Teaching NeuroImage: Selectively Bright Inferior Cerebellum in Christianson Syndrome

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Case report:

A 4-year-old male presented with developmental delay (non-verbal, unable to walk or sit independently) and recurrent tonic-clonic seizures beginning at one 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 mutations 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 two diagnoses as the prognosis in CS is worse than in AS.

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 FLAIR images (B-C) show cortical hyperintensity selectively in the inferior cerebellum, known as a “bright inferior cerebellum” (arrowheads, B and C).

References:


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