Teaching NeuroImage: Selectively Bright Inferior Cerebellum in Christianson Syndrome

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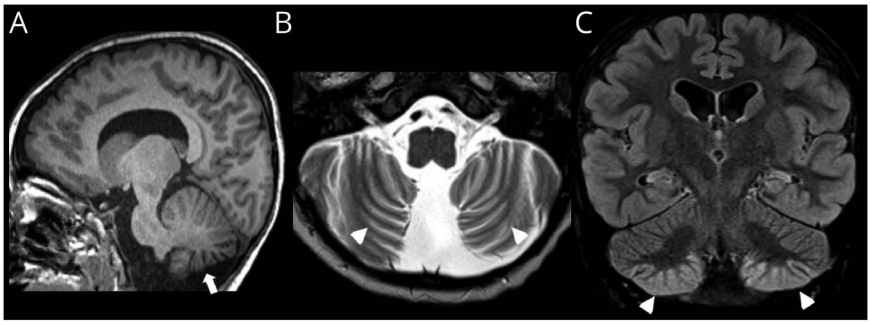
Figure Bright Inferior Cerebellum



Brain MRI. Sagittal T1-weighted image (A) demonstrates asymmetric cerebellar atrophy with predominant inferior cerebellar involvement (arrow, A). Axial T2-weighted and coronal fluid-attenuated inversion recovery images (B and C) show cortical hyperintensity selectively in the inferior cerebellum, known as a "bright inferior cerebellum" (arrowheads, B and C).

Case Report

A 4-year-old boy presented with developmental delay (nonverbal and unable to walk or sit independently) and recurrent tonic-clonic seizures beginning at 1 month of age. EEG demonstrated underdeveloped background organization and intermittent focal right occipital slowing. Brain MRI revealed inferior cerebellar atrophy with T2-WI and FLAIR cortical hyperintensity (Figure). A *SLC9A6* c.803+3_803+6del (intronic) pathogenic variant was detected, confirming a diagnosis of Christianson syndrome (CS). CS is characterized by severe cognitive dysfunction, behavioral disorder, seizures, ataxia, and microcephaly. The condition results from loss-of-function alterations affecting sodium Na+/H+ exchange enzymes. While some clinical features may overlap with Angelman syndrome (AS), inferior cerebellar atrophy is characteristic of CS. It is important to distinguish between the 2 diagnoses because the prognosis in CS is worse than that in AS.

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Teaching slides
links.lww.com/WNL/C294

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