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CORRESPONDENCE

Infantile Tremor Syndrome Masquerading as Glutaric Aciduria Type 1

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To the Editor: Infantile tremor syndrome (ITS) is characterized by pallor, delayed development, tremors, and sparse hypopigmented hair [1]. Glutaric aciduria (GA) Type 1 presents with dystonia, large head size, and neuroregression [2]. Here, we are reporting a rare phenotype of ITS presenting as GA Type 1.

A 15-mo-old boy presented with developmental delay, stiffness of limbs, and tremors in the last 15 d. The child attained sitting, cooing, and a social smile by 11 mo of age. In the last month, there was a loss of the above milestones. On examination, pallor, knuckle hyperpigmentation, hypopigmented hair, normal weight (12 kg) and head circumference (47 cm), spasticity, dystonia, and tremors were noted. On investigations, low hemoglobin (7.2 g/dL) with megaloblastic anemia, low (69.59 pg/mL) vitamin B12 (normal: 211–911), and increased (45 micromol/L) homocysteine (normal: 3–15) levels were noted. MRI of the brain showed T2W hyperintensities in bilateral globus pallidus, white matter, and cerebral peduncles. On diffusion-weighted imaging, sections restricted diffusion was observed in bilateral globus pallidus and cerebral peduncles with a signal loss on apparent diffusion coefficient. Arterial blood gas, tandem mass spectrometry, gas chromatography/mass spectrometry, and exome sequencing (Supplementary Table S1 shows genes tested) were normal. Treatment with vitamin B12, was done initially intravenous 1000 µg/d (14 d) followed by intramuscular on alternate days (4 wk), and oral (6 mo). One month after treatment, the child attained new milestones, and his B12 increased (1250 pg/mL), and homocysteine level normalized (11 micromol/L).

This child had a batwing sign, prominent sylvian fissure with symmetric signal changes in basal ganglia mimicking as GA Type 1. The points that favored ITS were normal head size, skin and hair changes, and low vitamin B12 levels. The differentiation of both the conditions is important as GA Type 1 is a chronic disorder whose management is strict diet restriction as compared to ITS which requires a high protein, vitamin B12–rich diet, which comes predominantly from a nonvegetarian diet.

Supplementary Information The online version contains supplementary material available at https://doi.org/10.1007/s12098-021-04067-y.

Declarations

Conflict of Interest None.

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