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

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, a heterogeneous group of autosomal recessive disorders linked to several chromosomal loci. Classic features are hypotonia, psychomotor 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.

REFERENCE
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