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IS GENETIC ETIOLOGY AN ABSOLUTE CONTRAINDICATION FOR EPILEPSY SURGERY? DESCRIPTION OF TWO PATIENTS WITH GENETIC-RELATED EPILEPSY WHO UNDERWENT PRE-SURGICAL STUDY

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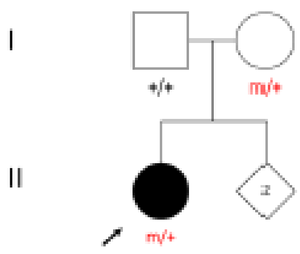
Purpose

Focal epilepsy (FE) was long thought to originate from focal lesions, whereas genetic factors caused generalized epilepsy. This historic over-simplification has been disrupted by new evidence that FE can be due to genetic mutations. A growing number of papers reported malformation of cortical development connected to pathogenic mutation of DEPDC5 and SCN1A. Some anecdotal studies recently described surgical series of patients with mainly lesional epilepsy related to DEPDC5 and SCN1A; preliminary results seem to indicate that the surgical approach could be effective in DEPDC5-related epilepsy, while it provided no benefits in SCN1A-related epilepsy [1,2]

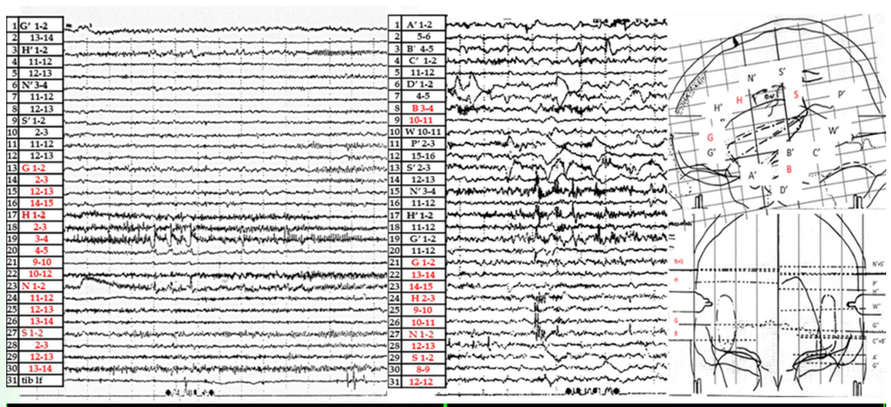
Case Series

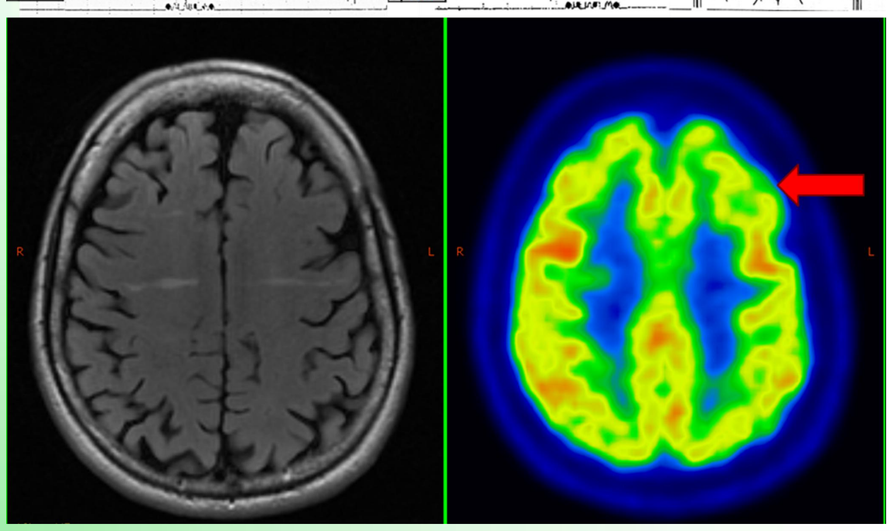
• PATIENT 1 ♀ 43y

- Drug-resistant sleep-related hypermotor epilepsy since she was 15.
- Interictal EEG: bilateral epileptiform activity with left frontal prevalence
- Ictal EEG: masked by muscular artifacts.
- Multiple MRI examination showed no alteration



+ DEPDC5 c.492delTCGTT; p.R165Yfs*13
Discovered years later than SEEG study



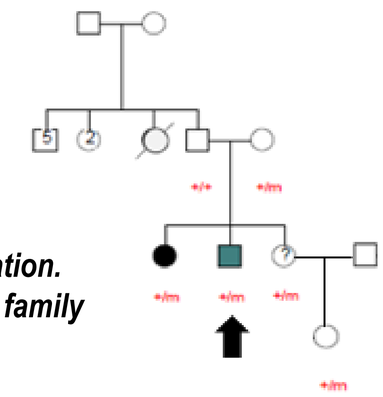


- *SEEG showed bilateral epileptiform activity, prevailing on the right anterior cingulate cortex, suggestive of focal cortical dysplasia.*
- *Unclear anatomo-electroclinical correlation, surgical intervention was rejected.*
- *Recently a PET study identified a focal hypometabolic area in the left frontal lobe.*

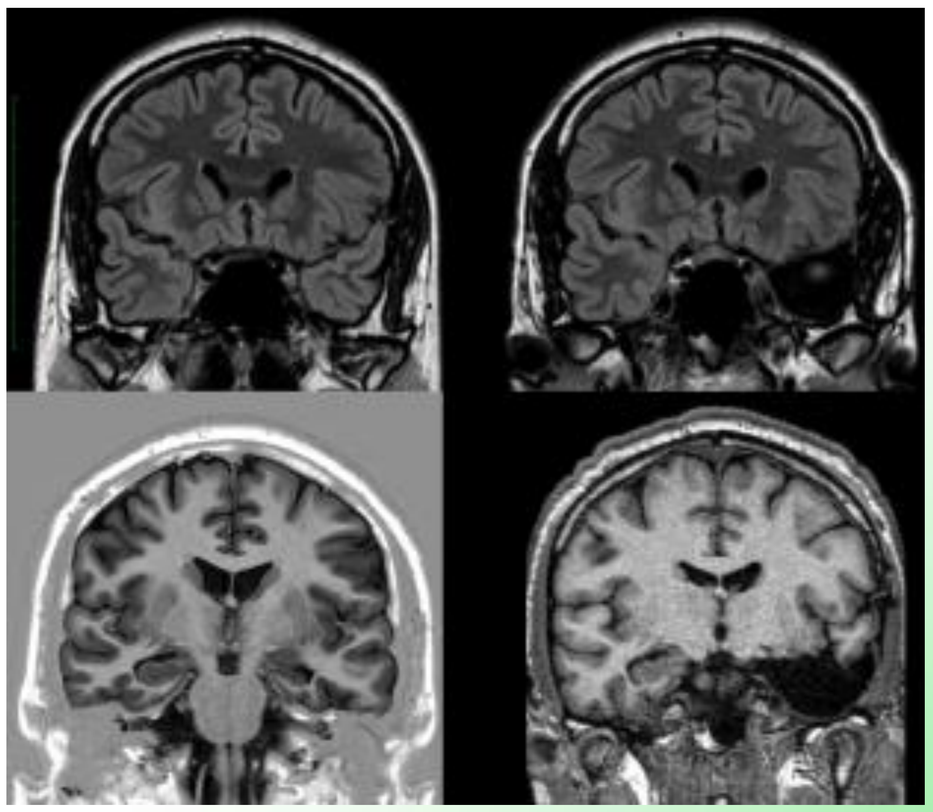
• PATIENT 2 ♂ 27y

- Drug-resistant temporal lobe epilepsy since he was 20.
- Interictal EEG: epileptiform activity on left temporal regions
- Ictal EEG: typical episode related to left temporal ictal discharge
- MRI: left hippocampal sclerosis

● Dravet Syndrome
○ Epilepsy with FS



+ SCN1A (p.Glu1881Lys) missense mutation.
Generalized Epilepsy Febrile Seizures + family



- *Concordant anatomo-electroclinical correlation.*
- *He underwent left temporal lobectomy; seizure-free for 1 year..*
- *Histology: Hippocampal sclerosis type 1.*

Discussion

Focal genetic epilepsy could be related both to a lesional and to an apparently non-lesional etiology. We suggest that a genetic analysis should be performed in patients with undefined anatomo-electroclinical correlations and non-lesional drug-resistant epilepsy. Surgical approach should be considered in the presence of a clear epileptogenic zone.

The genetic etiology is not an absolute exclusion criterion for surgery, but may require a more comprehensive surgical risk/benefit assessment

1 Baulac S, Ishida S, Marsan E et al. Familial focal epilepsy with focal cortical dysplasia due to DEPDC5 mutations. Ann Neurol. 2015 Apr;77(4):675-83
2 Skjei KL, Church EW, Harding BN et al.Clinical and histopathological outcomes in patients with SCN1A mutations undergoing surgery for epilepsy. J NeurosurgPediatr. 2015 Dec;16(6):668-74.