Montenegro MA. Pelizaeus Merzbacher disease: dysmyelination versus demyelination. *Arq Neuropsiquiatr*. 2016 Jan;74(1):81. <https://doi.org/10.1590/0004-282X20150170>
2. Steenweg ME, Vanderver A, Blaser S, Bizzi A, Koning TJ, Mancini GM, et al. Magnetic resonance imaging pattern recognition in hypomyelinating disorders. *Brain* 2010 Oct;133(10):2971-82. <https://doi.org/10.1093/brain/awq257>

<sup>1</sup>Universidade Federal do Rio Grande do Norte, Departamento de Medicina Integrada, Natal RN, Brasil.

Matheus Augusto Araújo Castro  <https://orcid.org/0000-0003-0335-8799>; Pedro Henrique Almeida Fraiman  <https://orcid.org/0000-0002-5250-8602>; Clecio de Oliveira Godeiro-Junior  <https://orcid.org/0000-0002-4312-1633>

**Correspondence:** Clecio Godeiro-Junior; Av. Nilo Peçanha, S/N – Petropolis; 59012-300 Natal RN, Brasil; E-mail: clecijunior@yahoo.com.br

**Conflict of interest:** There is no conflict of interest to declare.

Received 23 January 2019; Received in final form 19 February 2019; Accepted 10 March 2019.