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SCIENTIFIC LETTER

Aphonia in Infantile Tremor Syndrome

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To the Editor: A 9-mo-old girl presented with regression of milestones for 2 mo. She had lost her ability to vocalize, and had a silent cry for 2 wk. She was exclusively breastfed by a vegetarian mother. On examination, she had pallor, sparse thin depigmented hairs, and hyperpigmentation over dorsum of hands and feet. Her serum vitamin B12 (200 mmol/L) was low and plasma homocysteine (24 pg/ml) was elevated; blood thiamine levels were normal (0.44 µg/dl). She was treated with intramuscular Hydroxycobalamin 1 mg per day. On day 3 of treatment, she developed tremors, and on day 7, started to regain her cry. At follow-up 6 mo later, she had gained milestones, and her voice was normal.

Another 15-mo-old girl presented with global developmental delay with regression of milestones for 3 mo. She had lost her ability to vocalize, and had a silent cry 10 d. The baby had been exclusively breastfed by a vegetarian mother. On examination, she was lethargic, had depigmented scalp hair, and hyperpigmentation over dorsum of hands and feet. Her serum vitamin B12 was very low (50 mmol/L), plasma homocysteine was high (172 pg/ml), and thiamine levels were normal (0.40 µg/dl). On Hydroxycobalamin 1 mg/d, she developed tremors and orofacio-lingual dyskinesis on day 4 and on day 7, child regained her cry. While on nasogastric feeds, she was orally fed and possibly aspirated and died at home on day 10.

Aphonia though reported in thiamine deficiency (infantile *beriberi*) [1], has not been described in vitamin B12 deficiency. We postulate that severe vitamin B12 deficiency leads to aphonia due to dysfunction of basal ganglia. Globus pallidi involvement has been documented in vitamin B12 deficiency [2]. Bilateral lesions of the Globus pallidi can interrupt the flow of impulses in the striato-pallido-thalamo-cortical loop and to the phonatory motor nuclei (nucleus ambiguus), resulting in vocal fold paralysis [3, 4].

Declarations

Conflict of Interest None.

References

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