Teaching NeuroImages: Autosomal recessive spastic ataxia of Charlevoix-Saguenay

Typical MRI findings

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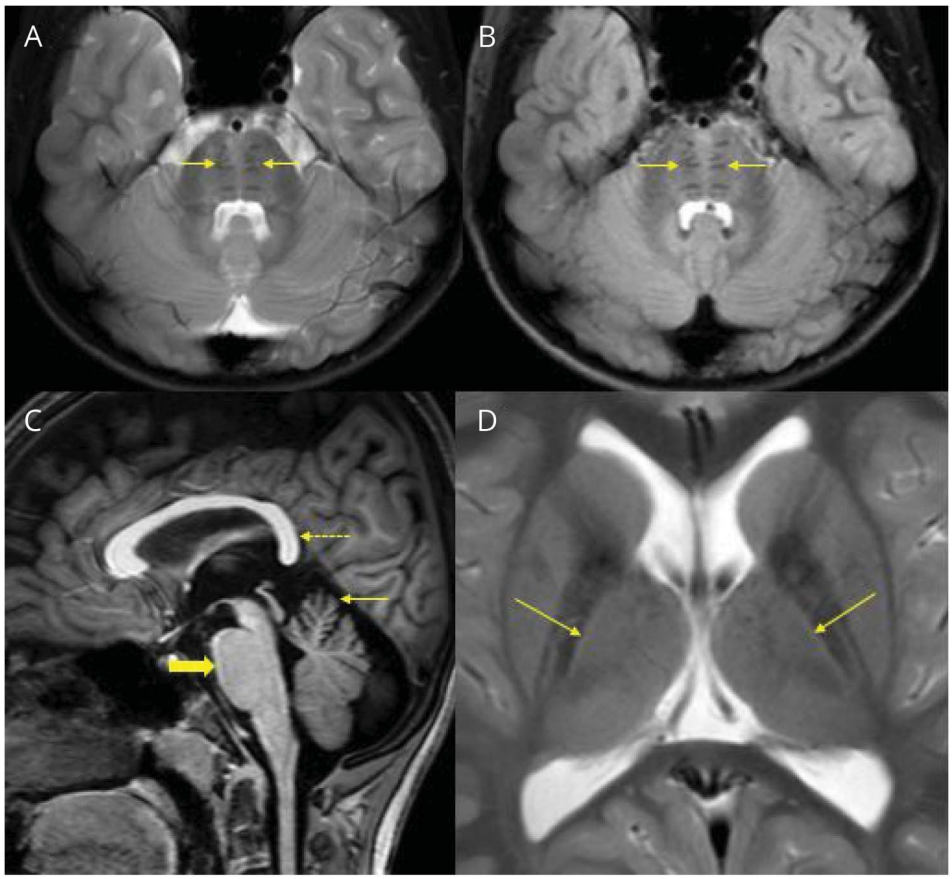
Figure MRI shows T2-hypointense pontine striations, superior vermian atrophy, and T2 hyperintensity around the thalami



(A, B) Axial T2 and fluid-attenuated inversion recovery images show an enlarged pons with linear hypointense striations on either side of the midline (arrows). (C) Sagittal T1-weighted image shows the enlarged pons (large arrow), superior vermian atrophy (arrow), and thinning of the splenium of corpus callosum (dashed arrow). (D) Axial T2-weighted image shows a rim of hyperintensity around the thalami (arrows).

A 9-year-old boy presented with a slowly progressive spastic ataxic syndrome. Sensorimotor polyneuropathy was detected on nerve conduction studies. MRI (figure) was highly suggestive of autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS).

ARSACS is a neurodegenerative disorder characterized by progressive cerebellar signs, spasticity, and peripheral neuropathy.[1] Although the majority of cases have an early age at onset (3.4 ± 1.55 years),[2] considerable variability of age at onset (infancy to >40 years) has also been described in the literature.[1] Long repetition time linear pontine hypointense striations (pathophysiology of which is unknown) and superior vermian atrophy are characteristic imaging

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findings.[1,2] Other useful imaging features include T2 hyperintense rim around the thalami and thinning of the cervical spinal cord.

Author contributions

Asthik Biswas: concept, image interpretation, writeup of manuscript. Mugil Varman: image interpretation and preparation. Sangeetha Yoganathan: clinical input, critical revision for intellectual content. Patel Khushboo Subhash: clinical input. Sunithi Mani: critical revision for intellectual content.

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