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CORRESPONDENCE

An Inherited Cause of Stroke Mimic in a Toddler

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To the Editor: Stroke mimic is one of the common causes of neurological deficits in young children. A 2-y-old-boy born to nonconsanguineous parents presented with recurrent weakness of left upper and lower limbs alternating with right upper and lower limbs, each event lasting for 48–72 h. The child was delivered at 36 wk of gestation with a birth weight of 2130 g. He had daily episodes of abnormal twisting posture of limbs and trunk with preserved awareness from first month of age. Mild developmental delay was observed. Left eye convergent squint and right hemiplegia were observed. Possibilities of alternating hemiplegia of child (AHC), mitochondrial encephalomyopathy with lactic acidosis and stroke (MELAS) and Moyamoya syndrome were considered. Brain magnetic resonance imaging with magnetic resonance angiography and electroencephalography were normal. Next generation sequencing revealed a likely pathogenic heterozygous missense variation in the exon 17 of *ATP1A3* gene (c.2440G>A; p.Asp814Asn; ENST00000545399.6) confirming the diagnosis of alternating hemiplegia of childhood-2.

AHC is a rare inherited cause of stroke mimic in infants and children. The key differentiating features of stroke mimics from stroke are gradual onset symptoms, presence of altered sensorium, nonvascular territory involvement, absence of vascular risk factors, absence of ischemic or hemorrhagic lesions on neuroimaging, and rapid recovery [1]. *ATP1A3*-related neurological disorders include rapid onset dystonia-parkinsonism (RDP), AHC and cerebellar ataxia, areflexia, pes cavus, optic atrophy, and sensorineural deafness (CAPOS) [2]. AHC typically has onset before 18 mo with repeated episodes of neurological symptoms of variable duration. Neurological symptoms include alternating hemiparesis, quadriparesis, dystonic spells, oculomotor abnormalities, developmental delay, ataxia and autonomic symptoms [3]. Our patient had developmental delay, paroxysms of dystonia, and alternating hemiparesis. The pathophysiological mechanisms described in *ATP1A3*-related neurological disorders include dysfunctional neuronal sodium–potassium ATPase pump and imbalance of glutaminergic-GABAergic activity [4]. Treatment includes avoidance of sleep deprivation and stress, flunarizine, topiramate, benzodiazepines, steroid, amantadine, memantine, aripiprazole, adenosine triphosphate, coenzyme Q, acetazolamide, dextromethorphan, ketogenic diet and vagus nerve stimulation [4].

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Declarations

Conflict of Interest None.

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