Epileptic Encephalopathy, Early Infantile, 38

Alternative Names
EIEE38

Record Category
Disease phenotype

WHO-ICD
Diseases of the nervous system> Episodic and paroxysmal disorders.

Incidence per 100,000 Live Births
Unknown

OMIM Number
617020

Mode of Inheritance
Autosomal recessive

Gene Map Locus
1q42.2

Description
Epileptic encephalopathy is a heterogeneous group of epilepsy syndromes associated with severe cognitive and behavioral disturbances characterized by spontaneous, recurrent seizures and neurodevelopmental impairment. Epileptic encephalopathy include eight syndromes: Ohtahara syndrome, Dravet syndrome, West syndrome, myoclonic status in nonprogressive encephalopathies, Landau-Kleffner syndrome, Lennox-Gastaut syndrome, and epilepsy with continuous spike waves during slow-wave sleep. These syndromes vary in their age of onset, seizure types, developmental outcome, etiologies, EEG patterns, neuropsychological deficits, and prognosis. The disease prognosis is very poor; most affected children either die or are severely neurologically impaired.

Molecular Genetics
Mutations in the ARV1 gene, located on 1q42.2 chromosome, have been associated with early infantile epileptic encephalopathy 38. This gene encodes a transmembrane protein that mediates sterol transport from the endoplasmic reticulum (ER) to the plasma membrane.

Epidemiology in the Arab World

Lebanon
Palmer et al., (2016) reported a girl who presented at six weeks of life with persistently irritable with roving eye movements and demonstrated visual inattention, central hypotonia, extensor posturing, peripheral hypertonia, and dystonia. She was the third child of consanguineous Lebanese parents. At four months of age she developed intractable seizures, and didn't obtain developmental milestones. She died at 12 months of age due to intractable seizures and recurrent respiratory infections. A novel homozygous (c.294+1G>A) mutation in the ARV1 gene was found in the effected girl. This gene resulted in a splice site mutation that leads to an in-frame deletion of 40 amino acids (p.Lys59_Asn98del).

Saudi Arabia
Alazami et al.,(2015) identified a novel homozygous missense mutation (c.565G>A) in the ARV1 gene in a male born to consanguineous Saudi parents. This gene resulted in a Gly189Arg amino acid substitution. He presented with severe intellectual disability, poor head control, early onset epileptic encephalopathy, and ataxia. He had a brother who died at the age of four years due to similar neurodevelopmental disorder. Also his two second cousins (a female and a male) had the same (c.565G>A) mutation with similar symptoms including: epileptic encephalopathy, ataxia, intellectual disability, and visual impairment.

References
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Related CTGA Records

ARV1, S. Cerevisiae, Homolog of

External Links

http://emedicine.medscape.com/article/1179970-overview

Contributors

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