Teaching NeuroImages:

Molar tooth sign with hypotonia, ataxia, and nystagmus (Joubert syndrome) and hypothyroidism

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Figure MRI

Axial (A, B), midsagittal (C), and parasagittal (D) T1-weighted MRI demonstrating deepened interpeduncular fossa (arrowhead, A) and horizontally oriented superior cerebellar peduncles (arrows, A, D) forming the “molar tooth” sign, as well as cerebellar vermis hypoplasia and fourth ventricle enlargement with elevated roof (*, C) and abnormal cerebellar structure (C, D).

A 2-year-old girl with congenital hypothyroidism, diagnosed by neonatal screen, and thyroid dysplasia was referred for developmental delay, hypotonia, ataxia, and nystagmus. She had neonatal episodic hyperpnea and renal cysts. At 3 she understood simple language and signed 6 words. Imaging revealed vermian hypoplasia, the “molar tooth” sign (figure), considered pathognomonic for Joubert syndrome,¹ a heterogeneous group of autosomal recessive disorders linked to several chromosomal loci. Classic features are hypotonia, psy-

chomotor delays, vermian hypoplasia, episodic hyperpnea or apnea, and abnormal eye movement. Joubert variably includes polymicrogyria, retinal, renal, or hepatic abnormalities, as well as polydactyly and orofacial dysmorphisms.¹

REFERENCE

1. Valente EM, Brancati F, Dallapiccola B. Genotypes and phenotypes of Joubert syndrome and related disorders. *Eur J Med Genet* 2008;51:1–23.

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Disclosure: Dr. Graber has received honoraria and funding for travel from the Discovery Institute for Medical Education. Dr. Lau and Dr. Sathe report no disclosures.

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Neurology 2009;73:e106

DOI 10.1212/WNL.0b013e3181c679ba

This information is current as of December 14, 2009

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