Teaching Video NeuroImages:

Spastic ataxia syndrome

The Friedreich-like phenotype of ARSACS

Paula Saffie, MD
Marcelo A. Kauffman,
MD, PhD
Jose Manuel Fernandez,
MD
Ignacio Acosta, MD
Alberto J. Espay, MD,
MSc
Andrés de la Cerda, MD

Correspondence to
Dr. Espay:
alberto.espay@uc.edu

Figure Key clinical and MRI findings

Friedreich-like pes cavus (A, B) and suggestive MRI findings: linear 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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From the Corporacion de Investigacion de Neurologia de Santiago (CINSAN), Clinica Davila (A.d.I.C.), Universidad de los Andes and CETRAM (P.S.), Santiago, Chile; Consultorio y Laboratorio de Neurogenética (M.A.K.), Centro Universitario de Neurología "José María Ramos Mejía" y División Neurología, Hospital JM Ramos Mejía, Facultad de Medicina, UBA, and Programa de Medicina de Precision y Genomica Clinica, Instituto de Investigaciones en Medicina Traslacional, Facultad de Ciencias Biomédicas, Universidad Austral-CONICET, Argentina; Facultad de Medicina Clínica Alemana-Universidad del Desarrollo (J.M.F., I.A.), Servicio de Neurología Clínica Alemana, Santiago, Chile; and Department of Neurology (A.J.E.), UC Gardner Neuroscience Institute, Gardner Center for Parkinson's Disease and Movement Disorders, University of Cincinnati, OH.

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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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