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Visual Development in a Patient with Infantile-Onset Saccade Initiation Delay

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Abstract **Case Report**

Congenital ocular motor apraxia (COMA), also known as infantile-onset saccade initiation delay (ISID), is characterized by the inability to initiate volitional horizontal saccades. This five-month-old girl showed constant rapid movements to right or left one week after birth. After four months, she began thrusting her head from left to right when following target objects. She was diagnosed with COMA/ISID. At 4 years of age, head thrust became less evident. She first uttered meaningful words at 5 years of age, and spoke in sentences by 6 years of age. At 6 years of age, visual acuity was 1.0 in both eyes at 1 m. Head thrust disappeared at almost 6 years of age. She could not start crawling and sustaining a sitting position due to hypotonia. Clinicians should be aware of visual development in COMA/ISID and should look for them during the assessment of these children.

Keywords: Infantile-onset saccade initiation delay, congenital ocular motor apraxia, head thrust.

INTRODUCTION

Congenital ocular motor apraxia (COMA), also known as infantile-onset saccade initiation delay (ISID), is characterized by the inability to initiate volitional horizontal saccades [1, 2]. Developmental delay, hypotonia, and ataxia occur frequently in COMA/ISID, suggesting more global brain impairment and not just a saccadic disorder.

There are few reports of the development of visual functions [1-5]. Here, we report visual development in a patient with COMA/ISID. The pediatric aspect including video record of this case was previously described [6].

CASE REPORT

This five-month-old girl who was born at 40 gestational weeks with a body weight of 3398 g with tetralogy of Fallot showed constant rapid movements to right or left one week after birth. Therefore, her parents could not establish eye contact with her. After four months, she began thrusting her head from left to right when following target objects. Her limb movements did not exhibit ataxia or tremor. Ophthalmological examination did not show any abnormalities in the retina. Electroencephalography and visual evoked potentials were normal. Brain magnetic resonance imaging did not show any abnormalities, such as elongated superior cerebellar peduncles or hypoplasia of the cerebellum and brainstem observed in Joubert syndrome. She was diagnosed with COMA/ISID. Mutations were not observed in *APTX*, *ATM*, *PIK3R5*, *PNKP*, and *SETX* genes, which reportedly account for most cases of this disorder.

At 11 months of age, her visual acuity was 20/270 in both eyes examined by Teller Acuity Card at 38 cm. At 2 years of age, head thrust became less evident. At 3 years and 6 months of age, she became interested in things around her and could understand the meaning of words. At 4 years of age, head thrust became less evident. She first uttered meaningful words at 5 years of age, and spoke in sentences by 6 years of age. At 6 years of age, visual acuity was 1.0 in both eyes at 1 m. Head thrust disappeared at almost 6 years of age. However, she could not crawl due to hypotonia. She was also unable to sustain a sitting position and was unable to stand.

DISCUSSION

Developmental abnormalities may occur in COMA/ISID including motor delay, speech or language delay, and cognitive delay. However, there are few reports of the long-term development of visual function and intellectual prognosis in COMA/ISID [1-5].

Salman [1] analyzed 66 articles with information on 288 patients with COMA. According to their review, head thrusts were reported in 84.7%. Blinks without head thrusts were used to initiate saccades in 41%. The fast phases of the optokinetic response and vestibulo-ocular reflex were impaired in 69.8% and 34.4% respectively. Smooth ocular pursuit was abnormal in 33%. Global developmental delay occurred in 41.3%, speech or language delay in 36.5%, cognitive delay in 17%, hypotonia in 35.8%, motor delay in 48.6%, and ataxia/clumsiness in 49.3% of patients. Neuroimaging was performed on 197 patients and was normal in 39.1%. Abnormalities involved the cerebellum (24.9%), cerebrum (15.7%), other infratentorial structures (11.7%), and corpus callosum (6.1%).

Head thrusts are a common, but not universal finding in COMA/ISID [1, 2]. They appear from infancy and gradually get improvements. There is a clear association between delayed development and COMA/ISID in about 40% of the patients [1]. Although normal development is often attained, patients who start walking after the age of two years may have intellectual disabilities and delayed linguistic development [2]. It can be global, or selectively involve motor skills, speech and language, cognition or reading. The frequency of these abnormalities points to a more widespread brain dysfunction in COMA/ISID than is currently appreciated. In this present patient, she could not start crawling and sustaining a sitting position due to hypotonia.

CONCLUSION

Clinicians should be aware of visual development in COMA/ISID and should look for them during the assessment of these children.

DISCLOSURE

The author declares no conflict of interest.

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