DIAGNOSTIC UTILITY OF ELECTROENCEPHALOGRAM IN DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHIES

Dr Asha S, Dr Smilu Mohanlal, Dr Divya Pachat, Dr. Preetha Remesh, Dr. Tushar VP
Department of Pediatric, Genetics, Pediatrics and Adult neurosciences, Aster MIMS hospital, Calicut

18TH ICNC 2024 Footprints to the future
INTERNATIONAL CHILD NEUROLOGY CONGRESS
Cape Town, South Africa I May 6-10, 2024

IINTRODUCTION

- Developmental and epileptic encephalopathy (DEE) are conditions where cognitive functions are influenced by seizures, interictal epileptiform activity and the neurobiological process behind the epilepsy
- Many DEEs are related to gene variants
- Electroencephalography (EEG) plays a vital role in defining the epilepsy syndrome
- The EEG in DEE are normal in early stages but characteristic abnormalities can evolve that provides clue in tailoring the investigations towards diagnosis
- We describe our experience in the diagnosis of various DEE with the help of EEG

OBJECTIVES

To diagnose various developmental and epileptic encephalopathies with the help of electroencephalogram (EEG)

Neuroimaging(n=18/30)

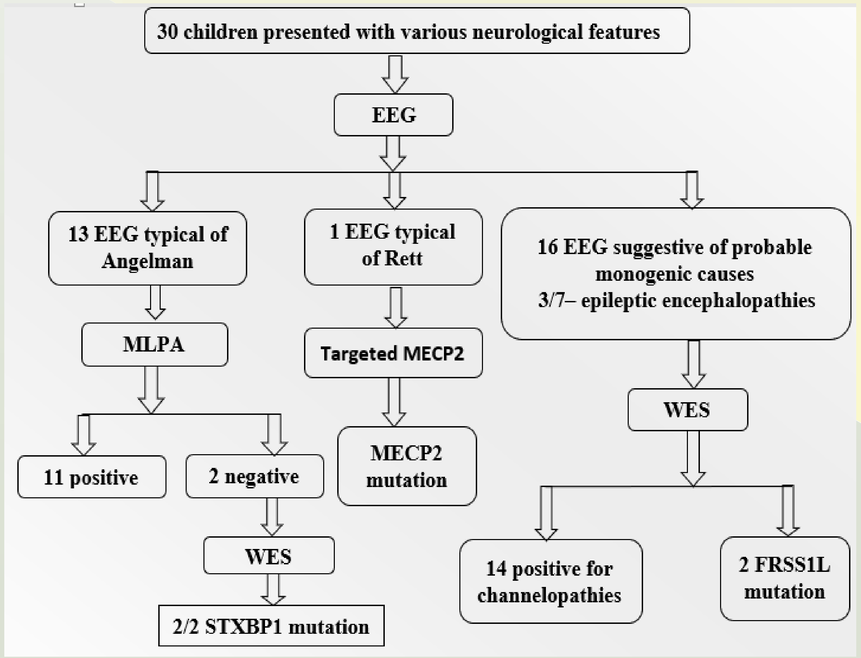
Specific	Periventricular leukomalacia	3
	Minimal volume loss in left hippocampus	1
Non Specific	Bilateral fronto-temporo-parietal atrophy Diffuse white matter volume loss Thinning of corpus callosum	1
	Wide cortical sulci hyperintense signals in periventricular white matter	1
	Right mesial temporal sclerosis	1
Normal		11

EEG findings(n=30)

High amplitude waves & notched appearance over frontal area	5	Multiplex ligation dependent probe amplification (MLPA) for Angelman syndrome
High amplitude wave in posterior head region	4	
High amplitude rhythmic slow wave activity	4	
Centro parietotemporal spike	1	Targeted mutation analysis for MECP2
Multi-focal discharges	14	Whole exome sequencing
Hypsarrhythmia pattern	2	
Normal	1	

MATERIALS AND METHOD

- Observational longitudinal study
- Aster MIMS Calicut in the department of Pediatrics & Pediatric neurology
- Study period: from January 2022 - June 2023
- Study population : 30 children who presented with developmental delay (cognitive delay > motor delay) with or without seizures
- Method:
  - Initially investigated with EEG
  - With the EEG pattern appropriate modality of genetic testing was done



RESULTS

- 30 children- age 1-15 years recorded.
- Male: female - 13:17.
- Predominant clinical features
  - 10 with seizures and developmental delay
  - 12 with predominant cognitive delay
  - 8 with autistic traits & development delay
- Neuroimaging done in in 21 of which 7 were abnormal
- EEG done and based on the EEG pattern appropriated genetic evaluation sent

Fig A- High amplitude waves in posterior head region

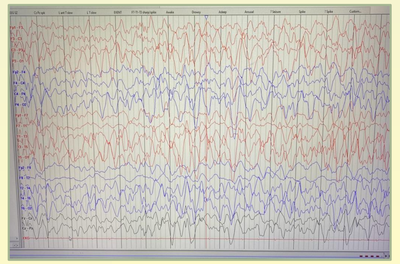
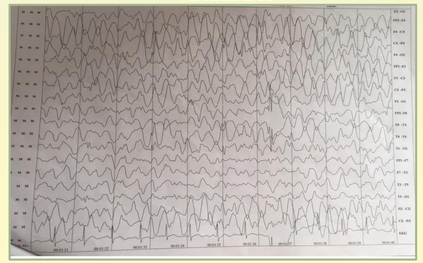
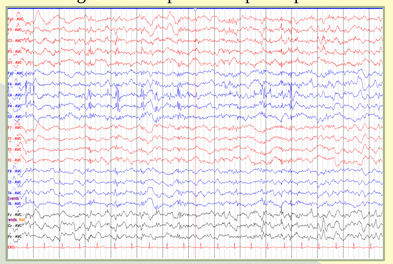


Fig B- High amplitude waves with notched appearance ov frontal area



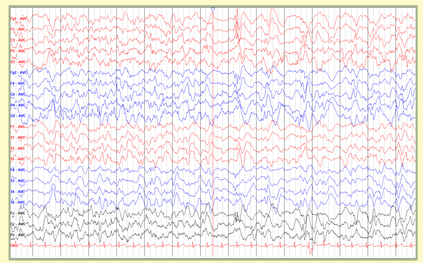
EEG pattern suggestive of Angelman syndrome

Fig C- Centro parietotemporal spike



EEG pattern suggestive of Rett syndrome

Fig D- Multifocal spike and slow wave discharges



EEG pattern suggestive of FRSS1L mutation

CONCLUSION

EEG patterns play a key role in tailoring the investigations for the genetic diagnosis of DEE.

Referrences
1.Laan, Laura & Vein, Alla. (2005). Angelman syndrome: Is there a characteristic EEG?. Brain & development. 27. 80-7. 10.1016/j.braindev.2003.09.013.
2. Portnova G, Neklyudova A, Voinova V, Sysoeva O. Clinical EEG of Rett Syndrome: Group Analysis Supplemented with longitudinal Case Report. J Pers Med. 2022 Nov 29;12(12):1973. doi: 10.3390/jpm12121973. PMID: 36556193; PMCID: PMC9782488.