Familial Congenital Ocular Motor Apraxia

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Congenital ocular motor apraxia (COMA) is a unique ocular motor disorder which is characterized by a deficit in initiation of voluntary horizontal eye movement with reserved reflex eye movement. Although a portion of cases with COMA were found to be associated with other abnormalities, COMA in most patients is an isolated disorder. The most characteristic appearance of these patients is compensatory head thrusts which usually become less evident with increasing age.

Since Cogan first described COMA in 1952, many cases have been reported. The majority of these occurred sporadically with only a few exceptions. We report on 4 patients with COMA. Two of them were siblings, and the other 2 patients were father and daughter. The ocular motility status is described in detail. (Chang Gung Med J 2002;25:411-4)

Key words: congenital ocular motor apraxia, familial.

Congenital ocular motor apraxia (COMA) has also been labeled "congenital saccade initiation failure", "congenital saccadic palsy" or "Cogan's apraxia" after the first description by Cogan in 1952.(1) It is thought to be a supranuclear disorder of the initiation of voluntary horizontal saccade. Voluntary horizontal pursuit is not necessarily normal, but vertical eye movement is exclusively normal. The vestibulo-ocular reflex is preserved but may be defective.(2) The most characteristic appearance of affected children is the compensatory head thrusting as neck control develops at the age of 3 to 4 months. Patients use the vestibulo-ocular reflex to rotate the eyes to fixate on targets. While maintaining fixation on the target, the head slowly moves back until the eyes are pointing straight forward.

Because of the difficulty with voluntary eye movements, patients with COMA usually have difficulty reading. Delayed developmental milestones, oral motor difficulties, and being clumsy with the hands may also be present with this disorder.(2-4) Fortunately, this disorder tends to spontaneously resolve in the first decade of life. Most adult patients have only occasional random head thrusts, but many still have difficulty reading as well as oral motor problems.(3,5)

Classically, COMA is considered to be a sporadic disorder. Familial cases have been reported but are unusual in occurrence. We diagnosed COMA in 2 siblings and a girl and her parent. Results of motility tests are also presented in detail.

CASE REPORTS

Cases 1, 2

A 3.5-year-old boy and his 5-month-old younger brother were brought to the first author's clinic by their mother with the chief problem of 'odd head movements' since 3 months old in the older boy. An 'undetermined neurologic problem' was the diagnosis of a neurologist, and psychotherapy was suggested by an ophthalmologist for the elder brother. The worried mother was also concerned about the delayed motor development in her first son.

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Since her second son showed the same problem of head thrusting a few days before the visit, she was determined to find an answer.

On examination, both children appeared to be normal healthy boys. Voluntary horizontal saccade was absent from both boys. Voluntary pursuit was present but hypometric in the elder brother and totally absent in the younger brother. The hand-held optokinetic drum induced only random responses in the older brother and no response at all in the younger brother. Vertical eye movements were normal in both children.

Because the mother was concerned about central nervous system problems, the elder brother underwent a magnetic resonance imaging study of the brain. Due to the normal results of the imaging study, no further testing was performed on the younger brother. The mother denied a family history of this disorder.

**Cases 3, 4**

A 1-year-old girl was brought to the second author’s clinic by her parents with the problem of head thrusts since 3 months old. The mother claimed that the girl also had delayed development. Horizontal voluntary saccade was absent, and pursuit was present but hypometric. The optokinetic drum induced only occasional responses. Vertical eye movements were normal.

The father showed occasional head thrusts in the clinic and admitted that he had had similar problems since he was a child. He refused to be evaluated, and we could get no family history from him. The girl was their only child.

**DISCUSSION**

The pathogenesis of COMA is still obscure. Although many associated neurological abnormalities (absence or hypoplasia of the corpus callosum and/or inferior vermis, for example) have been reported, most cases are generally healthy and have normal mental development.\(^6\)\(^-\)\(^9\) No neuropathologic studies have been made on specimens obtained from autopsies, but delayed myelination of the ocular motor pathways for conjugate gaze is a reasonable conjecture.\(^10\)

The first case of COMA was described by Cogan in 1952, but the occurrence of COMA among family members was not reported until 1966 by Robles.\(^11\) At least an additional 17 families with COMA have been reported since then.\(^2\)\(^,\)\(^5\)\(^,\)\(^10\)\(^-\)\(^18\) Most of the reports describe affected siblings or identical twins and unaffected parents. Only 4 reports have documented COMA in 2 successive generations of a single family. Cogan reported a mother and 2 sons with this disorder.\(^10\) The pedigree was not available. Vassella described an affected father and daughter.\(^2\) Noticeably, the father and mother were first cousins. The father had normal parents and a normal younger sister. The mother's parents, 1 brother, and 2 sisters were all normal. The affected daughter had a normal brother. Phillips diagnosed COMA in 4 children of an affected father.\(^5\) The father was adopted, and the medical histories of his biological parents were unknown. He had 4 children, including 1 boy and 3 girls, in total. All of the children were affected. Borchert and associates also reported a possible instance which included a pair of identical twins with COMA whose father "appeared to have subtle abnormalities in saccade". The father would not allow the author to evaluate him, and the pedigree was also unavailable.\(^11\)

We report on COMA in 2 families. The first family included 2 affected siblings which is not unusual in the literature. However, the second family included an affected father and daughter which is the fifth documented family, to our knowledge, with COMA in 2 successive generations. Because the pedigrees of the affected families are not available in most reports, including the present one, it is difficult to determine the transmission modes.

In conclusion, due to its tendency for spontaneous resolution and the possible existence of mild forms of this disorder, the familial incidence of COMA is highly likely to be greatly underestimated. Parents and siblings of a child with COMA should be routinely examined for possible residual signs of this disorder and questioned about a history of childhood symptoms to begin to elucidate the modes of transmission.

**REFERENCES**

2. Vassella F, Lutschg J, Mumenthaler M. Cogan's congeni-
家族性先天眼球運動失用症

徐紅娜  楊孟玲  賴慧群

先天性眼球運動失用症 (congenital ocular motor apraxia) 是一種少見的眼球運動障礙。其特徵為自主性眼球運動啓動困難，但反射性眼球運動則可能正常。垂直性運動除了常見病例報告外均為正常。病患之外觀特徵明顯 - 代償性頭部甩運動，以前庭眼球反射來達到眼球轉動之目的。罹病者常有閱讀困難及學習障礙。幸運的是，絕大多數病患隨著年齡增長有逐漸改善之趨勢。

自從西元1952年首例報告以來，陸續有零星病例被提出。其中除了少數為家族性外，均為散發性。本報告將提出四位病患，其中二位為兄弟，另外二位則為父女。對於其眼球運動檢查結果亦有詳細描述。(長庚醫誌 2002;25:411-4)

關鍵字：先天性眼球運動失用症，家族性。