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Letter to the Editor

Nutritional Vitamin-B12 deficiency masquerading as a mitochondrial disorder

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Dear Editor,

Secondary mitochondrial dysfunction is known to occur in cobalamin deficiency.[1] Here, we report Vitamin-B12 deficiency masquerading as mitochondrial disease. An 11-month-old boy presented with decreased activity for 3 weeks, fever for 1 week, tachypnoea and refusal of feeds for the past 1 day. The child was on a predominantly breastfed diet. The child lost his attained milestones of sitting and recognition of parents for the past 3 weeks.

On examination, the child was drowsy, heart rate of 130/min, respiratory rate of 60/min, weight (9 kg), length (74 cm) and head circumference (46 cm) was normal. He had pallor, knuckle hyperpigmentation, hypopigmented sparse hair, lethargy, hypotonia, power 3/5 in all four limbs, gallop rhythm and hepatomegaly. On investigations, haemoglobin was low 2.4 g/dL, total leukocyte counts 18,300 cells and low platelet count 16,000 with high mean corpuscular volume (104 fL), high lactate 16.65 mg/dL, low Vitamin-B12 <50 pg/mL and high homocysteine levels 36.5 umol/L. Peripheral smear and bone marrow showed features of the megaloblastic picture. Arterial blood gas was suggestive of high anion gap severe metabolic acidosis (pH: 6.898, Pco2: 12 and pHCO3–: 2.3 mmol/L with base excess: 29.4). MRI brain was suggestive of cerebral atrophy. TMS was suggestive of high alanine 1105.12 (74–613) with an elevated glutamate/lysine ratio of 982.67 (211–683). Whole exome and mitochondrial genome sequencing were not shown any variants. Treated with supportive care, injection vitamin-B12, and packed red blood cell transfusion. After 14 days, the haemoglobin improved to 10.2 g/dL, TMS and pH normalised.

The above changes are probably due to elevated MMA in Vitamin-B12 deficiency which leads to inhibition of carbamoyl phosphate synthetase I, pyruvate carboxylase and the dicarboxylate carrier needed for malate shuttle.[2] Hence, Vitamin-B12 deficiency is to be considered in differentials when features are suggestive of mitochondrial disorders.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

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