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Retinitis pigmentosa in pantothenate kinase-associated neurodegeneration

Retinose pigmentar na neurodegeneração associada à pantotenato quinase

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A 16-year-old boy presented to our hospital with 4-year-history of generalized dystonia (predominantly cranio-cervical and upper limbs) (Figure 1) and visual loss. Brain MRI revealed globus pallidus hypointensity with central hyperintense signal (eye-of-the-tiger) (Figure 2). Retinitis pigmentosa was observed in ophthalmologic evaluation (Figure 3). Genetic test confirmed mutation in PANK2 gene.

Pantothenate kinase-associated neurodegeneration (PKAN) is classically characterized by early-onset dystonia and pyramidal signs but other features may include parkinsonism, choreoathetosis and dementia[1]. Brain MRI typically depicts the eye-of-the-tiger pattern[1]. When retinitis pigmentosa, an unusual finding[3], is observed in the clinical spectrum of PKAN, we must consider variants[2,3,4], such as HARP syndrome (hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration).

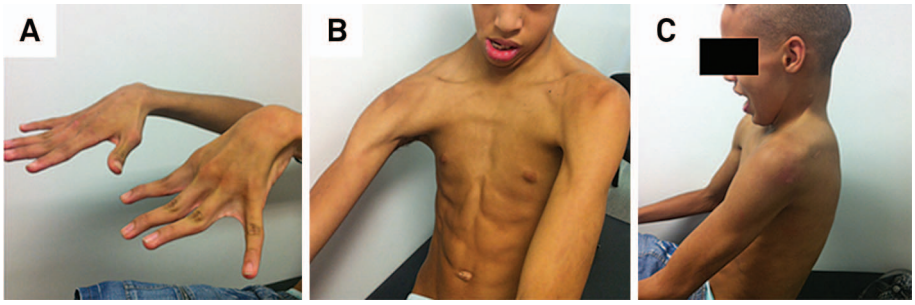

Figure 1. Dystonia in upper limbs (A). Note marked dystonia involving cranio-cervical segment and facial "grimacing" (B and C).

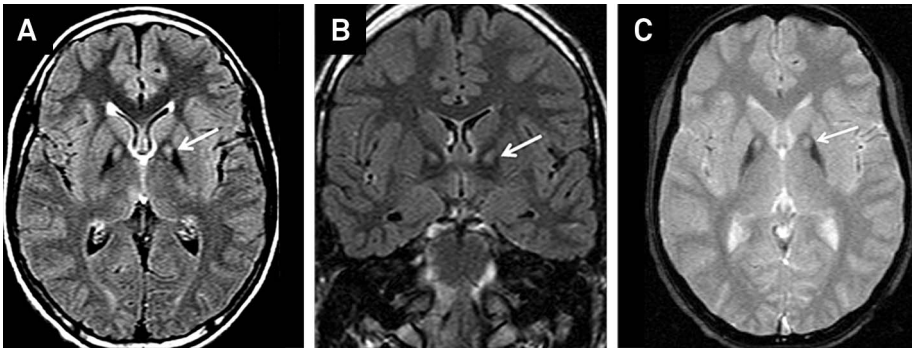

Figure 2. Axial FLAIR (A), coronal FLAIR (B) and spin echo (C) sequences brain MRI disclose marked hypointense signal of the globus pallidus with central hyperintense signal (eye-of-the tiger appearance) (arrows).

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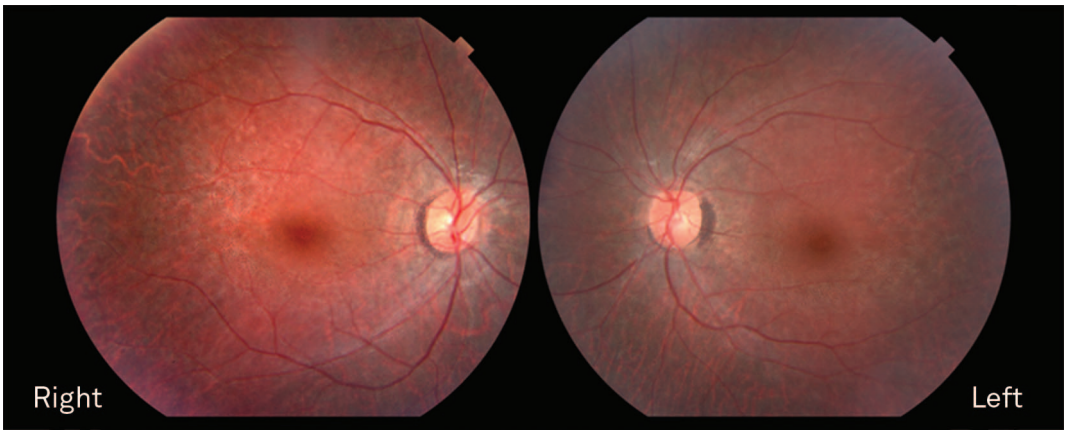

Figure 3. Note grainy appearance of the retinal pigmented epithelium, with fine dots, arteriolar thinning and peripapillary pigmentation on the temporal optic disc border. Those are mild signs of retinal degeneration, due to early retinitis pigmentosa.

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