Epileptic encephalopathy of genetic origin clinical-electro-radiological correlations in a Tunisian pediatric population

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INTRODUCTION

- Developmental and epileptic encephalopathy (DEE) (1) constitute an heterogeneous group of age-related severe epileptic syndromes, of predominantly **genetic** origin.
- The development of **next generation-sequencing (NGS)** (2) techniques has shortened the diagnostic pathway.

OBJECTIVES

- Our aim was to study **the clinical, electrophysiological and radiological** features of DEEs in a Tunisian pediatric population.

MATERIALS AND METHODS

- **Type of study :** A retrospective and descriptive study
- **Place:** Pediatric Neurology in National Institute Mongi Ben Hmida of Neurology, Tunisia.
- **Period:** Between 2005 and 2023
- **Target population:** Patients with DEE in whom genetic etiology was confirmed by the Whole Exome Sequencing.

DEMOGRAPHIC DATA :

- Twenty-four cases were included
- Sex-ratio: 2
- Number of families : 24

RESULTS

CLINICAL DATA :

- Psychomotor development was normal : 1 case
- Mean age of onset of seizures: 12.32 months [40 days , 8 years]
- Neonatal onset: 7 cases.
- **Seizures type:** focal motor in the majority of cases (n=10).
- **Epileptic syndromes**:
  - Dravet syndrome(n=6)
  - Epilepsy of infancy with migrating focal seizures (n=4)
  - West syndrome (n=1)
  - Progressive myoclonic epilepsy (n=1)
- Examination results are summarized in Figure 1.

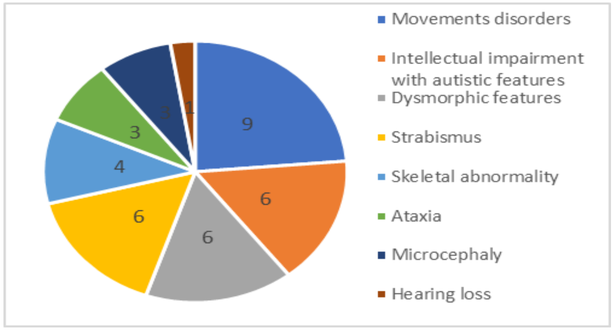

Figure 1: Clinical feautres in our patients

ELECTROPHYSIOLOGICAL DATA (Figure 2)
RADIOLOGICAL DATA (Figure 3)

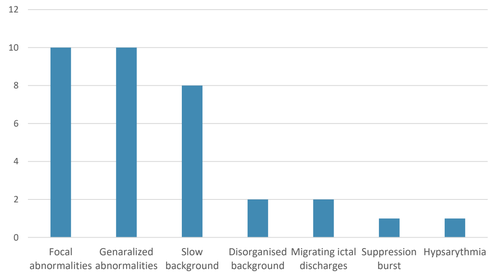

Figure 2: Electophysiological assessments in patients with DEE

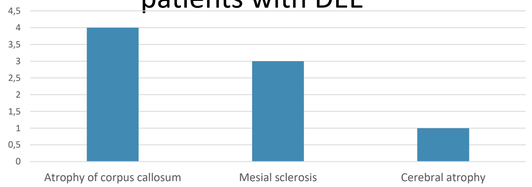

Figure 3: MRI assessments in patients with DEE

- Gene identified(figure 4)

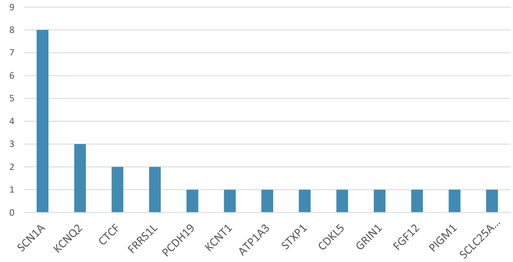

Figure 4: Genes identified in patients with DEE

- Lamotrigine was exacerbating all cases of DEE linked to the SCN1A gene.
- Valproic acid was used in all cases, with marked improvement in patients followed up for DEE linked to the CTCF gene.

DISCUSSION

- Dravet Syndrome (1) is the most common epileptic encephalopathy in our population as described in the literature.
- Epilepsy genes often present phenotypic pleiotropy, where a specific mutated gene can present various phenotypic characteristics. This is exemplified in our patients with SCN1A mutation.
- Genetic heterogeneity (1) is also commonly observed in electroclinical syndromes, when different genes can produce one phenotype.
- It has been noted in patients with Dravet syndrome due to SCN1A (2) and PCDH 19 mutations(1) .

CONCLUSIONS

- Interpretation of genetic findings in clinical context can often be complex. It should be robust, considered in the context of the electro-clinical presentation.
- Our study highlights heterogeneity of DEE and the importance of genetic screening(2) .
- Advances in genetics have made it possible to develop specific therapies, predict prognosis and establish genetic counseling.

REFERENCES

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