Infantile Tremor Syndrome Presenting as Stroke

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To the Editor: Infantile tremor syndrome (ITS) is characterized by pallor, developmental delay, tremors, skin and hair changes [1]. We report a child of ITS presenting as stroke.

A 1-y-old boy presented with left upper and lower limb weakness of 3 d following loose stools. The child was on exclusive breast-feeding and attained only head control and social smile. Following loose stools, there was loss of social smile and neck control. On examination, weighed 8.5 kg (0 to -2 z score), length 67 cm (-2 to -3 z score), and head circumference 42.5 cm (-2 to -3 z score), lethargic with severe pallor, sparse, hypopigmented hair, and knuckle hyperpigmentation, power (medical research council grading) of 2/5 on the left side, and tremors.

On investigation, severe megaloblastic anemia (hemoglobin - 2.9 g/dL), low serum vitamin B12 levels 65 pg/mL (normal - 200–300 pg/mL), and elevated serum homocysteine level - 68 µmol/L (normal - 3–15) were noted. Mother's investigations showed megaloblastic anemia (hemoglobin - 8 g/dL), with low B12 level (120 pg/mL). MRI of the brain showed T2W hyperintensities in right globus pallidus and crus cerebri and DWI and ADC maps show diffusion restriction in the right globus pallidus and crus cerebri. Arterial blood gas, tandem mass spectrometry, gas chromatography-mass spectrometry, and exome sequencing were normal. Stroke was attributed to transient hyperhomocysteinemia secondary to vitamin B12 deficiency. With intravenous vitamin B12 1000 mcg, improvement in development, left-sided power, and hemoglobin increased to 10.5 g/dL and homocysteine level decreased to 12 µmol/L.

Here we present a case of ITS with stroke following acute gastroenteritis. We considered stroke due to inborn errors

of cobalamin metabolism; abnormalities in homocysteine metabolism, organic acidemias were excluded. The elevated homocysteine can cause increased coagulation, increased cholesterol synthesis, and oxidative stress with reduced apolipoprotein A1 synthesis which leads to endothelial injury [2, 3]. In this case, we postulate the elevated homocysteine as the cause of stroke, that reversed after Vitamin B12 administration. ITS can present as stroke, in addition to classical presentation of developmental delay, tremors, and anemia.

Declarations

Conflict of Interest None.

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