Teaching NeuroImages: Figure of 8

The clue to the diagnosis of AMPD2 pontocerebellar hypoplasia (PCH9)

Mariasavina Severino,
MD

Federico Zara, PhD
Andrea Rossi, MD
Pasquale Striano, MD,
PhD

Correspondence to
Dr. Striano:
strianop@gmail.com

Figure Typical figure of 8 midbrain appearance of PCH8 subtype

(A, B) Axial T2-weighted images of figure of 8 midbrain appearance (dotted oval), small hyperintense basal ganglia, and hypointense thalami (arrowheads). (C) Sagittal T1-weighted image: small pons (arrow), absent corpus callosum (empty arrow). (D) Coronal T2-weighted image: cerebellar hemispheric atrophy (arrowheads) and periventricular leukoencephalopathy (empty arrows).

A 2-year-old girl, born after uneventful pregnancy from healthy nonconsanguineous parents, presented with failure to thrive, microcephaly, facial dysmorphism, strabismus, nystagmus, axial hypotonia, lower limb spasticity/hyperreflexia, dystonic movements, and no active postures. Since the first months of life, developmental delay without any motor skills acquisition, drug-resistant epilepsy, and progressive spasticity had emerged. Neuroimaging revealed pontocerebellar hypoplasia with the figure of 8 midbrain appearance

(figure), a distinctive sign for PCH8 or AMPD2 deficiency.^{1,2} Sanger sequencing of the adenosine monophosphate deaminase 2 enzyme gene (*AMPD2*) revealed the first compound heterozygous mutation (c.1664C>T; c.1112+1 G>A). The patient died at age 3 years due to respiratory insufficiency.

AUTHOR CONTRIBUTIONS

Pasquale Striano, Mariasavina Severino: drafting the manuscript. Andrea Rossi, Federico Zara: critical revision. All authors approved the final version.

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From the Neuroradiology Unit (M.S., A.R.), Laboratory of Neurogenetics (F.Z.), and Pediatric Neurology and Muscular Diseases Unit, Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health, University of Genoa (P.S.), Istituto "Giannina Gaslini," Genoa, Italy.

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