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CORRESPONDENCE

Molybdenum Cofactor Deficiency (MoCD) Masquerading as Stroke-Like Episodes

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To the Editor: Molybdenum cofactor deficiency (MoCD) is characterized by neonatal-onset refractory seizures, and developmental delay [1]. In this report, we present a novel phenotype of stroke-like episodes due to MoCD.

A two-year-old boy, born to consanguineous parentage with normal birth history presented with right-sided focal seizures, right-sided weakness, and encephalopathy. His developmental milestones were delayed with attainment of neck holding at 8 mo, rolling over at 10 mo, palmar grasp at 10 mo, cooing at 8 mo, and social smile at 8 mo. On examination, a head circumference of 45 cm [<-3 standard deviation (SD)], length of 72 cm (<-3 SD) and weight of 7 kg (<-3 SD), dysmorphism in the form of frontal bossing, bitemporal wasting, elongated palpebral fissures, long philtrum, and puffy cheeks, dystonia right more than left, the power of 3/5 (MRC Grade) on right side were noted.

Investigations revealed anemia (hemoglobin- 10 mg/dl), decreased serum uric acid- 0.8 mg/dl (3.4-7) and normal homocysteine, 10.35 micmol/L (<15). Liver and renal function tests, and arterial blood gas were within normal limits. Magnetic resonance imaging of brain showed T2 hyperintensities and T1 hypointensities in the bilateral globus pallidus-left more than right, and left half of ventral midbrain with diffusion restriction on diffusion weighted imaging. Whole exome sequencing revealed a known homozygous splice acceptor variant c.646-6G>A in Intron-5 of the *MOCS1* gene, thus confirming the diagnosis. The child was treated with levetiracetam, vitamin B12, pyridoxine, low-protein diet and supportive management.

The clinical and radiological manifestations arise due to sulfite accumulation and cases are often misdiagnosed. Sulfite accumulation results in deranged cellular metabolism, impedes the biosynthesis of sphingolipids for myelination and has neurotoxic effects [2]. Urine sulfite dipstick test may serve to screen neonates for this disease [3]. To conclude, MoCD should be considered in children presenting with stroke-like episodes, in addition to classical radiological description of hypoxic ischemic encephalopathy.

Declarations

Conflict of Interest None.

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