

# Eyelid retraction is not a pathognomonic sign of Machado-Joseph disease in the context of spinocerebellar ataxias

Retração palpebral não é um sinal patognomônico da doença de Machado-Joseph no contexto das ataxias espinocerebelares

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We describe three patients with spinocerebellar ataxia (SCA) and marked eyelid retraction: A 33-year-old woman with ataxia, pyramidal signs and eyelid retraction; genetic test confirmed SCA1 (Figure A). A 41-year-old man with ataxia, ophthalmoplegia, neuropathy and eyelid retraction; genetic test confirmed SCA2 (Figure B). A 68-year-old man with ataxia, neuropathy, nystagmus and eyelid retraction; genetic test confirmed SCA3 or Machado-Joseph disease (MJD) (Figure C).

Patients with SCA, other than MJD, in special SCA1, SCA2 and SCA10, may rarely present with eyelid retraction<sup>1,2,3</sup>. This neurological feature presumes a more widespread degenerative process involving the midbrain<sup>3</sup>. SCA patients with eyelid retraction and negative genetic test for MJD should be tested for other SCA subtypes, particularly SCA1, SCA2 and SCA10<sup>2,3</sup>.



Figure. All three patients, SCA1 (A), SCA2 (B) and SCA3 (C) have widening of the eyelid opening with white parts of the sclera appearing above and below the limbus, characterizing eyelid retraction or Collier's sign.

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