Neuro-ophthalmology 0165-8107/90/US$ 3.50 (Accepted 10 October 1989)

Congenital ocular motor apraxia: a neurodevelopmental and neuroradiological study

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ABSTRACT. Since its first description in 1952 neither the pathogenesis nor the localization of congenital ocular motor apraxia (COMA) have been cleared. There are reports indicating that COMA is more variable in its clinical expression and neuroradiological findings than previously assumed. In eight children with COMA neurological examination, developmental testing and neuroradiological investigation by CT or MRI were carried out. Ataxia was found in five, developmental delay in six of the patients. CT or MRI were normal in four children, the others had variable malformations such as hypoplasia of the cerebellum or corpus callosum, heterotopias of gray matter and maturational delays in the basal ganglia. No correlations between the clinical symptoms and the neuroradiological findings were found. The authors suggest that COMA is not an entity but rather a symptom, variably associated with malformations and maturational disorders of the brain.

Key words: congenital ocular motor apraxia; neuroimaging; neurological and developmental disorders

INTRODUCTION

Congenital ocular motor apraxia (COMA) is a congenital disorder characterized by disturbance of initiating fast saccades of horizontal eye movements. During the first months of life affected infants learn to follow objects by typical jerky, overshooting head movements, and therefore they are able to adjust their gaze quickly to a new target. With fixated head they are unable to follow fast moving objects. Horizontal optokinetic and vestibular nystagmus are impaired, but vertical eye movements and slow horizontal pursuits are intact[1, 2].

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These ocular problems tend to improve with age, but children with COMA are reported to have other neurological problems such as ataxia and/or developmental delay[2-5].

The exact pathogenesis as well as the localization of these abnormalities are still unknown, although Zee *et al.*[1] as well as Pierrot-Deseilligny *et al.*[6-8] and Eda *et al.*[3] pointed out several possible locations.

We report eight children with COMA. By trying to compare the clinical symptoms and the neuroradiological findings we would like to show the great variability of this condition.

METHODS

During the last eight years we have seen eight children with COMA. Children with acquired ocular

motor apraxia as seen in M. Gaucher type III, ataxia teleangiectatica and Leigh's disease were excluded.

All children had a detailed neurological examination. Five underwent developmental testing (Griffith-test[9] or Snijders-Oomen-nonverbal-intelligence-test[10]), the development of three children already attending school was judged according to their school performance. All of them had neuroradiological investigations, four with CT (General Electric CT 9800 system), four with MRI (using a 2.35 T superconducting magnet for combined MR-imaging and spectroscopy[11]). The investigations have been performed during the first three years of life. The children had sedative drugs and were examined during sleep.

RESULTS

The relevant data are summarized in Tables 1 and 2. Five of eight children (cases 1-5) suffered from ataxia affecting trunk and limbs, in combination with variable degrees of hypotonia. Ataxia was always associated with a delay in motor development. Although ataxia did not improve significantly in most cases, the motor development was improving up to school age. Three children aged four to eight months (cases 6-8) were too young to be assessed for possible ataxia, but two of them showed developmental delay.

Six of the children (cases 1, 2, 4, 5, 6, 8) dis-

TABLE 2. Mental development and school performance

Case	*Test*	*Age at testing (years)*	*DQ/IQ*		*School performance*
			Motor	*Mental*	
5	G	8/12	93	83	
6	G	10/12	50	60	
7	G	5/12	114	100	
8	G	4/12	79	66	
1	S	7 1/2	delayed	50	
2		7	delayed		below grade level
3		8	delayed		at grade level
4		6	delayed		below grade level

G = Griffith test: Developmental quotient.
S = SON-Y9: Intelligence quotient.

TABLE 1. Neurological and neuroradiological findings

Case	*Sex*	*Age at diagnosis*	*Clinical findings*		*Neuroimaging*	
			Ataxia Hypotonia	*Other symptoms*	*Method*	*Finding*
1	F	13 mo	+ +		CT	cerebellar hypoplasia heterotopias of gray matter
2	M	26 mo	+ +	torticollis	CT	vermian hypoplasia
3	M	5 mo	+	macrocephaly	CT	normal
4	F	5 mo	+ +		CT	normal
5	M	6 mo	+ +		MRI	normal
6	M	8 mo	–	cleft lip and palate	MRI	enlarged subdural space T2-hyperintensity putamen
7	F	5 mo	–	spastic hemiplegia	MRI	heterotopias of gray matter hypoplasia of corpus callosum
8	F	4 mo	–		MRI	normal

played a general developmental delay of variable severity. The children we had the opportunity to see before the age of six months showed problems of hand and eye coordination and cognitive delay, apparently resulting from their ocular motor problems; further developmental delay could not be excluded with certainty at this age. Five of our eight children (cases 1, 2, 4, 5, 6) also showed variable severe mental delay. Four of the eight have already reached school age, but only one boy was able to attend a normal school. In addition to the mental retardation two of our school-aged children had severe speech problems.

In four of eight children (cases 3, 4, 5, 8) neuroradiological findings were normal. In two children we observed abnormalities in the posterior fossa, cerebellar (case 1/Fig. 1) and vermian hypoplasia (case 2/Fig. 2), respectively. Two children were found to have heterotopias of the gray matter, combined with hypoplasia of the cerebellum (case 1/Fig. 3) or with hypoplasia of the corpus callosum (case 7/Figs. 4 and 5). In one boy we observed symmetrical hyperintensity in the putamen in T2-weighted images, probably due to delayed maturation (case 6).

DISCUSSION

Since Cogan described congenital ocular motor apraxia in 1952 a number of cases have been

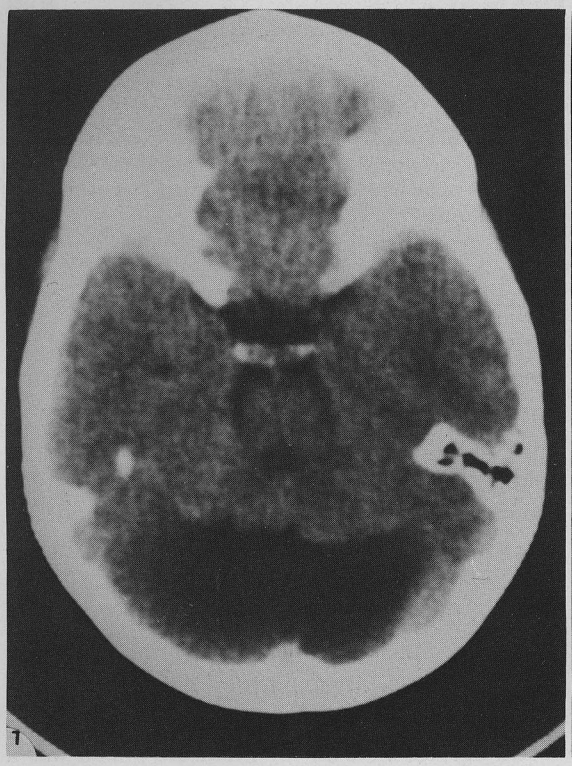

Fig. 1. Hypoplasia of cerebellar vermis and hemispheres, enlarged cisterna magna. CT, axial section, case 1.

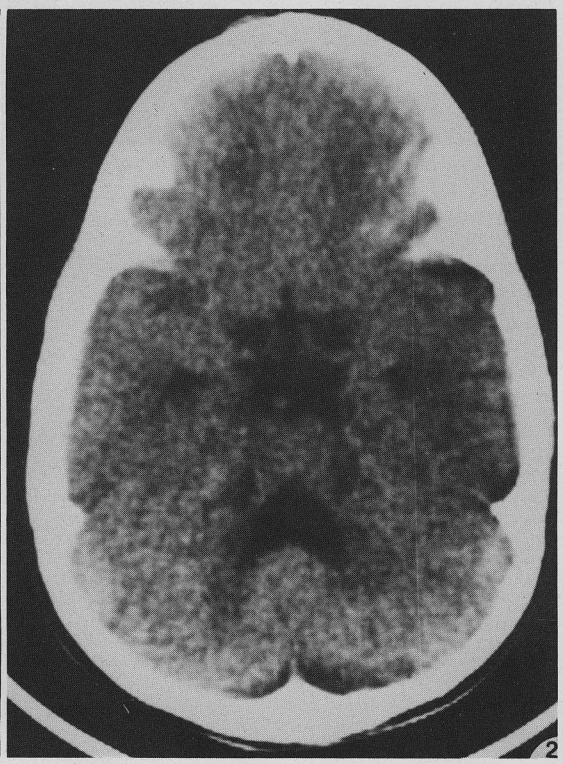

Fig. 2. Enlarged fourth ventricle, vermian hypoplasia. CT, axial section, case 2.

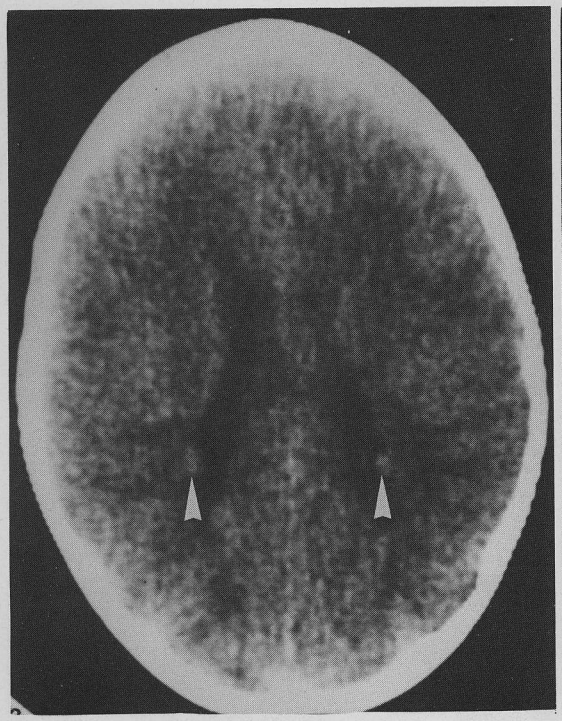

Fig. 3. Evidence of heterotopias of gray matter in periventricular area. CT, axial section, case 1.

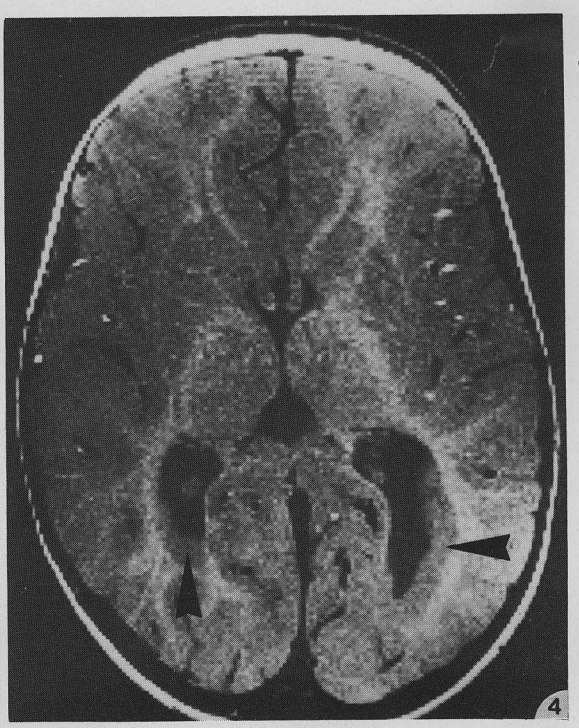

Fig. 4. Heterotopia of gray matter around both occipital horns, hypoplasia of corpus callosum. MRI-T1-weighted axial section, case 7.

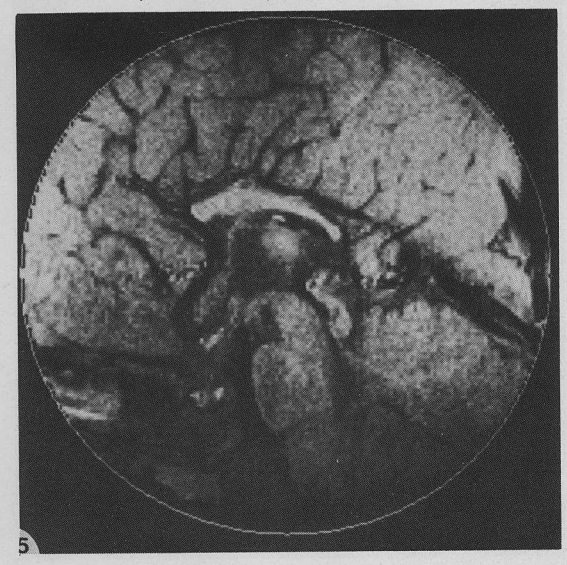

Fig. 5. Hypoplasia of the corpus callosum with absent genu and thin, small splenium. MRI-T1-weighted sagittal section, case 7.

published[1-5, 12-14]. The children usually come to attention between the ages of four and 12 months, because of suspicion of reduced vision or developmental delay. Around six months of age they develop the 'strategy' of the typical overshooting head movements. The ocular signs tend to improve with increasing age and at school age they may be very distinct or even may have disappeared. In our series of eight children we confirmed these findings. But most infants showed relevant impairment of other neurological signs such as ataxia, developmental delay and speech problems.

In agreement with the literature ataxia was a common (but not constant) finding in COMA in our series, usually combined with hypotonia and

motor delay. Although the motor developmental problems are somewhat improving until school age, ataxia persists in most cases.

There are scant reports about general development of children with COMA[2, 4, 5], and we are not aware of any reports on developmental testing. In our series we found variable developmental delay – other than motor – in six of eight children; half of them were below one year of age.

Fielder *et al.*[2] reported about special speech problems – we can confirm this in two of our cases, although only four of our children were beyond one year of age.

Up to now neither the exact pathogenesis nor the localization of COMA are known. Zee *et al.*[1], Pierrot-Deseilligny *et al.*[6-8] and others pointed out several possible locations for the disorder: paramedian pontine reticular formation, colliculi superiores, pons, cerebellum; but none of them could be proven. Eda *et al.*[3] reported abnormalities in the posterior fossa in the CT-examination of all their four cases. We were not able to confirm these findings, but we agree with Fielder *et al.*[2] who showed a great variability of neuroradiological findings in eight patients. We found cerebellar malformations, hypoplasia of the corpus callosum, heterotopia of the gray matter and maturation disorder in the basal ganglia – in variable combinations.

In only two of our five cases with ataxia did we find cerebellar hypoplasia as a possible explanation for the clinical symptoms. Heterotopias of the gray matter and hypoplasia of the corpus callosum could not be connected for certain with any clinical findings, as both conditions are well known to exist without any specific pathological symptoms.

Therefore we did not find significant correlations between neurodevelopmental disturbances and neuroradiological findings. We assume that COMA is not a well-defined entity, but rather a symptom which occurs in association with different disorders of maturation and/or malformation of the brain.

In COMA further neurological, developmental as well as neuroradiological investigations are indicated for a better understanding of its variability and long-term consequences.

ACKNOWLEDGEMENTS

Supported by Swiss National Foundation grant No. 3.917-0.88.

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