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Novel Mutation 0f ATP6V1A Gene Associated with Infantile Spasms and Microcephaly

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Abstract

We describe a seven-month-old boy who presented with flexor spasms of two weeks duration. He was first born child to unrelated parents. Term delivery with uneventful pregnancy and normal development milestones till seven-months of age. On examination he had small head with closed anterior fontanels. EEG was suggestive of hypsarrythmia with multifocal inter ictal discharges.

Etiologic investigations: Magnetic resonance imaging (MRI), chemical and physical examination of cerebrospinal fluid, pyruvic acid and lactic acid on plasma and cerebrospinal fluid, plasma and urinary aminoacids, urinary organic acids, CSF glycine and aminoacids , biotinidase levels were normal. Exome sequencing of a multigene epilepsy panel for EIEE detected ATP6VA1 mutation.

Child was treated with ACTH with complete cessation of spasms however he had recurrence after five months of stopping steroids. Currently he is on multiple anti epileptic drugs with partial response.

Four cases with ATP6V1A mutations have been reported till date, all of them had preexisting developmental delay and fever associated seizures, followed by epileptic encephalopathy. This child was developmentally normal and there were no episodes of fever triggered seizures. He presented with infantile spasms and neuroregression followed by refractory seizures.

This is the first case report of ATP6V1A mutation presenting as infantile spasms and refractory Epilepsy in a developmentally normal child.

The present case adds to the few cases of epileptic encephalopathies so far reported that were caused by ATP6V1A gene mutations and expands the clinical spectrum of ATP6V1A mutation associated epilepsy.

Biography:

Razia Adam Kadwa attained her MD-Pediatrics, DM Pediatric Neurology. She is Currently working as Consultant Pediatric Neurologist at Ankura Hospital for Women and Children, Hyderabad , India.

Speaker Publications:

1. Kadwa R. A, Nair A. K. "Isolated left ventricular non-compaction with moyamoya syndrome and renal agenesis-a rare association and a rare cause of recurrent stroke in a young boy"; Child's Nervous System : Chns : Official Journal of the International Society for Pediatric Neurosurgery. 2020 /36(8):1581-1582.

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