RESIDENT & FELLOW SECTION

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Teaching Video NeuroImages: Spastic ataxia syndrome

The Friedreich-like phenotype of ARSACS

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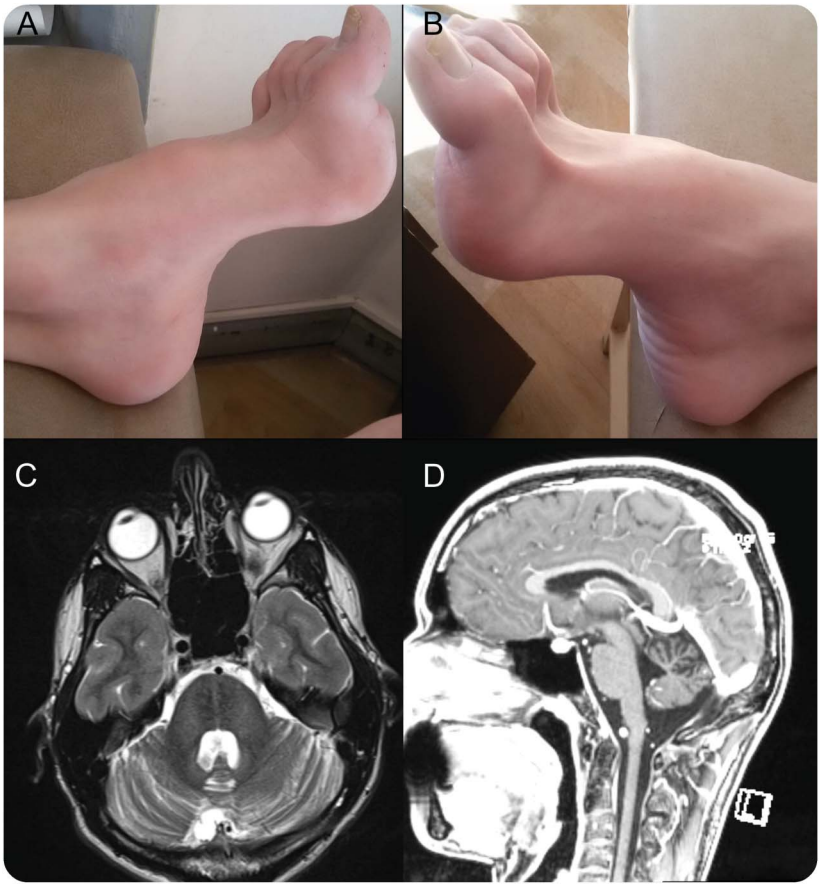
Figure Key clinical and MRI findings



Friedreich-like pes cavus (A, B) and suggestive MRI findings: liner hypointensity on axial T2-weighted brain MRI (classic abnormality) (C) and cerebellar anterosuperior vermal cerebellar atrophy (nonspecific; D).

A 24-year-old Chilean man with slowly progressive ataxia since age 2 presented with spastic ataxia, hyperreflexia, pes cavus, axonal polyneuropathy, incomplete right-bundle branch block on ECG, and impaired glucose tolerance test, suggesting Friedreich ataxia (figure; video at Neurology.org). However, the combination of hyperreflexia and cerebellar (rather than cervical cord) atrophy with T2-weighted linear hypointensity in the pons on brain MRI suggested autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS). Biallelic mutations were found (c.4492C>T p.[R1498X] and c.2388dupA p.[L797Ifs*4]) in the *SACS* gene (NCBI sequence NM_001278055). ARSACS is the second most common cause of autosomal recessive spastic ataxia syndrome (*SACS* mutations account for 37% of

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Friedreich-negative cases)[1] and should be considered in any population with suggestive MRI abnormalities.[2]

AUTHOR CONTRIBUTIONS

Drs. Saffie, Fernandez, Acosta, and de la Cerda: acquisition of data, analysis and interpretation, critical revision of the manuscript for important intellectual content. Dr. Kauffmann: genetic analysis, critical revision of the manuscript for important intellectual content. Dr. Espay: report analysis and interpretation, critical revision of the manuscript for important intellectual content.

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