Congenital Ocular Motor Apraxia: Sporadic and Familial
Support for Natural Resolution

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Four patients aged between 9 months and 17 years were detected to have congenital ocular motor apraxia (COMA) over a 10-month period. Three of them were siblings. All exhibited the classical signs of a horizontal saccadic palsy. However, the signs were less pronounced with increasing age of the patient. This supports the observations of other authors who have noted an age-related resolution. There was no evidence of other motor developmental delay in any of the patients and computed tomography revealed no abnormalities. Both these findings are in contradistinction to prior reports of central nervous system abnormalities and motor and speech difficulties in COMA. The family of three appears to be the largest sibling cluster reported so far. Key Words: Congenital ocular motor apraxia—Familial—Resolution.

Cogan (1) first described a condition, beginning between the ages of 4 and 7 months (2), characterized by an inability to execute normal voluntary horizontal saccades when the head is stationary. Head thrusts are used to accomplish ocular refixations, and this may give the appearance of spontaneous random movements. Horizontal saccades used in both voluntary movements as well as in the quick phase of vestibular nystagmus are defective, leading some to coin the term "congenital saccadic palsy" (3). Smooth pursuit and all vertical gaze movements are preserved. We report a series of four cases, three of them siblings, to illustrate the familial nature of the disorder and its tendency to improve with age.

CASE HISTORIES

Case 1
A 9-month-old boy from Najran, South Western Saudi Arabia was referred for "abnormal eye movements/squint," which had begun a few months ago. He was the firstborn and there was no significant natal nor family history. The eyes were straight. He showed head thrusts to either side spontaneously. On attracting his attention with toys to either side, head thrusts with overshoot were followed by refixation and realignment of the head. The first phase of the optokinetic nystagmus (OKN) was abnormal, whereas the slow phase and both phases of the vertical OKN were normal. Developmentally, he had reached all corresponding milestones with no delay.

Case 2
A 10-year-old boy from Qatif, Eastern Saudi Arabia sought relief from itchy eyes and persistent
rubbing of the eyelids. On observing him, he manifested small, “spontaneous” head thrusts in either direction. Each thrust was preceded by a blink. In response to commands, typical absence of horizontal saccades were demonstrated and recorded on videofilm. Smooth pursuit movements (SMP) and vertical gaze were executed with ease. Closer monitoring suggested that his rubbing of the eyes were substitutes to blinks at the beginning of refixations. A computed tomography (CT) scan revealed no abnormalities. The neurologic examination was otherwise normal. He had six siblings; one older sister and an older brother had had similar problems in the past. Permission was sought from and granted by the father to bring them for an examination.

Case 3

The 15-year-old sister of Case 2. She showed no abnormal spontaneous movements. She studiously avoided voluntary refixations, keeping her head and gaze straight ahead. In this fashion, she had learnt to curb the cosmically embarrassing head thrust. On command, a small head thrust accompanied by a barely perceptible blink resulted in a slow, strained limited horizontal saccade. All other movements were full.

Case 4

The 17-year-old brother of Cases 2 and 3. No abnormal spontaneous movements were present. Voluntary and command horizontal saccades were executed in a slow, strained fashion to full excursion. Toward end-gaze positions, the globes appeared to retract and the eyelids quivered, in obvious effort. Neither head thrust nor blink accompanied the saccadic movements. Spontaneous pursuit movements and vertical gaze were normal.

DISCUSSION

In its commonest form, congenital ocular motor apraxia (COMA) is a pure horizontal saccadic palsy. The head moves toward the position of the new target. An active, uninhibited vestibuloocular system causes the eyes to rotate conjugally in the opposite direction. The head has to overshoot the intended target to enable the contraversively deviated eyes to fixate on it. While maintaining fixation, the head slowly moves back until the eyes are straight forward. The eyelids close at the onset of the head movement as if to lower the gain of the vestibulo-ocular reflex and thus reduce the amplitude of the head thrust. This defect is always bidirectional, although it may be asymmetrical (2,4). As the head thrusts do not develop until 4 months of age, or until head control is achieved, COMA may be mistaken for cortical blindness, with the child seemingly unable to “look” at an object held away from straight gaze (5). Rarely, the disorder is a vertical one (6).

The structural defect in COMA is not known, although associated abnormalities in the form of gray matter heterotopias and hypoplasia of the corpus callosum, cerebrum, and cerebellum have been demonstrated (2,4,7). Infrequently, a brainstem neoplasm has mimicked the condition (8). Hence CT scans of these cases were carried out revealing no abnormalities.

A familial occurrence is unusual (7,9,10). Autosomal dominant, recessive, and sex-linked inheritance patterns have been postulated. Marriages in this region of Saudi Arabia occur within closely knit communities. An autosomal recessive pattern of inheritance could be responsible for the involvement of just three siblings of a large family. A prospective long-term study of succeeding generations is necessary to reach a definitive conclusion. Fielder and coworkers (2) have reported in their series a sibling with partial agenesis of the posterior portion of the corpus callosum and hypoplasia of the right cerebellar hemisphere. This child died at the age of 3.5 years without manifesting the clinical signs of COMA. Thus, there appears to be the possibility of other family members with asymptomatic COMA. Our series of three siblings appears to be the largest single-family cluster so far.

Successive family members have shown a remarkable and progressive reduction in abnormal signs together with a strengthening of the horizontal saccades. The unrelated infant presented a dramatic clinical picture with violent head thrusts. The youngest sibling showed all the classical signs of a saccadic palsy, minus the violent thrusts. The sister could execute a horizontal saccade extremely slowly and to a small degree. With this partial recovery, the compensatory blink at onset was insignificant and barely noticeable. The eldest sibling was practically normal, with the exception of a slow, strained saccade albeit to a full degree. This phenomenon of improvement points to an age-related resolution of the disorder (2,11,12). The oldest patient described so far has been a 34-year-old man belonging to the original series of Cogan and associates (12). Despite “acquiring considerable vestibular and optokinetic responsiveness . . . he still had difficulty in mobilizing saccades.” Case 4 shows that resolution is possible at an even younger age.
It is tempting to speculate upon a delayed maturation of neural pathways as being the underly­ing defect in COMA. This is supported by the ob­servations that most affected children have delayed motor and speech development as well (2, 13). The cognitive development is commonly impaired, and many children require a special scholastic education even after the signs of COMA have begun to regress. The three siblings are doing quite well at school.

Finally, it is unusual to come across a series of 4 cases of this uncommon disorder in a short span of 10 months.

REFERENCES