AJNR

Cerebellar vermis and midbrain dysgenesis in oculomotor apraxia: MR findings.

E A Whitsel, M Castillo and O D'Cruz

AJNR Am J Neuroradiol 1995, 16 (4) 831-834
http://www.ajnr.org/content/16/4/831

Cerebellar Vermis and Midbrain Dysgenesis in Oculomotor Apraxia: MR Findings

Eric A. Whitsel, Mauricio Castillo, and O'Neill D'Cruz

Summary: The MR imaging findings in two patients with the clinical diagnosis of oculomotor apraxia are presented. Both cases showed dysgenesis of the cerebellar vermis, and the colliculi were fused in one patient. No supratentorial abnormalities were seen in either patient.

Index Terms: Cerebellum, vermis; Mesencephalon; Vision; Movement

Congenital oculomotor apraxia is a rare syndrome described in 1952 by Cogan (1). It is seen mainly in boys and is clinically characterized by rotational head thrusts that attempt to compensate for the lack of voluntary eye movements (2–5). We present the clinical and magnetic resonance (MR) imaging features in two children with congenital oculomotor apraxia.

Case Reports

Case 1

A 6½-year-old boy who was born at full term had a significant prenatal history of exposure to cigarettes, alcohol, and illicit drugs. At 6 months of age, he was seen because of "rolling" eyes and head "jerking." Clinically, bilateral convergent strabismus, nystagmus, generalized hypotonia, and hyperreflexia were present. Findings on computed tomography of the brain and orbits were reported as normal, and surgical correction of strabismus was done. At 10 months of age, motor milestones were clearly delayed. At 4 years of age, cognition was average (full scale IQ, 91). Visual acuity was 20/400 bilaterally at 2 years of age and improved to 20/50 bilaterally after correction of strabismus. An electroencephalogram was normal. Physical evaluation on presentation to our institution at 6½ years of age revealed choreoathetoid movements of the arms, complex motor tics (head jerking and nose wiping), wide-based gait, difficulty balancing, diffuse hypotonia, and hyperreflexia. Both eyes were deviated laterally. Head thrusts to the opposite side occurred if an object was placed in front of the patient. These compensatory head thrusts resulted in passive direction of the eyes toward the object of interest. Once fixation of gaze was accomplished, the head thrusts stopped. Dysmorphic features were present and included borderline microcephaly, prominent cranial vertex, flat occiput, long ears, mandibular prognathism, thoracic levoscoliosis, scapular winging, and pes planus. MR imaging showed a hypogenetic cerebellar vermis (Fig. 1A). The superior cerebellar peduncles were elongated and straight and the fourth ventricle was abnormally shaped (Fig 1B).

Case 2

A 5-year-old girl born at full term was first seen at 5 months of age because of episodic head "jerking." Physical examination at that time revealed telangiectasia in the lateral superior quadrant of the left eye, bilateral convergent strabismus, generalized hypotonia, and hyperreflexia. When referred to us at 5 years of age, motor milestones were delayed. Although no quantitative cognitive tests were performed, she was enrolled in preschool; her teachers noted delay in both acquisition of new cognitive abilities and development of social skills. She had a wide-based gait, generalized ataxia, and a dysarthric speech pattern. Her eyes were laterally deviated, and when an object was placed in front of her, rotational head thrusts ensued until the object of interest was located in her line of vision. Head circumference was normal. MR showed a hypogenetic cerebellar vermis with small residual rostral lobules (Fig 2A). The superior cerebellar peduncles were straight and elongated and the fourth ventricle abnormally shaped (Fig 2B). The collicular plate was abnormally shaped.

Discussion

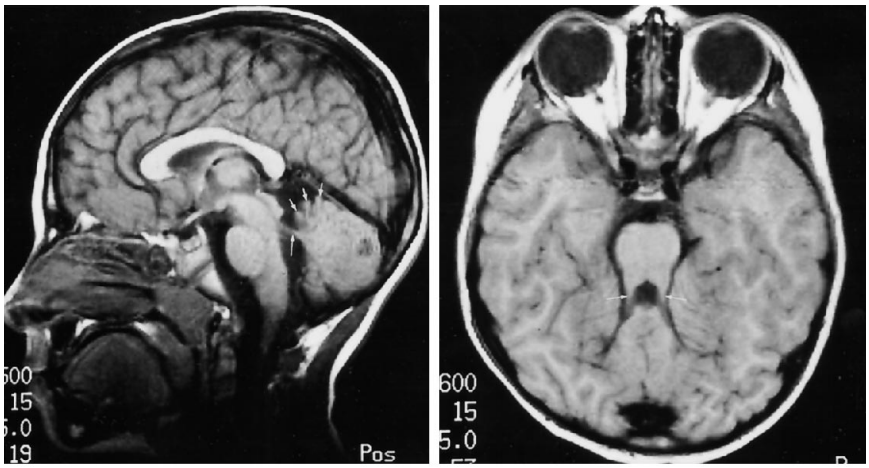
Congenital oculomotor apraxia is usually diagnosed between the fourth and eighth months of life; the most common initial symptom is the inability to follow objects visually (1, 2). The presence of normal visual evoked potentials

Received June 16, 1993; accepted after revision October 20.

From the Department of Epidemiology, School of Public Health (E.A.W.), and Departments of Radiology (M.C.) and Neurology (O.D.), School of Medicine, University of North Carolina, Chapel Hill.

Address reprint requests to M. Castillo, MD, Department of Radiology, CB 7510, University of North Carolina at Chapel Hill, Chapel Hill, NC 27599-7510.

AJNR 16:831–834, Apr 1995 0195-6108/95/1604–0831



A B

Fig 1. *A*, Midline sagittal T1-weighted MR image (600/15/2 [repetition time/echo time/excitations]) shows dysgenetic and small lingula, central lobule, and culmen (*small arrows*) of cerebellar vermis. The abnormal superior cerebellar peduncles (*long arrow*) produce flattening of the rostral fourth ventricle, which is abnormal in shape. The foramen of Magendie is open, and no hydrocephalus is present. The configuration of the midbrain and the corpus callosum is normal. The area of low signal intensity in the posterior cerebellum is of uncertain cause.

B, Axial T1-weighted image (600/15/2) shows the superior cerebellar peduncles (*arrows*) to be elongated and to project straight dorsally. The shape of the fourth ventricle is abnormal and the vermis is absent at this level. No fusion of cerebellar hemispheres was present on caudal sections.

serves to exclude the more common cortical blindness (6).

The striking ocular abnormalities begin with rotational head thrusts, which compensate for impairment of voluntary horizontal gaze (7). These head thrusts then trigger a reflexive deviation of the eyes toward the opposite direction, which is completed when the eyes rest passively on the initial target. The presence of these movements in combination with the absence of the fast phase of nystagmus on horizontal optokinetic testing and presence of contralateral eye deviation on rotation of the head about a vertical axis, helps to clinically confirm the diagnosis. Although this syndrome is typically thought of mainly as an ocular abnormality, it does have other clinical features, such as difficulty reading, gross motor delay, muscle incoordination, gait abnormalities, and a persistent expressive language delay (8, 9). In our cases, generalized motor-ataxic symptoms as well as developmental/motor delay were prominent. Our limited observations, based on the two cases presented here, support the fact that congenital oculomotor apraxia might not be limited to ocular abnormalities and that patients who have this syndrome might have generalized neurologic abnormalities. The clinical manifestations of this syndrome can diminish gradually with age.

Anatomically, the exact area of the brain responsible for the clinical findings is uncertain, and no consistent imaging abnormalities are evident (8). Similar symptoms can be seen with lesions involving the pons, mesencephalon, and frontal and parietal lobes (10). Lesions that primarily involve Broadmann's area 8 in the frontal lobes can produce saccadic (involuntary small and rapid eye movements that change point of fixation) abnormalities (11). Lesions in the parietal lobes (Broadmann's area 7) also cause increased latency of contralateral saccades and impair smooth ipsilateral pursuit. Indirect excitatory projections from both frontal and parietal lobes convey motor command and visual attention signals to the paramedian pontine reticular formation via the internal capsule, cerebral peduncles, and superior colliculi. Burst cells in the paramedian pontine reticular formation (level of the floor of the fourth ventricle) activate motor neurons in the oculomotor (III) and abducens (IV) nuclei, which are located at the level of the superior colliculi. These motor neurons excite the extraocular muscle agonists directly and indirectly via the interneurons in the medial longitudinal fasciculi. The amplitude of the motor neuron activity is modulated by input from the dorsal cerebellar vermis via projections coursing in the cerebellar peduncles. Also, because there are numerous transcallosal connections

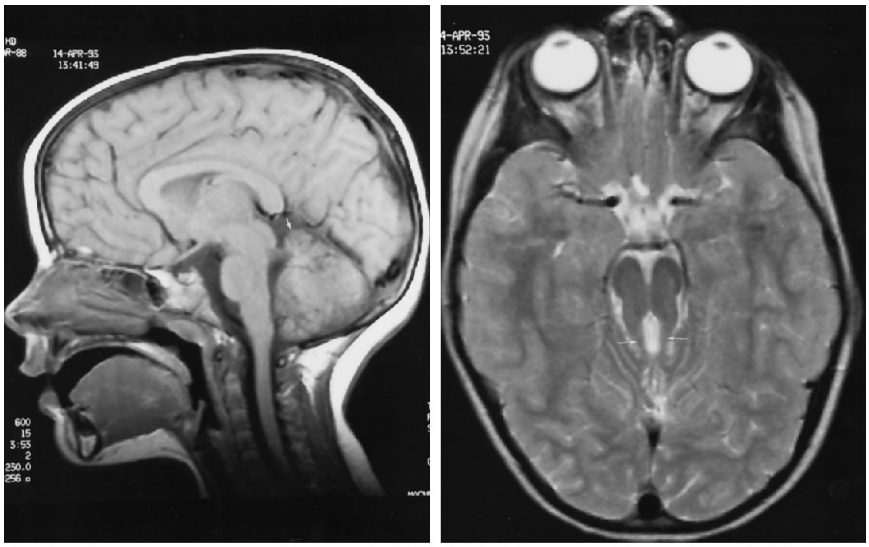


Fig 2. *A*, Midline sagittal T1-weighted image (600/15/2) shows dysgenetic rostral vermian lobules (*arrow*) and absence of the remaining vermis. The superior cerebellar peduncles are abnormal and produce flattening of the rostral fourth ventricle, which is abnormal in shape; the foramen of Magendie is patent. There is no intercollicular sulcus, and the superior and inferior colliculi appear to be fused. The corpus callosum is normal.

B, Axial T2-weighted spin-echo image (2500/90/1) shows a somewhat small midbrain and elongated superior cerebellar peduncles (*arrows*). No cerebellar fusion is present.

between the parietal lobes, dysgenesis of the corpus callosum can produce the syndrome. Indeed, Orrison and Robertson (4) reported two cases of congenital oculomotor apraxia in which the corpus callosum was absent. The presence of ataxia and tremor in some patients suggests abnormalities in the cerebellum, vermis, and their connections (12). In one case, MR imaging showed lesions at the mesencephalic-diencephalic junction (10). Although the origin of the lesions in that case were not proved, the authors postulated that the lesions were close to the rostral interstitial nucleus of the medial longitudinal fasciculus and that they might have interrupted their descending projections. These neurons are critical for vertical saccades and quick phases of nystagmus. In a different case, MR imaging showed only diffuse atrophy and thinning of the corpus callosum (11). Computed tomography showed a prominent fourth ventricle in four cases, but definite partial agenesis of the cerebellar vermis was present in only one case (12). From the above description, it is clear that congenital oculomotor aprexia is a syndrome that involves abnormalities of the collicular plate, fourth ventricle, cerebellar peduncles, cerebral hemispheres, and corpus callosum.

In our patients, the ocular and widespread clinical manifestations were compatible with the syndrome. In both patients, MR imaging features were similar. The cerebellar vermis was hypogenetic with preservation of dysplastic rostral areas (lingula, central lobule, and culmen) (Figs 1A and 2A). The superior cerebellar peduncles assumed a straight and elongated shape causing flattening of the rostral fourth ventricle (Figs 1A and B and 2A and B). The rostral fourth ventricle was also enlarged, reflecting dysgenesis of the superior vermis. The midbrain was normal in one case and in the second showed absence of the intercollicular sulcus with probable fusion of the superior and inferior colliculi (Fig 2A). We believe the imaging features seen in our patients clearly explain the clinical manifestations.

Clinically and by imaging, congenital oculomotor apraxia can be indistinguishable from the more common (albeit also rare) Joubert syndrome (13). Panting respiration and retinal dystrophy are seen more commonly in patients with Jouberts syndrome. Indeed, both syndromes might represent parts of a more complex spectrum of abnormalities involving the cerebellar vermis and brain stem. It is also interesting to note that patients with ataxia telan-

giectasia can also have dysgenesis of the cerebellar vermis (14), and one of our patients (case 2) harbored ocular telangiectasia. Because no cutaneous lesions, telangiectasias of the cerebrum or brain stem, cerebellar atrophy, or immune deficiency were seen in our patient, we believe she did not have ataxia telangiectasia. Some patients will have symptoms similar to those seen in congenital oculomotor apraxia and ataxia but will not have other criteria needed for the diagnosis of ataxia telangiectasia. These patients can be categorized as having the ataxia–oculomotor apraxia syndrome (15). Because both of our cases had nonprogressive ataxia and early developmental delay, we believe that these symptoms fit better the congenital form rather than the acquired form of oculomotor apraxia. Rare forms of acquired oculomotor apraxia are seen in association with ataxia telangiectasia and with Gaucher disease types 2 and 3 (16).

In conclusion, dysgenesis of the cerebellar vermis is present in a variety of clinical symptoms. When MR shows almost complete vermian agenesis in patients whose symptoms correspond to those describe here, the diagnosis of congenital oculomotor apraxia should be considered. It is conceivable that the dysgenesis of the vermis and midbrain seen in our cases reflects a wider (albeit not visible by MR) disorganization of the neural pathways involved in the production of normal and voluntary saccades.

References

1. Cogan DG. A type of congenital ocular motor apraxia presenting jerky head movement. *Trans Am Acad Ophthalmol Otolaryngol* 1952;56:853–862
2. Cogan DG. Congenital ocular motor apraxia. *Can J Ophthalmol* 1966;1:253–260
3. Vassella F, Lutschg J, Mumenthaler M. Cogan's congenital ocular motor apraxia in two successive generations. *Dev Med Child Neurol* 1972;14:788–796
4. Orrison WW, Robertson WC. Congenital ocular motor apraxia: a possible disconnection syndrome. *Arch Neurol* 1979;36:29–31
5. Gittinger JW, Sokol S. The visual-evoked potential in the diagnosis of congenital ocular motor apraxia. *Am J Ophthalmol* 1982; 93:700–703
6. Rosenberg ML, Wilson E. Congenital ocular motor apraxia without head thrusts. *J Clin Neuroophthalmol* 1987;7:26–28
7. Fielder AR, Gresty MA, Dodd KL, Mellor DH, Levene MI. Congenital ocular motor apraxia. *Trans Ophthalmol Soc U K* 1986;105: 589–598
8. Altrocchi PH, Menkes JH. Congenital ocular motor apraxia. *Brain* 1960;83:579–588
9. Rappaport L, Urion D, Strand K, Fulton AB. Concurrence of congenital ocular motor apraxia and other problems: an expanded syndrome. *Dev Med Child Neurol* 1987;29:85–90
10. Ebner R, Lopez L, Ochoa S, Crovetto L. Vertical ocular motor apraxia. *Neurology* 1990;40:712–713
11. Bochert MS, Sadun AA, Sommers JD, Wright KW. Congenital ocular motor apraxia in twins: findings with magnetic resonance imaging. *J Clin Neuroophthalmol* 1987;7:104–107
12. Eda I, Takashima ES, Kitahara T, Ohno K, Takeshita K. Computed tomography in congenital ocular motor apraxia. *Neuroradiology* 1984;26:359–362
13. Saraiva JM, Baraitser M. Joubert syndrome: a review. *Am J Med Genet* 1992;43:726–731
14. Woods CG, Taylor AMR. Ataxia-telangiectasia in the British Isles: the clinical and laboratory features of 70 affected individuals. *Q J Med* 1992;82:169–179
15. Aicardi J, Barbosa C, Andermann E, et al. Ataxia-ocular motor apraxia: a syndrome mimicking ataxia-telangiectasia. *Ann Neurol* 1988;24:497–502
16. Gross-Tsur V, Har-Even Y, Gutman I, Amir N. Oculomotor apraxia: the presenting sign of Gaucher disease. *Pediatr Neurol* 1989; 5:128–129