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Case Presentation

A rare case of Genetic Epilepsy with Febrile Seizure plus (GEFS+) presenting as Landau-Kleffner Syndrome

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ABSTRACT

Background: Developmental and Epileptic Encephalopathy (DEE) is a term used by the International League Against Epilepsy (ILAE) to describe a group of severe, early-onset epilepsies. In these conditions, a person's development is negatively impacted by two distinct factors: the underlying cause of the epilepsy itself and the frequent, severe seizure activity and abnormal brain waves (EEG).
Case Presentation: A 5-year-old male child presented with complaints of altered sensorium. He had h/o multiple episodes of abnormal body movements lasting for about 2-3 minutes and associated with LOC, Tongue bite and incontinence. These episodes started at the age of 11 months of age. At age of 11 months, child developed febrile seizures with semiology – unknown onset GTCS, occurring within 24 hr of fever, lasting up to 30 min. For the next 6 months, child continued to have febrile seizures. Since last 8 months, seizure frequency increased to about 2-3/month with each Seizure lasting 20-30 min & prolonged post ictal phase. Parents noticed that child was less active & had reduced speech output with poor eye contact with paucity of emotional display. EEG showed infrequent to frequent intermittent generalised front central dominant spike and slow waves with preserved sleep architecture. MRI brain showed mild diffuse cerebral atrophy. On genetic study SCN1A gene mutation was there. Patient showed some improvement with immunosuppressive therapy which is IVIg 2mg/kg once and Pulse Inj Methylprednisolone for 6 months.
Conclusion: This case highlights an unusual presentation of LKS Syndrome with multiple episodes of seizure starting as fever induced with mutism and some behavioural symptoms. SCN1A gene mutation was there but secondary mutism favors more towards LKS Syndrome rather than Dravet syndrome.

Keywords*: Developmental and Epileptic Encephalopathy (DEE), LKS Syndrome, Dravet Syndrome, SCN1A mutation.*

INTRODUCTION

Developmental and Epileptic Encephalopathy (DEE) is a term used by the International League Against Epilepsy (ILAE) to describe a group of severe, early-onset epilepsies. In these conditions, a person's development is negatively impacted by two distinct factors: the underlying cause of the epilepsy itself and the frequent, severe seizure activity and abnormal brain waves (EEG). [1]

Key Components of DEEs
According to the Child Neurology Foundation, DEEs involve a "complex interplay" of three factors:

Developmental Encephalopathy: Developmental delay or intellectual disability caused directly by an underlying genetic mutation or brain injury, even if seizures are not occurring.

Epileptic Encephalopathy: Frequent seizures and abnormal EEG activity that further worsen or cause a regression in developmental skills.

Severe Seizures: These are often drug-resistant and can appear as various types, such as infantile spasms, tonic seizures, or myoclonic jerks.[1]

Common DEE Syndromes
Many rare epilepsy syndromes fall under the DEE umbrella, classified based on the age they start and their specific seizure and EEG patterns:

1. **Infantile Epileptic Spasms Syndrome (West Syndrome):** Often begins before age 1 with "jackknife" spasms in clusters and a characteristic "hypsarrhythmia" EEG pattern.
2. **Dravet Syndrome:** Typically starts in the first year of life with prolonged, fever-triggered seizures, often linked to the SCN1A gene.
3. **Lennox-Gastaut Syndrome (LGS):** Usually starts between ages 1 and 8, characterized by multiple seizure types (including tonic "stiffening" seizures) and slow spike-wave EEG patterns.
4. **Early Infantile DEE (EIDEE):** A newer group that includes what were formerly called Ohtahara syndrome and Early Myoclonic Encephalopathy.
5. **Landau-Kleffner Syndrome (LKS)**: A childhood syndrome where a child suddenly or gradually loses language skills (aphasia) in association with abnormal brain waves during sleep.[2]

Causes and Management
Causes: The majority are genetic (over 900 linked genes like SCN1A, KCNQ2, and STXBP1), though they can also be caused by structural brain changes, metabolic disorders, or unknown factors.[3]

Multimorbidity: Beyond seizures, individuals often face complex challenges including movement disorders, autism, sleep apnoea, gastrointestinal issues, and an increased risk of Sudden Unexpected Death in Epilepsy (SUDEP).

Treatment: Management is holistic and includes anti-seizure medications, specialized diets (like the ketogenic diet), and sometimes vagus nerve stimulation (VNS) or brain surgery. [4]

While some patients with Dravet might develop features that overlap with other epileptic encephalopathies (like Lennox-Gastaut), LKS specifically represents a focal epilepsy syndrome associated with acquired aphasia.[5]

CASE PRESENTATION
A 5-year-old male child presented with complaints of altered sensorium. He had h/o multiple episodes of abnormal body movements lasting for about 2-3 minutes and associated with LOC, Tongue bite and incontinence. These episodes started at the age of 11 months of age. At age of 11 months, child developed febrile seizures with semiology – unknown onset GTCS, occurring within 24 hr of fever, lasting up to 30 min. For the next 6 months, child continued to have febrile seizures. Since last 8 months, seizure frequency increased to about 2-3/month with each Seizure lasting 20-30 min & prolonged post ictal phase. Parents noticed that child was less active & had reduced speech output.

The child also had poor eye to eye contact, just able to comprehend few simple commands. There was paucity of emotional display. He was also having difficulty in speech like mouthing of sounds able to produce only monosyllables.
There was history of repetitive movements in the form of inappropriate clicking sounds, grunting. Child was able to comprehend few simple commands from parents. No myoclonic jerks, head drops, absence episodes. Previously child was having normal developmental milestones and birth was uneventful.

EEG showed infrequent to frequent intermittent generalised front central dominant spike and slow waves with preserved sleep architecture. MRI brain showed diffuse cerebral atrophy.

Due to behavioural symptoms, we suspected LKS Syndrome and planned for immunosuppressive therapy which is IVIg 2mg/kg once and Pulse Inj Methylprednisolone for 6 months. Patient showed some improvement with the treatment.

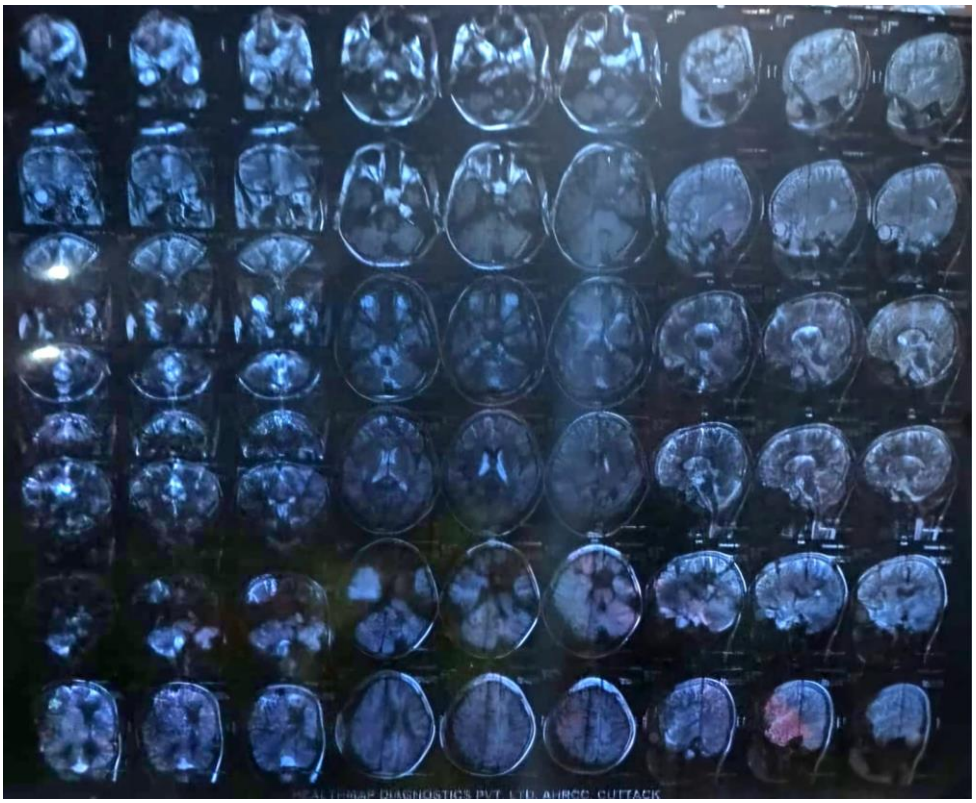
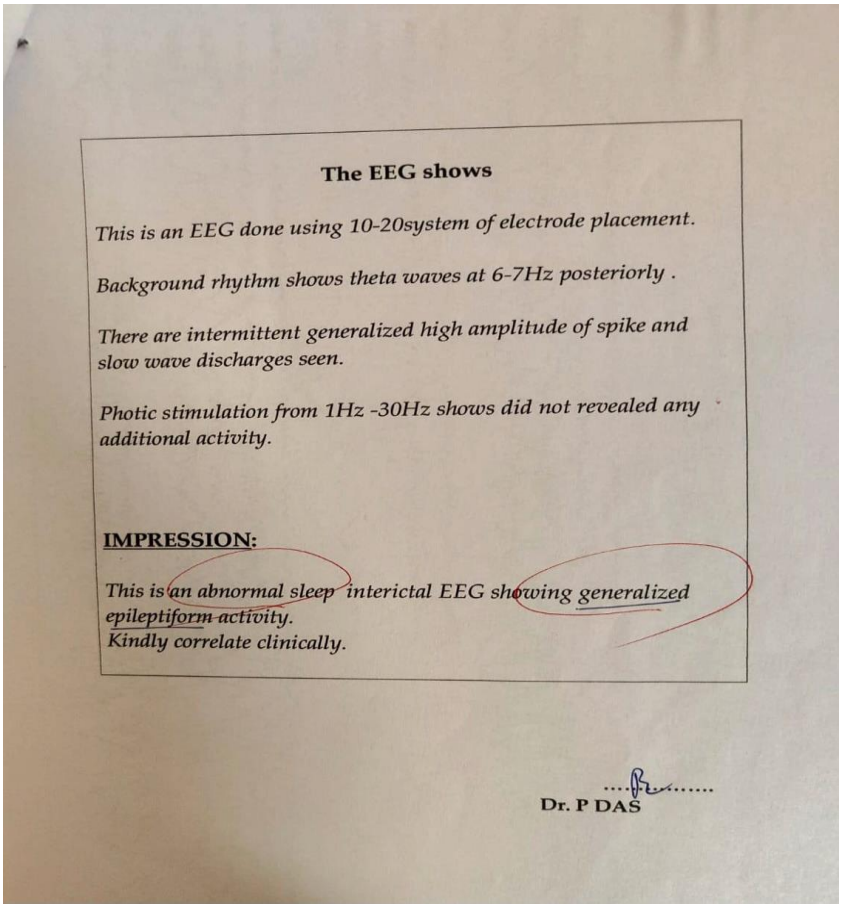


FIGURE 1: T2 Weighed MRI Brain showing Mild Diffuse Cerebral Atrophy



The EEG shows

This is an EEG done using 10-20system of electrode placement.

Background rhythm shows theta waves at 6-7Hz posteriorly .

There are intermittent generalized high amplitude of spike and slow wave discharges seen.

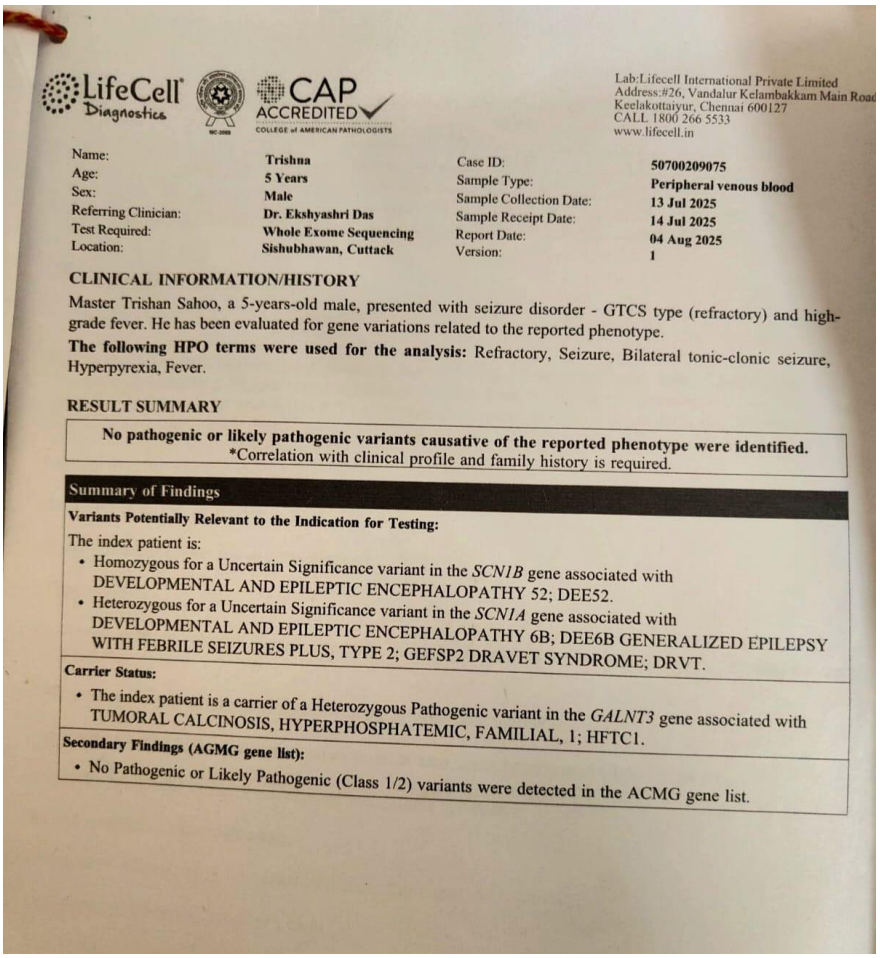
Photic stimulation from 1Hz -30Hz shows did not revealed any additional activity.

IMPRESSION:

This is an abnormal sleep interictal EEG showing generalized epileptiform activity.
Kindly correlate clinically.

Dr. P DAS

Figure 2: EEG Showing frequent intermittent generalised frontocentral dominant spike and slow waves with preserved sleep architecture.



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Name:	Trishna	Case ID:	50700209075
Age:	5 Years	Sample Type:	Peripheral venous blood
Sex:	Male	Sample Collection Date:	13 Jul 2025
Referring Clinician:	Dr. Ekshyashri Das	Sample Receipt Date:	14 Jul 2025
Test Required:	Whole Exome Sequencing	Report Date:	04 Aug 2025
Location:	Sishubhawan, Cuttack	Version:	1

CLINICAL INFORMATION/HISTORY

Master Trishan Sahoo, a 5-years-old male, presented with seizure disorder - GTCS type (refractory) and high-grade fever. He has been evaluated for gene variations related to the reported phenotype.

The following HPO terms were used for the analysis: Refractory, Seizure, Bilateral tonic-clonic seizure, Hyperpyrexia, Fever.

RESULT SUMMARY

No pathogenic or likely pathogenic variants causative of the reported phenotype were identified.
*Correlation with clinical profile and family history is required.

Summary of Findings

Variants Potentially Relevant to the Indication for Testing:

The index patient is:

- Homozygous for a Uncertain Significance variant in the *SCN1B* gene associated with DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY 52; DEE52.
- Heterozygous for a Uncertain Significance variant in the *SCN1A* gene associated with DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY 6B; DEE6B GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, TYPE 2; GEFSP2 DRAVET SYNDROME; DRVT.

Carrier Status:

- The index patient is a carrier of a Heterozygous Pathogenic variant in the *GALNT3* gene associated with TUMORAL CALCINOSIS, HYPERPHOSPHATEMIC, FAMILIAL, 1; HFTC1.

Secondary Findings (AGMG gene list):

- No Pathogenic or Likely Pathogenic (Class 1/2) variants were detected in the ACMG gene list.

Figure 3: Patient is having mutation at SCN1A,SCN1B and GALNT3 Gene locus

DISCUSSION

Landau-Kleffner Syndrome (LKS) is a childhood syndrome where a child suddenly or gradually loses language skills (aphasia) in association with abnormal brain waves during sleep whereas Dravet Syndrome Typically starts in the first year of life with prolonged, fever-triggered seizures, often linked to the SCN1A gene.

This case highlights an unusual presentation of LKS Syndrome with multiple episodes of seizure starting as fever induced with mutism and some behavioural symptoms. SCN1A gene mutation was there but secondary mutism favours more towards LKS Syndrome rather than Dravet syndrome.

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