

# Aniridia as a clue for the diagnosis of Gillespie syndrome

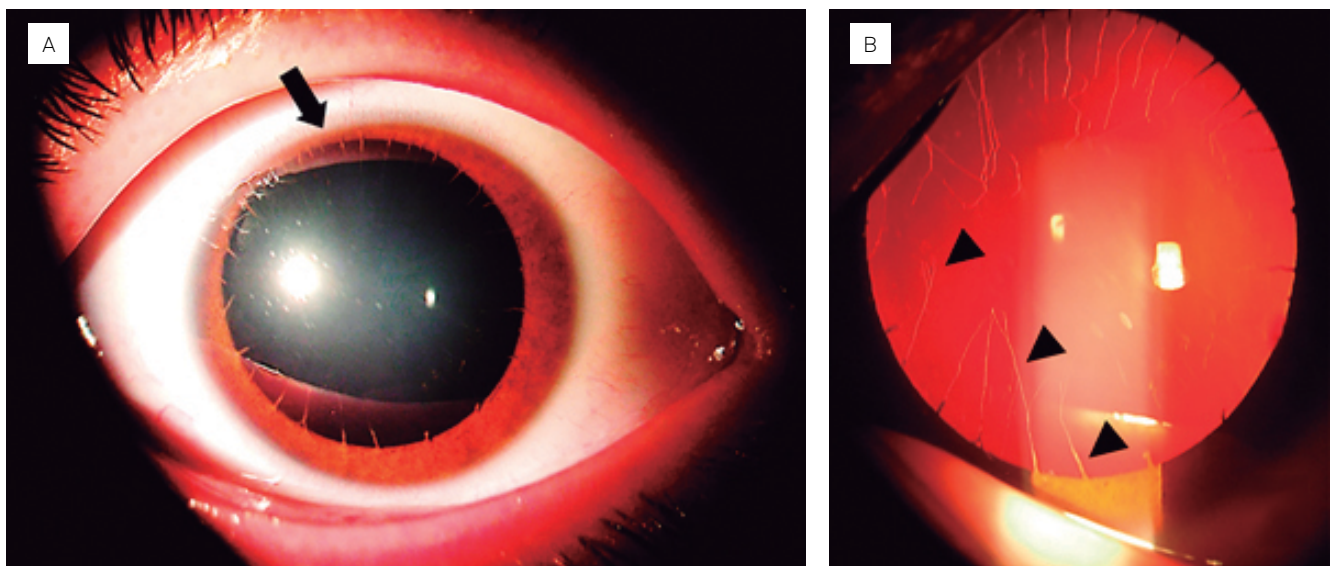
Aniridia: dica clínica para o diagnóstico da síndrome de Gillespie

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A 23-year-old woman presented with global developmental delay and non-progressive ataxia since birth. Parents were consanguineous. On examination, she had dysarthria, nystagmus and ataxia. Ophthalmological evaluation showed absence of part of the iris or aniridia (Figure 1) and magnetic resonance imaging (MRI) pointed mild cerebellar atrophy. Genetic testing revealed

homozygous mutations in *ITPR1* gene and confirmed Gillespie syndrome.

Absence of part or all the iris is called aniridia. This is a distinct ophthalmological feature observed in a restrict group of genetic diseases<sup>1</sup>. The triad of aniridia, congenital ataxia and intellectual disability characterize Gillespie syndrome, a recessive or dominant disease caused by *ITPR1* gene mutations<sup>2</sup>.








**Figure 1.** Ophthalmological findings in a 23-year-old woman with Gillespie syndrome. (A) Photograph of the right eye. (B) Slit-lamp examination. A thin rim of iridal tissue (arrow) results in a fixed and dilated appearance of the pupil, consistent with iris hypoplasia (A). Iridolenticular strands occurring at irregular intervals (arrowheads) constitute remnants of the pupillary membrane (B).

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