Solving a Puzzle: An Infant With Developmental Delay, Epileptic Spasms, and Petechiae

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Introduction:

Infantile spasm (IS) is an early onset epileptic encephalopathy with psychomotor impairment and characteristic hypsarrhytmia pattern on electroencephalography (EEG). More than 25 inborn errors of metabolism have been considered etiologic or predisposing factors for IS. Herein we report a rare cause of IS in a patient with underlying metabolic etiology.

Case Report:

An 8-month-old boy presented with poor head control, epileptic spasms, and petechiae over extremities. He was the third child of consanguineous Turkish parents and was born at 39 weeks gestation by cesarean section following an uneventful pregnancy. Physical examination revealed axial hypotonia, orthostatic acrocyanosis, and petechiae located predominantly over the lower extremities. The initial EEG demonstrated a disorganized, high-amplitude background and multifocal, interictal epileptiform discharges consistent with modified hypsarrhythmia (Figure 1). He was treated with levetiracetam and topiramate. Epileptic spasms resolved with intramuscular ACTH therapy. Brain magnetic resonance imaging (MRI) showed mild cerebral atrophy and increased T2 signal in the basal ganglia bilaterally (Figure 1). Metabolik work-up revealed elevated serum lactate levels, and increased levels of ethylmalonic acid 176,42 mmol/mol/creatinine (NV: 1,7-14,6) on urine organic acid analysis . Metabolik work-up revealed elevated serum lactate levels. The diagnosis of ethylmalonic encephalopathy (EE) was confirmed by molecular genetic analysis (c.554T> G; p.Leu185Arg homozygous variation in *ETHE1*). He was initiated on N-acetyl cysteine, metronidazole, and riboflavin.

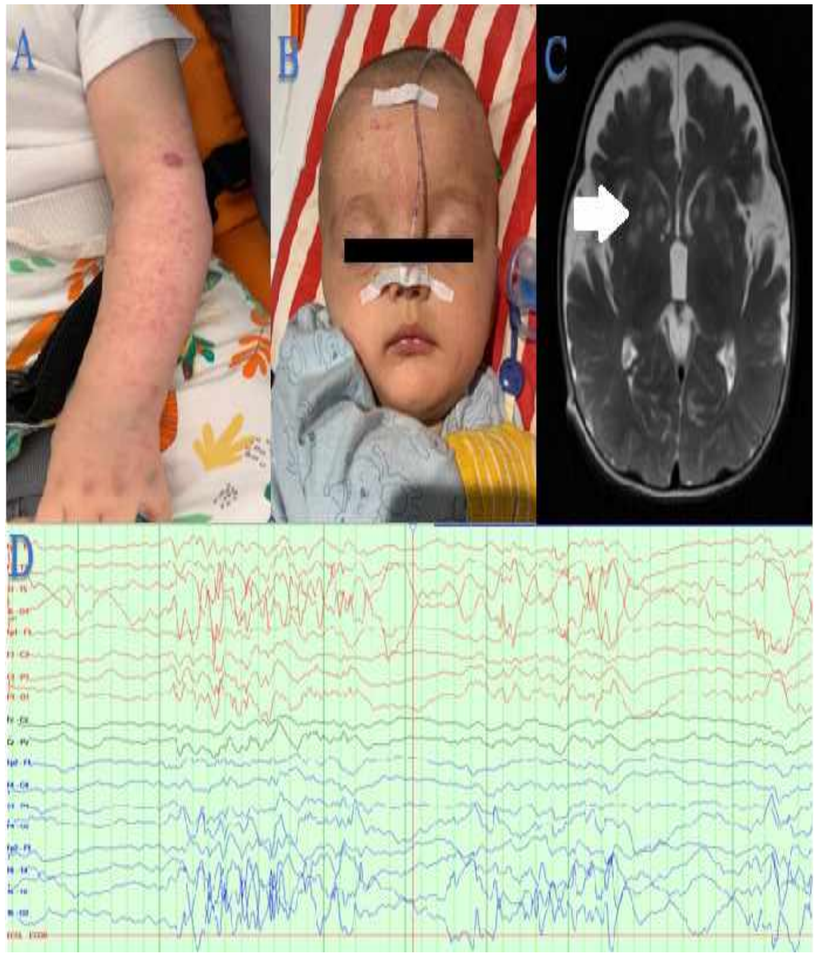

Figure 1: A-B: Patient photography revealing petechiae on his face and upper limbs. C: Axial T2-weighted imaging demonstrates signal hyperintensity at the level of the basal ganglia (white arrow). D: İnterictal EEG with modified hypsarrhythmia pattern.

Discussion:

Ethymalonic encephalopathy (EE) is an early-onset autosomal recessive severe metabolic disorder affecting the development of the brain, gastrointestinal tract, and peripheral blood vessels. Clinical features include neurodevelopmental delay, psychomotor regression, hypotonia, movement disorders, recurrent petechiae, orthostatic acrocyanosis, and chronic diarrhea. Treatment is primarily supportive including antiepileptic medications, physical therapy, maintenance of hydration or caloric intake, and multivitamin therapies for poor energy metabolism. Early diagnosis of metabolic disorders may lead to early initiation of appropriate treatment.

References:

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