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Visual Diagnosis

Involuntary Eye Movements Accompanied by Head Thrusting to View Objects

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This five-month-old girl 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 (Fig. 1 and 2). 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 congenital oculomotor apraxia.[1,2] Mutations were not observed in APTX, ATM, PIK3R5, PNKP, and SETX genes, which reportedly account for most cases of this disorder.[2]

Congenital oculomotor apraxia is characterized by the impairment of voluntary saccadic eye movements in the horizontal direction. One feature is "head thrust," wherein patients cannot voluntarily turn their eyes to watch objects and therefore, they rotate their heads toward the target object but often overshoot it. The eyes are then dragged toward the target position. Finally, patients turn their heads to the opposite side of where the eyes

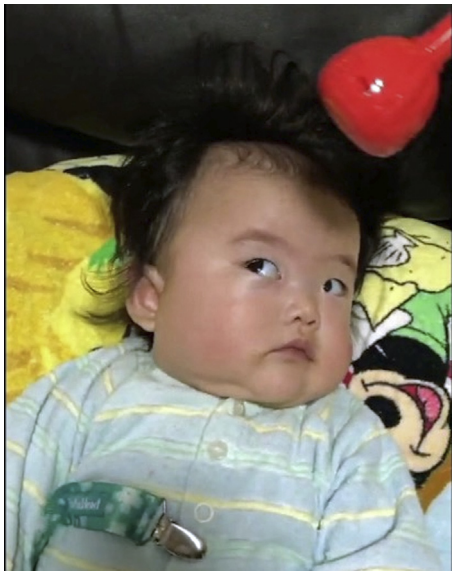

FIGURE 1. The patient rotates her head toward and overshoots the target object (left). The eyes are then dragged to the target position. Her head turns abruptly toward the opposite side where her eyes remain to gaze at the object. The video associated with this figure can be viewed online at doi:10.1016/j.pediatrneurol.2018.09.014.

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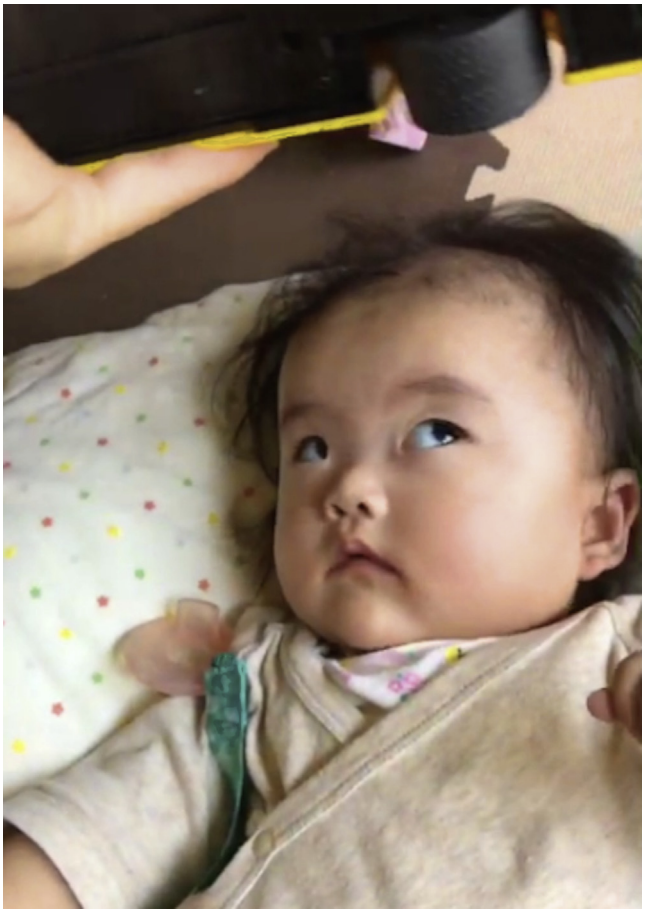

FIGURE 2. The eyes are then dragged to the target position. Her head turns abruptly toward the opposite side where her eyes remain to gaze at the object. The video associated with this figure can be viewed at doi:10.1016/j.pediatrneurol.2018.09.014.

remain to gaze using the vestibulo-ocular reflex. In infants, loss of vision is anticipated because patients do not follow the object, and physicians often misdiagnose cortical blindness. The pathogenesis remains either unknown or is considered to involve the brainstem or cerebellum or paramedian pontine reticular formation.[2,3] Although normal development is often attained, patients who start walking after the age of two years may have intellectual disabilities and delayed linguistic development.[3]

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