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Letter to the Editor: New Observation

Persistence of Infantile-Onset Saccade Initiation Delay (Congenital Ocular Motor Apraxia): An Update on a Young Adult

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Since its first description by Cogan in 1952,[1] much remains unknown about the pathophysiology and natural history of infantile-onset saccade initiation delay (ISID) (previously and erroneously known as congenital ocular motor apraxia).[2,3] The diagnosis is typically made in early infancy when head thrusts (after the infant achieves head control) or blinks are used to rapidly change the direction of gaze. Sparse case reports/series on the outcome of ISID are available in patients in late adolescence and adulthood.[3] (supplementary table), [4-7] There is an optimistic perception that it tends to attenuate or even resolves with age.[6,7] The reality is that while head thrusts to initiate saccades may become inconspicuous by early childhood and well before the end of the first decade, blinks are still needed to initiate medium to large amplitude (i.e., >20 degrees) saccades,[4,5,7] when the ocular motor system is examined carefully.

In 2010, we reported a case of a 7-year-old boy diagnosed just before the age 3 years with ISID. He also had mild developmental delays and clumsiness without ataxia. His brain MRI showed thinning of the intercollicular commissure, a unique neuroimaging finding.[8] In this report, we give an update on the long-term outcome of his condition and on his general daily functioning.

Case report update: At the age of 20 years, he still displayed horizontal head thrusts or blinks when he shifted his gaze rapidly horizontally either to the right or the left side. This has been stable over the years. He finished Grade 11. He has learning difficulties. He finds copying from the board hard because of his inability to "move his eyes fast." He is behind in Math by about two grades. He has a partial educational assistant and is enrolled in a Life Skills program. He has a driving license. A recent brain MRI just before his 20th birthday showed that his thin intercollicular commissure is stable with no new findings (Figure 1). On examination, his near visual acuity with glasses was 20/25 in each eye. His visual fields were normal. Pupils were equal and reactive to light. His funduscopy exam was normal.

His smooth pursuit was normal horizontally and vertically at slow visual target speed. Saccadic smooth pursuit with fast target speed was noted only horizontally. Convergence was normal. He displayed blinks or head thrusts horizontally intermittently when he shifted his gaze rapidly between right and left side for medium amplitude saccades (20–30 degrees). Vertical saccades were normal. Small amplitude (i.e., 5–10 degrees) saccades were performed normally. He only rarely performed medium to large amplitude saccades without head thrusts or blinks. When asked not to blink or move his head, his horizontal saccade velocity was slow.

His tone, strength, upper limbs coordination, and reflexes were normal. His sensation to pin prick, joint position sense, vibration sense, and touch were normal. He walked normally; however, heel to toe walking was clumsy/borderline ataxic.

ISID is generally said to improve or resolve with time;[6,7] however, this improvement was either not defined[6] or was defined as the ability to make saccades "with or without blinks".[7] We think that this conclusion is not correct since these patients are still using blinks to initiate saccades, at least sometimes. As they got older, there was a switch in strategy from head thrusts to blinks when medium or large amplitude saccades >20 degrees are made.

One can only conclude that - ISID has resolved when all the following three conditions are met: 1) no head thrusts are seen during the majority of medium-large range saccades (>20 degrees), 2) with the head restrained, no blinks are seen when most saccades are made, and 3) saccades' velocity remains fast when patients are asked not to blink or move their head while performing medium-large range saccades.

On the other hand, when a patient's attempt to initiate horizontal saccades changes from frequent head thrusts (typically as an infant) to occasional blinks with saccades (usually later on during childhood) then one can state that their condition has improved but not resolved.

In our 20-year-old patient, ISID has persisted since infancy. In addition, he continues to have learning difficulties and clumsiness. However, he functions reasonably well, can hold long conversations during his clinic visits, is independent, and can perform all activities of daily living without difficulty. He has a driving license.

In summary, a careful assessment of the ocular motor system may reveal that some clinical features of ISID can persist in adolescents and young adults.[5–7] It remains to be seen if ISID resolves in patients with normal brain MRI. An extensive review on ISID's features, neuroimaging findings, and outcomes is available for the interested reader.[3]

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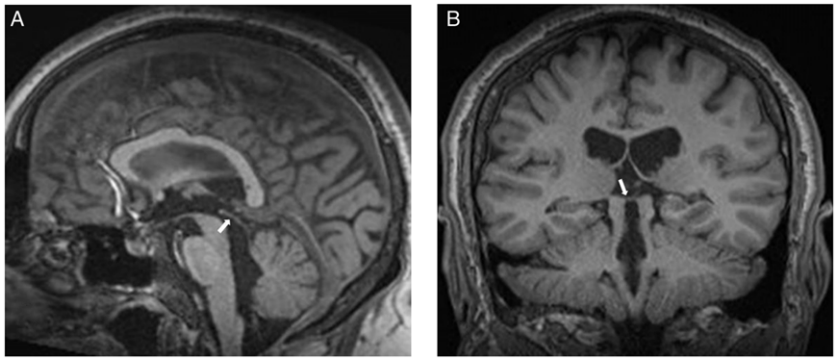

Figure 1: Midsagittal (A) and coronal at the level of the colliculi 3D MP RAGE (magnetization-prepared rapid gradient-echo imaging) (B) T1-weighted images (TR:1940, TE: 3.08) demonstrate thinning of the tectal plate/intercollicular commissure (arrows).

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Martin Bunge contributed the MRI figure and its legend and edited (review and critique) the manuscript.

Consent and Ethics Approval. The patient gave verbal and written consent for the case to be published. Ethical approval for reporting the case was given by the Research Ethics Board of the University of Manitoba.

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