Teaching Video NeuroImages: Characteristic head jerks in congenital oculomotor apraxia due to Joubert syndrome

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A 29-year-old man presented in our clinic due to repetitive head jerks since his first year of life, which were previously diagnosed as tics. Examination revealed difficulties initiating voluntary saccades in the horizontal plane and disturbed hand–eye coordination with compensatory head thrusts (video and figure, A). The diagnosis of congenital oculomotor apraxia was made. Brain MRI revealed cerebellar vermis hypoplasia with elongated superior peduncles ("molar tooth sign"; figure, B) characteristic for Joubert syndrome, a rare autosomal-recessive ciliopathy with genetic

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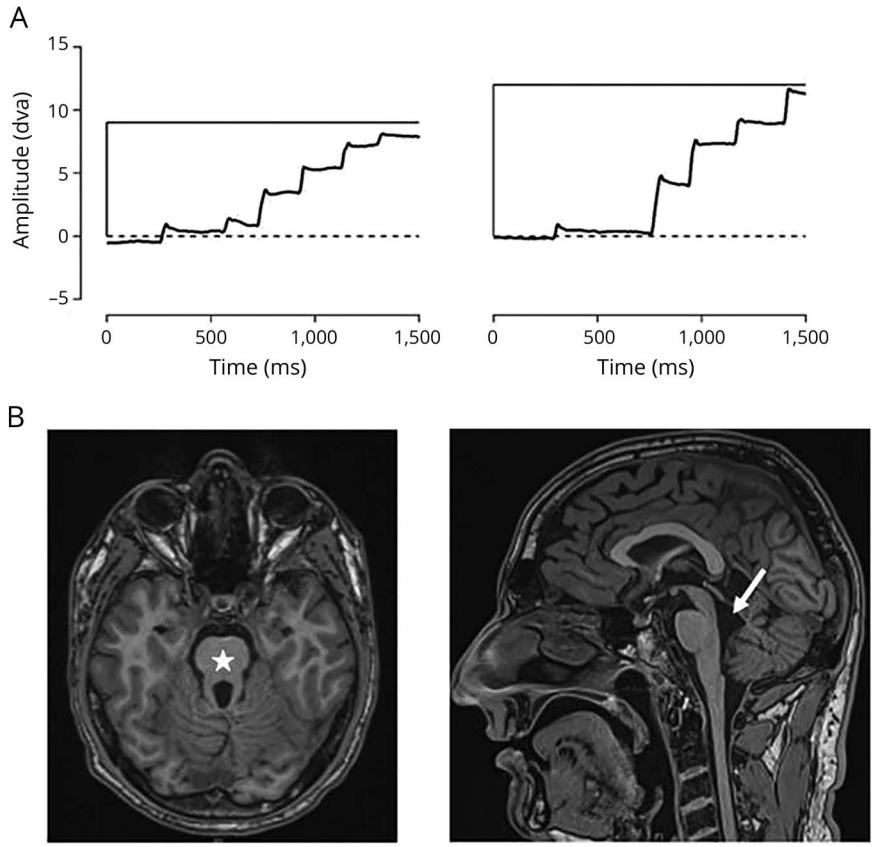
Video

Figure Video-oculographic recording and T1-weighted transversal and sagittal brain MRI



(A) Video-oculographic recording of exemplary saccades to a visual target, jumping rightward from initial fixation to a peripheral position (9/12 degrees of visual angle [dva]) at time "0," demonstrating prolonged latencies and hypometria of horizontal visually guided saccades. (B) T1-weighted transversal and sagittal brain MRI show hypoplasia of the cerebellar vermis with elongated superior cerebellar peduncles (molar tooth sign) (star) as well as enlargement of the fourth ventricle (arrow).

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heterogeneity.[1] Joubert syndrome is a common etiology of congenital oculomotor apraxia, a condition first described by Cogan,[2] who considered compensatory head jerks diagnostic.

Author contributions

F. Borngräber: drafting/revising the manuscript, data acquisition, analysis or interpretation of data, accepts responsibility for conduct of research and final approval, acquisition of data. Y. Peng: drafting/revising the manuscript, data acquisition, analysis or interpretation of data, accepts responsibility for conduct of research and final approval. F. Ostendorf: drafting/revising the manuscript, analysis or interpretation of data, accepts responsibility for conduct of research and final approval, acquisition of data. A.A. Kühn: drafting/revising the manuscript, accepts responsibility for conduct of research and final approval, study supervision. C. Ganos: drafting/revising the manuscript, data acquisition, study concept or design, analysis or interpretation of data, accepts responsibility for conduct of research and final approval, study supervision.

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Disclosure

The authors report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

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