Unilateral Congenital Ocular Motor Apraxia: A Case Report

Woo Jung Kim, M.D., Bong Leen Chang, M.D.
Department of Ophthalmology, College of Medicine, Seoul National University, Seoul, Korea

Congenital ocular motor apraxia (COA), first described by Cogan in 1953, is a rare disorder which shows characteristic defects of the horizontal voluntary saccades, and compensatory head thrust. Until now, most cases have showed a presumably congenital origin, bilaterality, and a tendency to various stages of recovery with aging. But the cause and mechanism of COA are not completely known. Occasionally, it combines with other neurologic abnormalities and metabolic diseases such as Gaucher's disease exhibit similar clinical characteristics to COA. We recently experienced a case of a 3-year-old girl who showed the clinical features of unilateral congenital ocular motor apraxia.

Key words: head thrust, unilateral congenital ocular motor apraxia, voluntary saccades.

INTRODUCTION

Apraxia is a term that means a defect in voluntary movement with save of other motor functions, to be distinguished from paralysis which means a defect in all movements.

In 1953, Cogan first described congenital ocular motor apraxia in 4 of his patients who showed a defect in voluntary ocular movement and compensatory head thrust.

This rare congenital anomaly is found in conditions which initiate horizontal gaze on both sides voluntarily. It seems to be caused by a defect of the supranuclear pathways with improving tendency as one grows older, but the location and mechanism of the lesion are not yet completely known.

Due to its typical clinical features, it is not hard to make a diagnosis. However, there may be combined neurological anomalies, and confusion with metabolic diseases such as Gaucher's disease.

In this article, we report a unilateral case of congenital ocular motor apraxia in a 3-year-old girl who presented with abnormal head turning to the right side.

CASE REPORT

A 42-month-old girl visited our pediatric ophthalmic clinic in Feb. 6th, 1991. Her parents complained that she showed abnormal head thrust to the right side when she tried to fix her eye to the right. From her past medical history, she was born one of twins, premature with a birth weight of 1950 gram, and brought up in an incubator for one month. The other twin died immediately after birth. At 4-months, she was hospitalized for 1 month due to meningitis.

General pediatric examination revealed delayed development of speech and intelligence compared with others of her age. There were normal findings in movement of the extremities, gait, and deep tendon reflex. But, her parents reported the history of her right side movement difficulty and improvement at the time of visit. On ophthalmic examination, visual acuity and re-
Fig. 1. The eyes are ortho in primary position.

Fig. 2. Normal voluntary eye movement to the left.

Fig. 3. When the patient is attracted in her right visual field (a) the head overshoots with blinking and eye levoversion. (b) Once fixation is obtained, (c) the head returned to the visual target and the eyes slowly achieved the central position in the orbits.

The refractive state of both eyes were almost the same and the eye position was ortho by corneal reflex test in primary position (Fig. 1). On ocular movement examination, voluntary movement to the left was normal (Fig. 2). But, when the patient was attracted in the right visual field, the head overshot toward the right side with blinking and levoversion of the eyes, instead of normal voluntary movement (Fig. 3a). Once fixation was obtained (Fig. 3b), the head returned to the visual target and the eyes slowly achieved a central position in the orbits (Fig. 3c). Vertical saccades were normal. Extraocular muscular paralysis was not found. Optokinetic nystagmus toward the right side was not found. No other pathologic findings were shown in fundus examination, visual evoked potential (VEP), magnetic resonance imaging (MRI) or sleep electroencephalogram (EEG).
DISCUSSION

The voluntary and saccadic movement of the eye is normally composed of version toward stimulus and following head turning to the same side. In ocular motor apraxia, the pathologic site is believed to be located on the supranuclear pathway reaching the paramedian pontine reticular formation (PPRF), as a result, a defect in voluntary ocular movement occurs. Additionally, to compensate this defect, typical reflex induced saccades, following blinking, are evoked by rapid turning of the head to the stimuli. In congenital form, the abnormality is found in both sides of the horizontal plane, and involuntary or pursuit movement is preserved. The saccadic velocity is also intact.

Generally, blinking occurs with saccadic movement or independently, but in COA, it precedes rapid head turning. The eyes get the position of fixation from the vestibular reflex following the overshooting head to a target. Once fixation is obtained, the head returns to the visual target and the eyes slowly achieve the central position in the orbits. That is, this phenomena is an adaptive strategy to provoke the gaze change. In severe cases, the quick phase of vestibular and optokinetic nystagmus might be also disturbed.

The cause and mechanism are not completely known, but pathologic findings in the cortical or subcortical areas of the frontal lobe have been exhibited, and possible lesions in the brain stem or cerebellum have been presented. Concurrence in identical twins or in one family has been reported.

To rule out intracranial lesions, a careful review of magnetic resonance images of the brain was performed, but no abnormality was found. Regarding the previous history of meningitis in the infant and the right hemiplegia noticed by her parents, the possibility of acquired ocular motor apraxia combined with localized lesion may be assumed. However, acquired ocular motor apraxia has been reported as a defect of voluntary movement in all directions until now.

The ability of voluntary movement to fix a visual target is considered a learned phenomenon, and the typical head thrust shown in COA is a form of compensation to overcome this defect. Therefore, COA might be missed before the age of 3 years when a normal baby can control his or her head, rather it can be misdiagnosed as visual impairment but can be differentiated by visual evoked potential test (VEP) findings.

As in our case, history of prematurity, hard labor, perinatal hypoxia, ischemia, and delayed psychomotor development are combined frequently in COA. Moreover, strabismus and impairment of motor control might also be found.

Though there is no difficulty in making a diagnosis due to the obvious clinical features attention should be given to other combined abnormalities and differential diagnosis. On radiologic examination, abnormal findings of interhemispheric connection including agenesis of the corpus callosum might be shown. Moreover, brain stem tumor, lesion in the occipital lobe, medulloblastoma, and abnormality in the brain stem or cerebellar vermis have been reported. But in most COA cases, localized abnormalities are not found as in this case.

As an important differential diagnosis, Gaucher’s disease, inborn error metabolism of glucocerebrosidase, reveal similar features to COA, but it is differentiated from COA by the symptoms and signs of the central nervous system, and visceromegaly. In addition, congenital and acquired vertical ocular motor apraxia have been reported.

In COA, as one grows older, apraxia more or less improves and exaggerated blinking may be combined. This fact is also believed to be related to reflex induced saccades. As in our case, attention to the possibility of combined psychomotor disturbance is also needed.

In the future, more studies will be required to elaborate the cause and mechanism of COA.

REFERENCES

3. Orrison, W. W., and Robertson, Jr. W. C.:
OCULAR MOTOR APRAXIA


