

Pantothenate Kinase Associated Neurodegeneration in Two Brothers

Neurodegeneração Associada a Pantotenato Quinase em Dois Irmãos



Bruno Niemeyer de FREITAS RIBEIRO¹, Tiago MEDINA SALATA², Lúvia de OLIVEIRA ANTUNES², Beatriz SALATA ANTUNES³
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Palavras-chave: Doenças Neurodegenerativas; Fosfotransferases (Aceptor do Grupo Álcool); Irmãos.

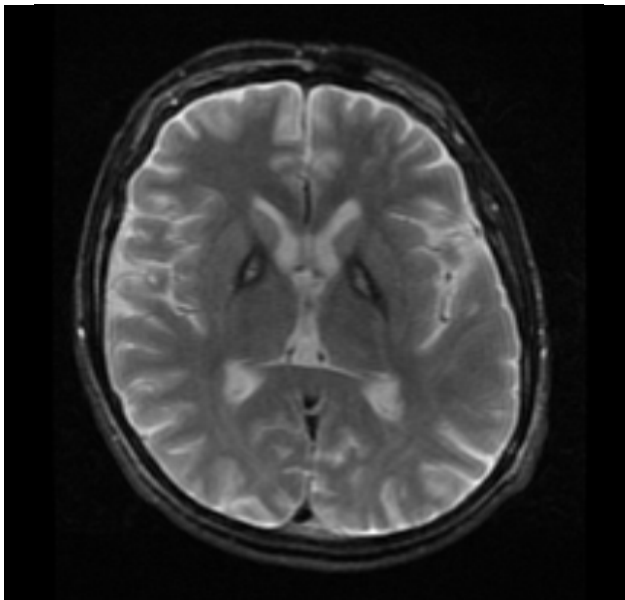


Figure 1 - Axial T2-weighted MR image demonstrates bilateral hyperintense pallidal areas on background areas of T2 shortening (eye-of-the-tiger sign)

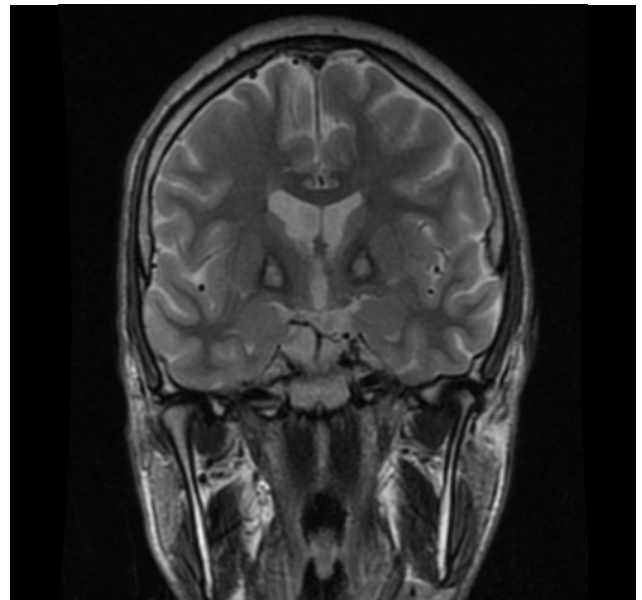


Figure 2 - Coronal T2-weighted MR image demonstrates bilateral hyperintense pallidal areas on background areas of T2 shortening (eye-of-the-tiger sign)

Two male brothers, aged 28 and 26 years, children of outbred parents, presented neurological symptoms. The oldest developed progressive spasticity, dysarthria, and aggressiveness for the previous four years. Concurrently his brother showed orofacial dyskinesia and cognitive decline. Family history was unremarkable. Laboratory testing was normal, including ceruloplasmin, ferritin, syphilis serology and viral markers (HIV, HBV, HCV, HTLV-1); the peripheral blood smear was negative for acanthocytes. Ophthalmoscopy was normal. Brain magnetic resonance imaging (MRI) revealed the 'eye of the tiger' sign on coronal and axial T2-weighted

images in both cases (Fig. 1 older brother; Fig. 2, younger brother). Molecular genetic testing revealed the G521R mutation in the PANK2 gene.

Pantothenate kinase-associated neurodegeneration (PKAN) is a rare autosomal recessive disorder, and a form of neurodegeneration with brain iron accumulation (NBIA).¹⁻³ PKAN cases can be either classical, with onset in the first decade, or atypical, with late-onset, both presenting extrapyramidal dysfunction.^{1,3} The 'eye of the tiger' sign is the characteristic MRI finding.^{1,4}

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1. Clinic 3D Diagnóstico por Imagem. Hospital Casa de Portugal. Rio de Janeiro. Brasil.

2. Department of Radiology. Hospital Casa de Portugal. Rio de Janeiro. Brasil.

3. Universidade do Grande Rio. Rio de Janeiro. Brasil.

✉ Autor correspondente: Bruno Niemeyer de Freitas Ribeiro. bruno.niemeyer@hotmail.com.

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Bruno Niemeyer de FREITAS RIBEIRO, Tiago MEDINA SALATA, Livia de OLIVEIRA ANTUNES,
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Tel: +351 218 428 215

E-mail: submissao@actamedicaportuguesa.com

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