Refractory Status Epilepticus as the First Presentation in an Infant with Alpers Disease

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ABSTRACT

Objective: Alpers Huttenlocher disease although is a rare mitochondrial disease, but is still present in different nervous systems and other system's features. Seizures and neurodegeneration are one of the early pictures encountered.

Case: A case of 11-month-old girl with acute onset of epileptic seizures has been reported along with her brother who died two years before this event following the same disease and symptoms. Her lab work values were normal for which she was planned for MRI. However, because of her general condition, only CT scan of brain was done which showed normal study.

Results: The electroencephalographic findings show a pattern of continuous, anterior, high-voltage, l-to-3-Hz spike-and-wave like activity that persists despite intermittent focal seizures. Death is not unusual in the setting of persistent seizures, and valproic acid should be avoided unless polymerase gamma γ level in genetic testing produces normal level. Whole exome sequencing identified the homozygous missense mutation c.3286C>T in *POLG* gene which corresponds to the p.R1096C amino acid change.

Conclusion: Alpers Huttenlocher disease should be considered in different diagnosis of refractory status epilepticus in infants even in absence of liver disease signs.

Keywords: Alpers Huttenlocher, Epilepsy, Infant, Refractory Status Epilepticus, Seizures

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