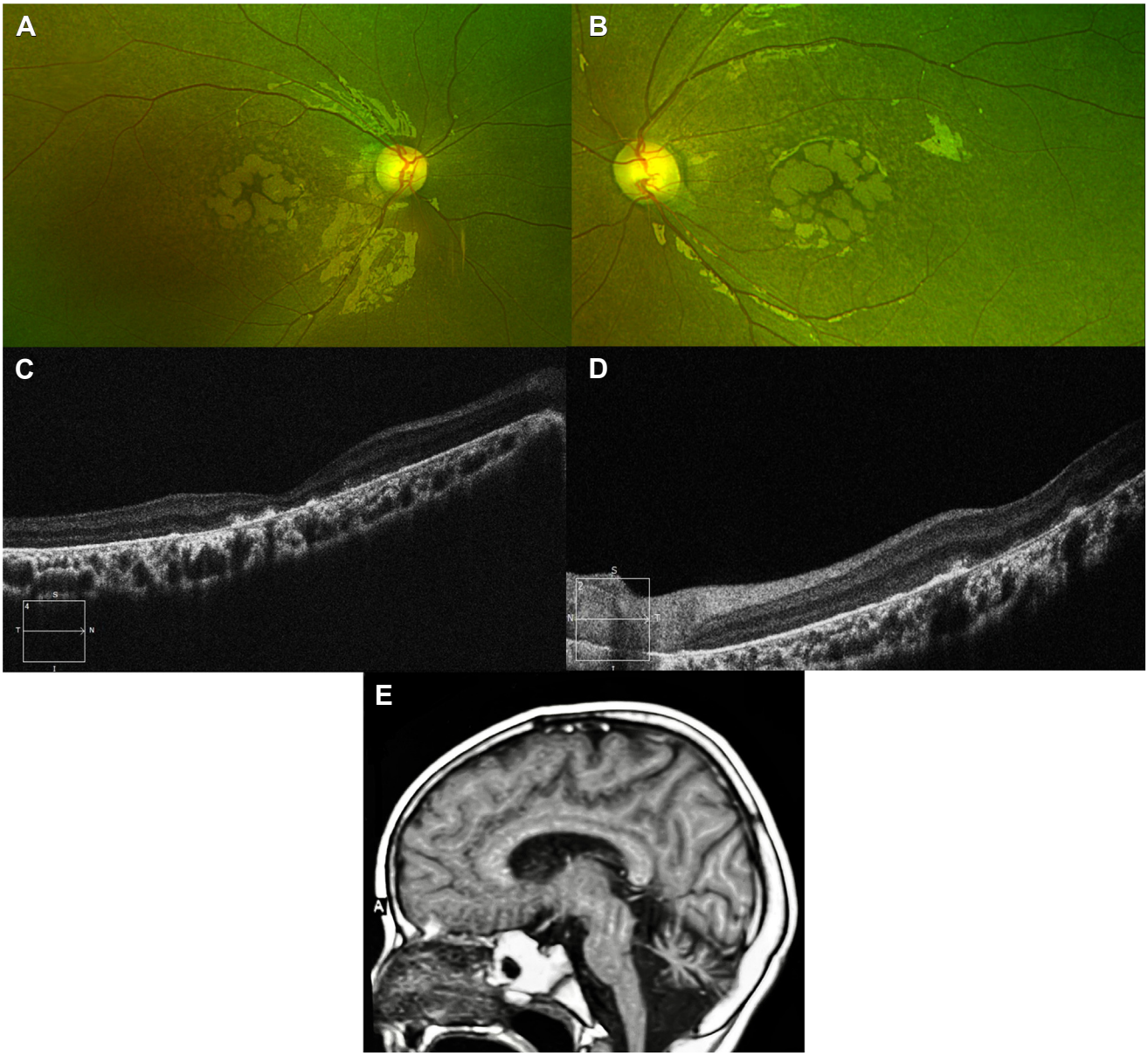

Pigmentary Retinopathy in Spinocerebellar Ataxia Type 7

The fundus examination of a 5-year-old girl with neuroregression symptoms and unremarkable family history revealed vascular attenuation and patchy pigment epithelial atrophy at the macula with intervening patches of pigment epithelium in both eyes (**A**, **B**). OCT evaluation revealed focal hyperreflective loci of clumped residual pigment epithelial cells in both eyes (**C**, **D**). Magnetic resonance imaging revealed diffuse cerebellar atrophy and prominent gyro-sulcal formations in the cerebral hemispheres (**E**). Genetic testing revealed 120 cytosine-adenine-guanine repeats at the second allele of the Ataxin 7 gene, confirming the diagnosis of full penetrant spinocerebellar ataxia type 7. The prognosis was explained, and the child continues to remain on follow-up. (Magnified version of Figure **A**-**E** is available online at www.ophthalmologyretina.org).

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