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A REVIEW OF DISTINCTIVE EEG PATTERNS IN PEDIATRIC GENETIC EPILEPSIES

Neuroscience

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ABSTRACT

Background: Epilepsy is a neurological condition that is common and persistent, and it is extremely prevalent in children. According to the International League Against Epilepsy (ILAE), around 30% of all epilepsies are of genetic origin, and over 0.4% of the general population is affected. **Methods:** A review was done using PubMed, Google Scholar and other related databases. Keywords like genetic epilepsy, EEG patterns in genetic epilepsy, ictal and interictal patterns in genetic epilepsy are used for the review. Inclusion criteria include the different EEG patterns of genetic epilepsy; 31 studies satisfied the inclusion criteria for the review. **Results:** This review foreshows the distinctive ictal and interictal EEG patterns linked to developmental and epileptic encephalopathies, as well as the main hereditary epilepsy disorders. Different electroclinical signatures were found in a variety of gene-related epilepsies, demonstrating recurring relationships between particular genetic variations and EEG characteristics. Differentiating seizure types, defining epileptic syndromes, and promoting early and precise diagnosis were all made possible by video-EEG monitoring. **Conclusion:** Video-EEG monitoring is a significant tool in evaluating hereditary epilepsy because it allows for better electroclinical characterisation and improved disease classification. The combination of genetic testing and Video-EEG results improves diagnosis accuracy, enables individualised treatment options, and adds to a deeper understanding of genotype-phenotype correlations.

KEYWORDS

Pediatric Genetic epilepsy, Video EEG, Classical semiology, Ictal EEG pattern, Interictal EEG pattern

INTRODUCTION

Epilepsy is one of the most prevalent and chronic neurological disorders, impacting most frequently in children, with an incidence of 144/100,000 persons during the first year of life, and 58/100,000 aged between 1-10 years are affected by epilepsy (1). The International League Against Epilepsy states that genetics is one of the main causes of epilepsy, and around 30% of all epilepsies are genetic , and over 0.4% of the general population is affected (2,3). The majority of genetic alterations associated with epilepsy have been detected in genes encoding ion channels (4).

Depending on the genes affected, seizure types and seizure complexity can vary significantly for each person who has genetic epilepsy . Video-EEG monitoring is an established technique to correlate clinical behaviour with EEG phenomena (5).

This review aims to summarise current knowledge on the genetic basis of epilepsy, with particular emphasis on ion channel–related epilepsies, and to highlight the role of Video-EEG in the characterisation and clinical management of genetic epilepsy syndromes.

Methods

A detailed literature search was commenced using PubMed, Google Scholar, and Scopus to find publications that reported electroencephalographic (EEG) characteristics in genetically proven epileptic syndromes. Searches were conducted using the keywords genetic epilepsy, video-EEG, ictal EEG, interictal EEG, and pediatric genetic epilepsy. Studies were considered if they showed ictal and interictal EEG abnormalities in patients with genetic epilepsy. The articles without EEG data and abstracts that did not include the complete data were excluded. Due to the variability of the study designs, data on genetic aetiology, clinical semiology, and ictal and interictal EEG patterns were retrieved and combined using a narrative qualitative technique. The final analysis includes 22 studies that met the inclusion criteria.

RESULTS:

An initial total of 272 articles were identified through the literature search. These data were then screened by reviewing their titles and abstracts to determine their relevance. After this initial screening, 220 records were excluded due to not meeting the inclusion criteria. As a result, 22 complete articles were assessed to determine their eligibility, describing EEG findings in genetically defined epilepsy syndromes that predominantly involve pediatric populations and developmental and epileptic encephalopathies is illustrated in the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) flow diagram provided [Figure 1]. Across multiple genetic etiologies, characteristic and recurring electroclinical patterns were identified.

Genetic Epilepsies And Their EEG Findings

KCNQ2-related epilepsy:

Lee IC et al, Kwak N et al Neonatal-onset developmental and epileptic encephalopathy in newborns typically showed better background in seizures. The interictal findings may show burst suppression and multiple focal spikes (6). The ictal EEG shows sudden rise in amplitude followed by a suppressed background.

LGI1 Related epilepsy:

Ottman R et al ,Michelucci R et al, Bonaventura CD et al LGI1 mutations have been associated with autosomal dominant lateral temporal lobe epilepsy, which is characterised by focal seizures (7,8) . The interictal EEG showed a significant epileptic activity characterised by spikes in concise runs [Figure 2]. The ictal EEG activity indicated the involvement of the left posterior lateral temporal cortex (9).

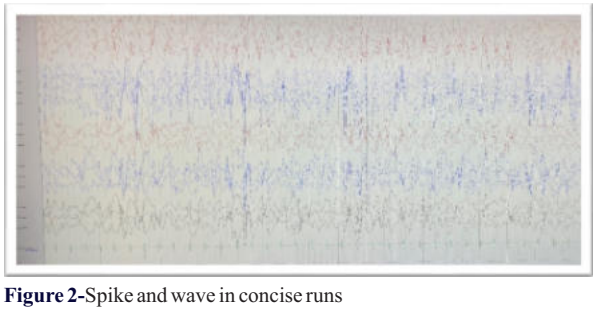

Figure 2-Spike and wave in concise runs

CHRNA4-related epilepsy:

Nielsen TØ et al Autosomal dominant sleep-related hypermotor epilepsy (ADSHE) seizures are generally brief (<2 min) with abrupt onset and offset. Attacks can encompass emotional outbursts with a frightened appearance and are typified by stereotyped movements. The video EEG monitoring revealed 3–4 Hz polyspikes. An evolving delta rhythm was seen in the ictal EEG (10).

CHD2-related epilepsy:

Clara-Hwang A et al, Chen J et al., early-onset disorders like developmental and epileptic encephalopathy and drug-resistant epilepsies, the interictal record showed Generalized spike and

polyspike[Figure 3] . The ictal record showed a sequence of myoclonic seizures and epileptic spasms [Figure 4] (11,12) .

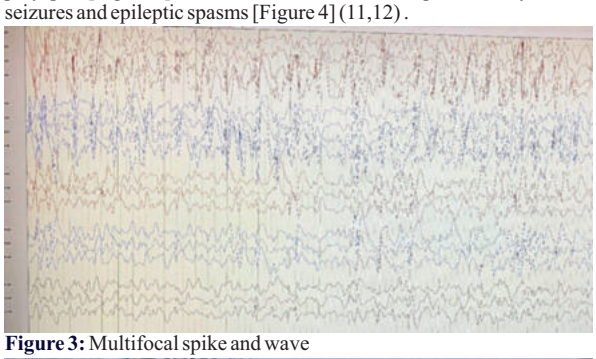

Figure 3: Multifocal spike and wave

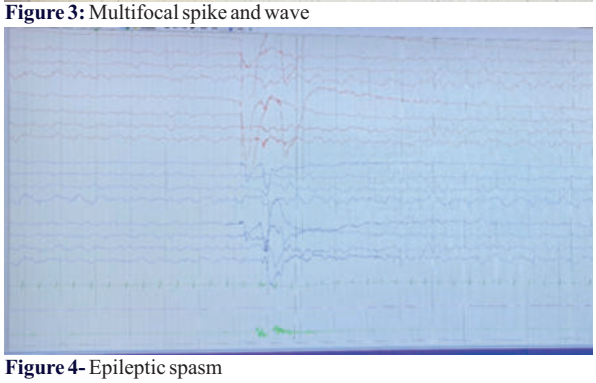

Figure 4- Epileptic spasm

SCN1A Related Epilepsy:

Kim SH et al Dravet syndrome. Interictal showed diffuse background slowing and a slower posterior dominant rhythm with multifocal spike discharge. A generalised discharge was the most commonly seen ictal EEG pattern. (13) [Figure 5]

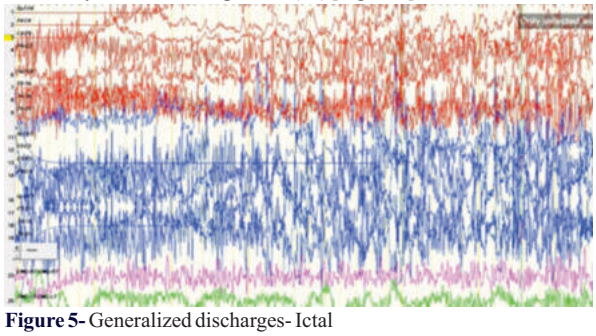

Figure 5- Generalized discharges- Ictal

Tang S et al, Oguni H et al Myoclonic atonic epilepsy, also called Doose Syndrome, generally begins in children with seizure onset (14). The interictal activity demonstrated intermittent sharp-slow complexes, and the ictal EEG revealed drop attacks, associated with epileptic spasms [Figure 6] (15) .

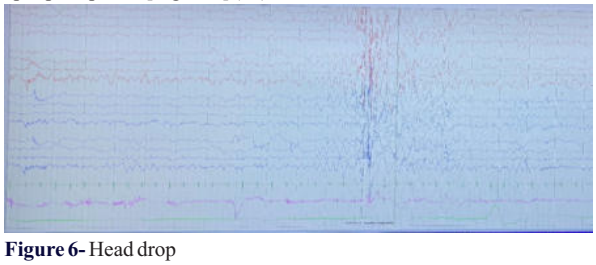

Figure 6- Head drop

Yoshitomi S et al, Coppola G et al Epilepsy of infancy with migrating focal seizures (EIMFS), the interictal pattern of Suppression burst was first obtained within one month of age, along with interhemispheric asynchrony (16). The ictal EEG pattern revealed rhythmic monomorphic activity in the range of alpha-theta frequency, although spike and slow waves can also be detected (17).

TSC1 AND TSC2 related epilepsy:

Savini MN et al Tuberous Sclerosis Complex, interictal revealed multifocal spikes, involving the left occipital region, the right frontotemporal region. Ictal EEG correlates generalised slow waves, a focal seizure that arises from the right frontal region (18).

CDKL5 gene-related epilepsy:

Melani F et al, Interictal EEG abnormality was characterised by sporadic sharp waves. Ictal EEG demonstrated an abrupt generalised decremental response during the initial spasms. High-amplitude slow waves with superimposed fast activity during the tonic contraction, followed by bilateral high-amplitude spikes and sharp waves during spasms (19).

SYNGAP1-related epilepsy:

Vlaskamp DR et al, Barco TL et al A developmental and epileptic encephalopathy (DEE) The interictal EEG showed multifocal low-voltage spikes with occipital predominance (20)**.** Ictal EEG demonstrated rhythmic delta activity in trains with a frequency of 3–3.5 Hz (21).

Grin-related epilepsy:

Ohba C et al, Interictal EEG showed nonspecific focal and diffuse epileptiform abnormalities. High-amplitude spike waves in the left or right central region spread to mid central region. Multifocal spike and waves were seen over bilateral frontal and central regions (22).

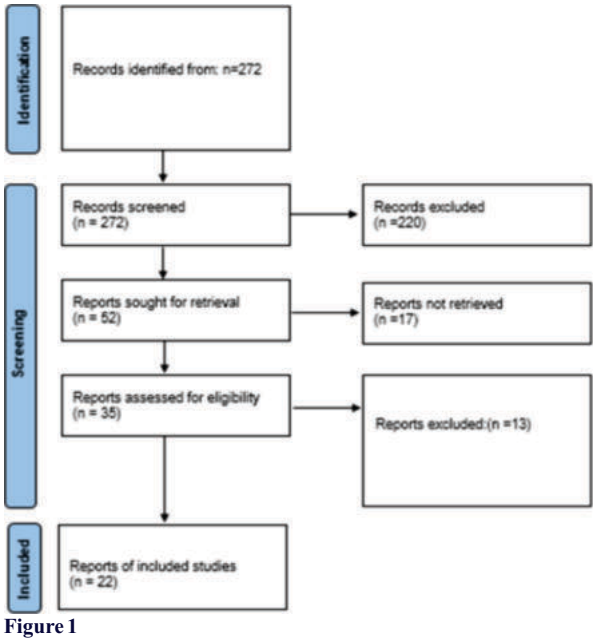

Figure 1

DISCUSSION:

This review synthesises current evidence on electroencephalographic characteristics associated with genetically defined epilepsy syndromes, with a particular focus on ion channel–related epilepsies and developmental and epileptic encephalopathies (DEEs). The integration of genetic testing with detailed Video-EEG analysis enhances syndrome recognition, facilitates early diagnosis, and supports precision-based therapeutic strategies.

Across multiple genetic etiologies, characteristic interictal and ictal EEG features were identified, reflecting underlying molecular and neurophysiological mechanisms. Ion channel gene mutations, such as in KCNQ2, SCN1A, and CHRNA4, predominantly affect neuronal excitability and synaptic transmission, leading to distinct electrographic signatures.

Similarly, SCN1A-related epilepsies, including Dravet syndrome and epilepsy of infancy with migrating focal seizures (EIMFS), demonstrated evolving EEG phenotypes over time. In Dravet syndrome, the progression from initially normal or mildly abnormal EEGs to diffuse background slowing and multifocal or generalized epileptiform discharges reflects the dynamic nature of this disorder.

In contrast, focal epilepsy syndromes associated with genes such as LGI1 and CHRNA4 show more localized EEG abnormalities consistent with their cortical network involvement. LGI1-related

autosomal dominant lateral temporal lobe epilepsy is characterised by posterior temporal interictal spikes and focal ictal onset, supporting its classification as a focal epilepsy syndrome with auditory features. Likewise, CHRNA4-related sleep-related hypermotor epilepsy often requires prolonged Video-EEG monitoring to capture ictal and interictal abnormalities, as routine EEGs may be normal.

Genes associated with broader neurodevelopmental dysfunction, such as CHD2, CDKL5, and SYNGAP1, were associated with generalized and multifocal EEG abnormalities, reflecting widespread cortical network involvement. The presence of such diffuse EEG abnormalities aligns with the severe developmental phenotypes observed in these conditions and supports the concept that EEG can serve as a surrogate marker of global cortical dysfunction in DEEs.

Importantly, this review highlights that no single EEG pattern is pathognomonic for a specific genetic epilepsy. Instead, the combination of clinical semiology, age of onset, EEG features, and genetic results is essential for accurate syndrome classification. Video-EEG monitoring plays a critical role in capturing ictal events, distinguishing seizure types, and identifying electroclinical syndromes, particularly in infants and young children where clinical manifestations may be subtle or atypical.

In conclusion, gene-specific EEG patterns provide important insights into the neurobiological mechanisms of genetic epilepsies and represent a critical component of modern epilepsy diagnostics. The continued integration of molecular genetics with detailed Video-EEG characterization will be essential for advancing personalized medicine approaches and improving outcomes for patients with genetic epilepsy syndromes.

CONCLUSION:

Video EEG monitoring plays a vital role in classifying genetic epilepsies, identifying seizure types, electroclinical features, including ictal and interictal EEG patterns, and also in treatment planning. Advances in neuroimaging, genetic testing, and computational EEG analysis continue to enhance our understanding of these patterns, enabling personalised therapeutic approaches. Future research should focus on integrating genetic data with electrophysiological findings to refine diagnostic precision and improve patient outcomes.

STUDY	YEAR	GENES	EPIELPSY/ SYNDROME TYPE	INTERICTAL EEG FINDING	ICTAL EEG FINDING
Lee IC et al; Kwak N et a	2021	KCNQ2	Neonatal-onset developmental and epileptic encephalopathy	Burst suppression; multiple focal spikes; paroxysmal centrotemporal	Sudden rise in amplitude followed by background suppression
Ottman R et al; Michelucci R et al; Bonaventura CD	2009	LGI1	Autosomal dominant lateral temporal lobe epilepsy	Isolated or brief runs of spikes in the left posterior temporal region	Left posterior lateral temporal cortex
Nielsen TØ et al	*2022*	*CHRNA4*	Autosomal dominant sleep-related hypermotor epilepsy	Long-term EEG: 3–4Hz polyspikes in the left frontotemporal region	Evolving delta rhythm beginning in the left frontotemporal region
Clara-Hwang A et al; Chen J et al	2024	CHD2	CHD2-related DEE and drug-resistant epilepsy	Generalised spike, polyspike, and slow waves.	Myoclonic seizures and epileptic spasms
Kim SH et al	2015	SCN1A	Dravet Syndrome	Diffuse background slowing; slow posterior dominant rhythm; multifocal spikes	Generalised discharges and focal patterns are more common in younger children
Tang S et al; Oguni H et al	2022	SCN1A	Myoclonic-atonic epilepsy (Doose syndrome)	Intermittent sharp–slow complexes; generalised sharp–slow discharges	Drop attacks associated with epileptic spasms
Yoshitomi S et al; Coppola G et al	2019	SCN1A	Epilepsy of infancy with migrating focal seizures (EIMFS)	Burst-suppression pattern; interhemispheric asynchrony	Rhythmic monomorphic alpha–theta activity; spike-and-slow waves
Savini MN et al	2020	TSC1 / TSC2	Tuberous sclerosis complex	Multifocal spikes and sharp waves centrotemporal predominance	Generalised slow waves; focal seizures , diffuse slowing with sporadic spikes
Melani F et al	2011	CDKL5	CDKL5-related DEE	Sharp waves over left frontocentral region	Generalized decremental response at spasm onset; high-amplitude anterior slow waves
Vlaskamp DR et al; Barco TL et al	2021	SYNGAP1	SYNGAP1-related DEE	Multifocal low-voltage spikes with occipital predominance	Rhythmic posterior delta (3–3.5 Hz) with superimposed low-voltage "notched" spikes.
Ohba C et al	2015	GRIN (GRIN1/2)	GRIN-related epilepsy	Nonspecific focal and diffuse epileptiform abnormalities; burst suppression; multifocal spikes	High-amplitude spike-waves in central regions

Availability Of Data And Materials: This is a review article all data are attained from published literature.

Ethical Approval: Not required

Declaration Of Patient Consent: Not applicable

Financial Support and Sponsorship: Nil

Conflicts Of Interest: Nil

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