

Pontocerebellar hypoplasia

Heather Borders, MD

06/27/2010

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.

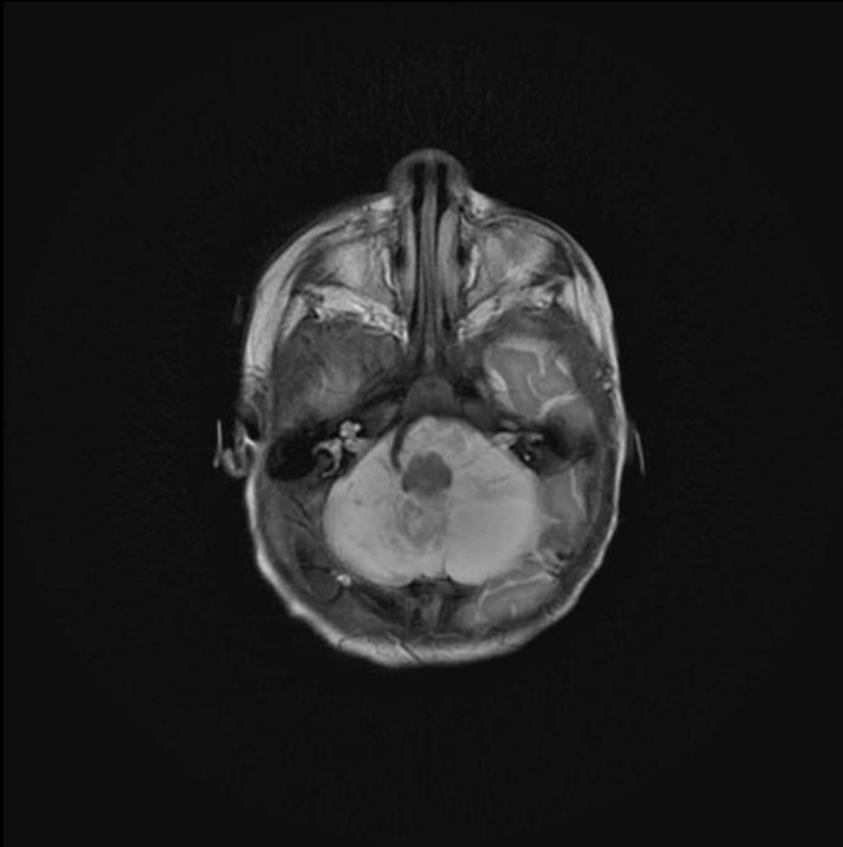
Findings

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







Sponsored By



Disclaimer

This teaching site is partially funded by an educational grant from GE Healthcare and Advanced Radiology Services, PC. The material on this site is independently controlled by Advanced Radiology Services, PC, and GE Healthcare and Spectrum Health have no influence over the content of this site
Content Download Agreement

The cases and images on this website are owned by Spectrum Health. Permission is granted (for nonprofit educational purposes) to download and print materials to distribute for the purpose of facilitating the education of health professionals. The authors retain all rights to the material and users are requested to acknowledge the source of the material.

Site Disclaimer

This site is developed to reach healthcare professionals and medical students. Nothing this site should be considered medical advice.

Only your own doctor can help you make decisions about your medical care. If you have a specific medical question or are seeking medical care, please contact your physician.

The information in this website is provided for general medical education purposes only and is not meant to substitute for the independent medical judgment of a physician relative to diagnostic and treatment options of a specific medical condition.

The viewpoints expressed in these cases are those of the authors. They do not represent an endorsement. In no event will Advanced Radiology Associates, PC, Spectrum Health Hospitals (Helen DeVos Children's Hospital) or GE Healthcare be liable for any decision made or action taken in reliance upon the information provided through this website.