ORIGINAL RESEARCH PAPER

Paediatrics

DEVLOPMENTAL AND EPILEPTIC ENCEPHALOPATHY

KEY WORDS:

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BACKGROUND:
Developmental and epileptic encephalopathy-13 is caused by heterozygous mutation in the SCN8A gene (voltage-gated sodium channel, type VIII, alpha subunit gene) on chromosome 12q13. Developmental and epileptic encephalopathy-13 is a neurologic disorder characterized by the onset of intractable seizures in the first year of life. Some patients may present with seizures in the first days, whereas others present later (between 2 and 7 months of age) after normal or only mild developmental delay. Affected individuals have profoundly impaired development or developmental regression after the onset of seizures, and show severe intellectual disability, poor or absent language, hypotonia, and are usually unable to walk. Some patients may have progressive microcephaly and poor eye contact. Most patients have intellectual disability, hypotonia, impaired balance, developmental regression, speech and language regression.

Case Report:
A 3 months old, male child, presented with complaint of 1 episode of convulsion, GTCS type lasting for 15-20 minutes. No c/o fever.

Patient had birth h/o perinatal asphyxia and history of NICU admission for same. No episode of convusion during that period. On day 7 of life, patient had 1st episode of convulsion and patient was started on AED. MRI Brain and EEG was done and both were normal. 2nd episode of covulsion at 2.5 month of age and patient admitted again and during course of treatment patient had multiple episodes of convulsion during hospitalisation. AED were added and dose adjusted accordingly. Till 3 months of age, patient was developmentally normal, Anterior fontanelle was open and at level. Repeat MRI was done and it was normal and repeat EEG suggestive of generalized intermittent spike and wave discharges. Patient was having refractory convulsions despite being on multiple AED. Patient was started on tab pyridoxine and metabolic workup was sent. TMs was normal. Tablet topiramate and clobazam were added. Considering possibilities of genetic epilepsy, whole exome sequencing was sent which came out positive as devlopmental epileptic encephalopathy, heterozygous, autosomal dominant in nature. During the course of hospital stay, patient started to have developmental regression, anterior fontanelle fused and activity reduced. Patient adviced to continue aed and for regular follow up.

DISCUSSION:
Developmental and Epileptic Encephalopathy (DEE) refers to a group of severe epilepsies that are characterized both by seizures, which are often drug-resistant, as well as encephalopathy, which is a term used to describe significant developmental delay or even loss of developmental skills. In the DEEs, there are two factors that contribute to the developmental delay.

Most DEEs begin early in life, often starting in infancy. Children often have very frequent and severe seizures which can be of multiple types. Often epileptic spasms, tonic or atonic seizures and myoclonic seizures can be seen. In most cases, seizures are life long, although less commonly can abate with time with certain syndromes or specific causes.

The electroencephalographs (EEGs) in children with DEEs are typically very abnormal, showing diffuse slowing of the background, and frequent seizure discharges. However, in some cases, the EEGs done early on (before or very shortly after the seizure onset) may not show abnormalities.

CONCLUSION:
In cases in which the encephalopathy is caused by both developmental impairment and epileptic activity, a developmental and epileptic encephalopathy should be considered. Despite the inevitable developmental sequelae for many of the DEEs, early recognition and intervention permits optimal and often improved outcomes.

Interventions for people with DEE should be based on a balance between control of epileptic activity and avoidance of unacceptable adverse effects.