Pontocerebellar hypoplasia

Heather Borders, MD

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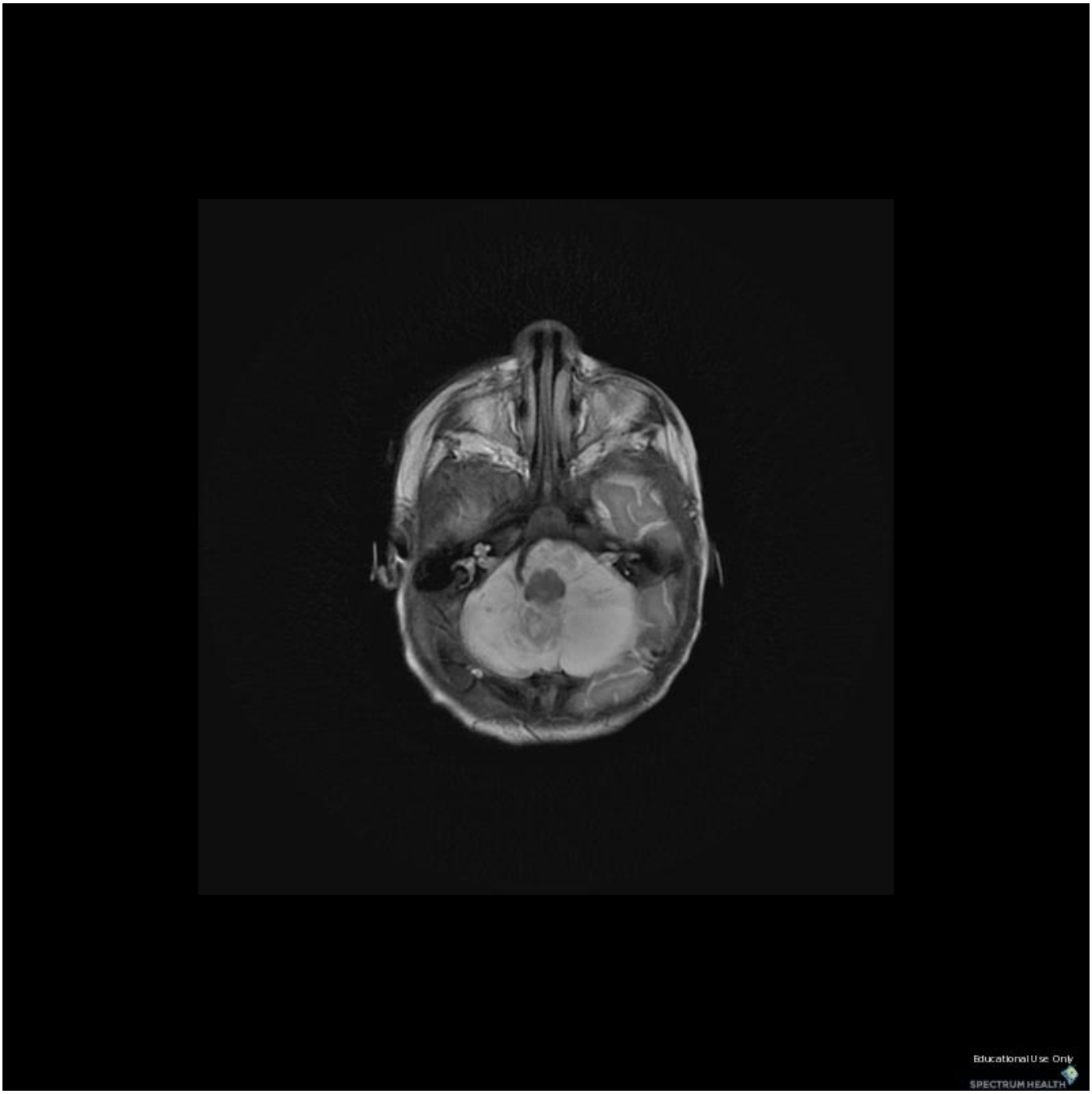
History
14 month old with microcephaly, delayed development, nystagmus

Diagnosis
Pontocerebellar Hypoplasia

Discussion
The association of cerebellar hypoplasia with pontine hypoplasia is not unusual, likely due to shared origin of neurons in the rhombic lips. Numerous connections between the pons and cerebellum likely also play a role.
This is a heterogeneous problem with genetic and acquired etiologies. Researchers have described six forms of pontocerebellar hypoplasia. All forms of this condition are characterized by abnormal brain development, problems with movement, delayed development, and intellectual disability. The signs and symptoms are usually present at birth, and in some cases they can be detected before birth. Many children with pontocerebellar hypoplasia live only into infancy or childhood, although some affected individuals have lived into adulthood.
The MRI features in a study by Uhl, et al. included: (1) Hypoplastic cerebellum situated close to the tentorium. (2) The cerebellar hemispheres are reduced to bean-like or wing-like structures. (3) Markedly hypoplastic ventral pons. (4) Slight atrophy of the supratentorial gyral pattern. (5) Dilated cerebromedullary cistern and fourth ventricle. (6) Delayed myelination of the white matter. (7) No significant disorganisation of brain architecture and no severe corpus callosum defect.

Findings
Markedly hypoplastic cerebellum with a small remnant of vermis along the tentorium and no cerebellar hemisphere tissue. Hypoplastic pons. Normal sized posterior fossa.

Reference
Barkovich, Pediatric Neuroimaging p. 172.
MR findings in pontocerebellar hypoplasia Journal Pediatric Radiology 1432-1998 (Online)Issue Volume 28, Number 7 / July, 1998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