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SCIENTIFIC LETTER

Super-Refractory Status Epilepticus Progressing to Infantile Epileptic Spasms Syndrome Secondary to Very Long Chain Acyl-CoA Dehydrogenase Deficiency

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To the Editor: Very long chain acyl-CoA dehydrogenase deficiency (VLCAD) is an autosomal recessive disorder due to pathogenic variants in *ACADVL* gene. Clinical presentation includes cardiomyopathy, encephalopathy, myopathy, and hypoketotic hypoglycemia [1]. We are reporting on a rare phenotype of VLCAD presenting as super-refractory status epilepticus (SRSE) and later developing to infantile epileptic spasms syndrome (IESS).

An 11-mo-old girl, with normal birth history and development till 9 mo of age presented with fever followed by status epilepticus requiring more than five antiseizure medicines and midazolam infusion and mechanical ventilator support. Following this acute event, the baby had loss of all attained milestones and new onset spasms in clusters. On examination the weight was 6.1 kg (<3 standard deviation), head circumference 42 cm, bilateral esotropia, and hypotonia with brisk reflexes was found. On investigation, hypoglycemia (glucose 20 mg/dL) without ketosis and metabolic acidosis (pH 7.33, pCO_2 22.5 mmHg, base excess of 12.8 mEq/L and bicarbonate 11.7 mEq/L) was found. Ammonia, lactate, liver function tests, echocardiography, and creatine phosphokinase were normal. Tandem mass spectrometry showed elevated blood propionyl carnitine (C3) and heptadecanoyl carnitine (C17). CT brain during acute event showed diffuse cerebral edema. MRI brain showed diffusion restriction in periventricular region with diffuse cerebral atrophy. Electroencephalography was suggestive of very low voltage activity with multifocal interictal epileptiform discharges initially, later modified hypsarrhythmia. Whole exome sequencing identified a known pathogenic homozygous missense variant c.406C>T, p.(Leu136Phe) in exon 6 of *ACADVL* gene. The child improved partially after starting steroids, vigabatrin, riboflavin and low fat, high medium chain triglyceride with high protein diet.

Inborn errors of metabolism and mitochondrial disorders were considered and ruled out after metabolic workup. The lack of acetyl CoA and ketone bodies is the mechanism behind seizures in VLCAD [2, 3]. Metabolic causes should be considered in infancy with SRSE and IESS in addition to common causes as outcome depends on appropriate specific management.

Declarations

Conflict of Interest None.

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