

Septo-optic dysplasia with late-onset seizure: MRI and ophthalmological features

Displasia septo-óptica com epilepsia tardia: neuroimagem e alterações oftalmológicas

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A 31-year-old man presented with tonic-clonic seizures, started at 15-year-old, that improved with carbamazepine. Fundoscopy disclosed small optic discs and arterial tortuosity. Neurological examination and visual acuity were normal. Brain MRI showed hypoplasia of the optic nerves, absence of septum pellucidum and polymicrogyria (Figure 1). Optical coherence tomography showed bilateral

retinal nerve fiber layer thinning (Figure 2). Septo-optic dysplasia was diagnosed.

Septo-optic dysplasia is usually reported in children, and is characterized by optic nerve hypoplasia, absent septum pellucidum, cortical malformations and pituitary dysfunction¹. This patient had no endocrinopathy, and presented with late onset epilepsy, which is unusual in septo-optic dysplasia^{1,2}. The typical MRI changes and optic abnormalities were the clues for the diagnosis².

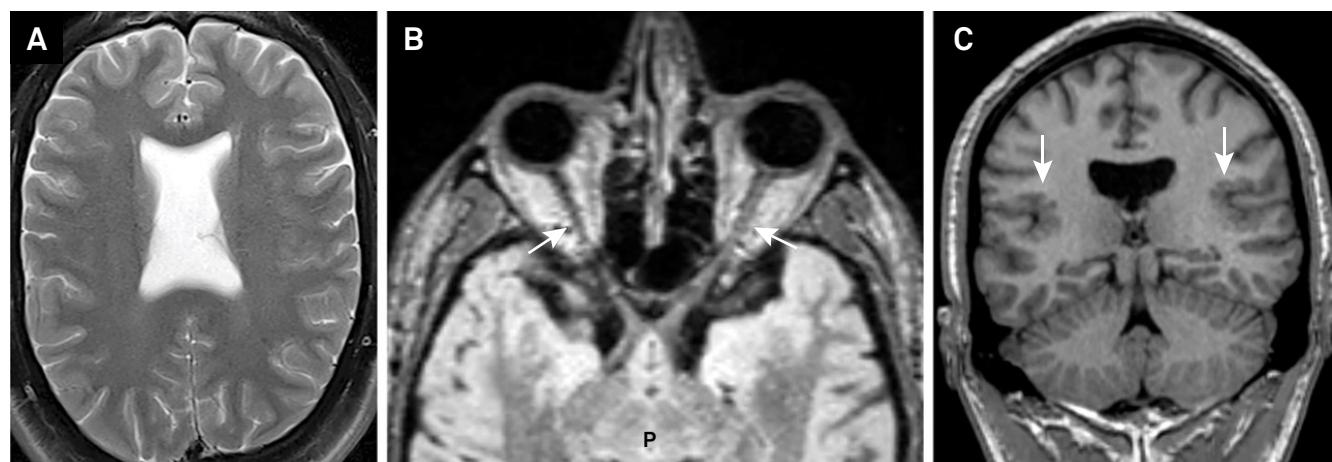


Figure 1. (A) Axial T2-weighted image shows absence of septum pellucidum. (B) Axial FLAIR sequence demonstrates hypoplasia of the optic nerves, more marked on the right (arrows). (C) Coronal T1-weighted image shows absence of septum pellucidum and polymicrogyria in posterior sylvian fissures (arrows).

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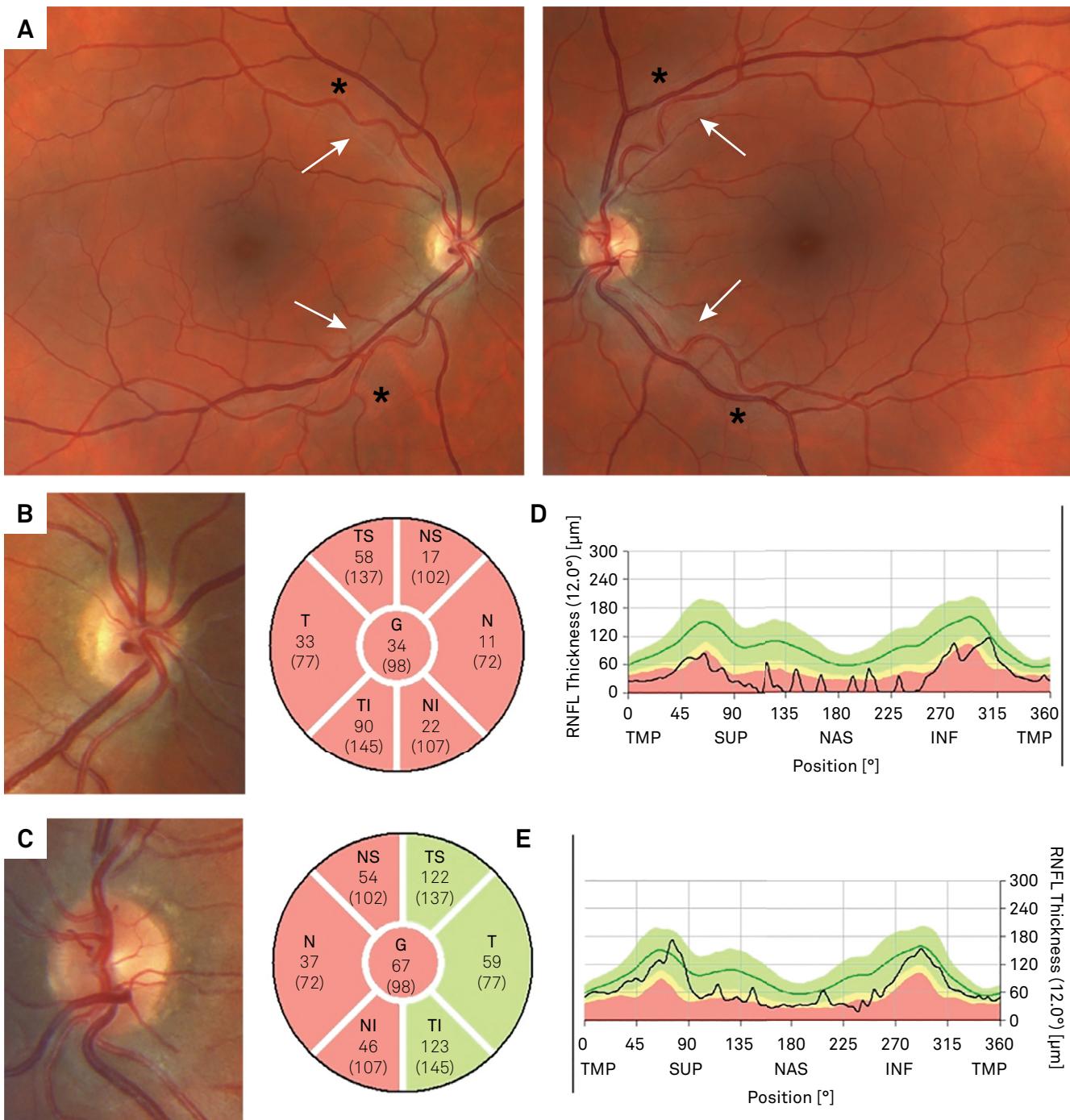


Figure 2. A: Retinography shows hypoplastic optic disc in OD and small disc in OS, arterial tortuosity (asterisks) and reduced visibility of the retinal nerve fibers (arrows) bilaterally. B and C: Absent cup in OD and bilateral temporal pallor. D and E: Thickness maps and graphs show retinal nerve fiber layer thinning in both eyes.

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