FOCAL CORTICAL DYSPLASIAS SECONDARY TO DEPDC5 WITH GOOD RESPONSE TO KETOGENIC THERAPY

Álamo R., Gonzalez M. G., Jorrat P., Diez C., Querze D., Villanueva M, Arroyo H., Schteinschnaider A.
Neuropediatric Departament, FLENI. Buenos Aires, Argentina.

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INTRODUCTION

The DEPDC5-gene participates in the mTOR-pathway, its mutation initially related to autosomal dominant non-lesional familial focal epilepsy, is currently associated with focal cortical dysplasias (FCD).(2)

OBJECTIVE

Our objective is to describe two patients with refractory focal epilepsy secondary to DCF due to gene mutations with good response to Ketogenic Therapy (KT).

METHOD

Retrospective and descriptive review, based on digital patient medical records.

CONCLUSION

Epilepsy related to DEPDC5-gene in 54% can become drug-resistant (3). KT acts by inhibiting the mTOR-pathway, which is why it could be considered useful while waiting for an eventual surgical resolution.(4)

The reported patients exemplify the improvement in seizure control after the start of KT, allowing in the second one, a stable evolution until the surgery is scheduled and in both cases, improving quality of life.

PATIENT 1

A 2 y-old boy, begins at 2 months with focal motor seizures, compromised consciousness progressing to epileptic spasms.

EEG: Epileptic encephalopathy and left temporal discharges.

Neuroimaging: Brain MRI with eft temporal DCF.

Genetic Test: heterozygous pathogenic mutation in DEPDC5-gene in an exome sequencing panel.

Evolution: Turns refractory to drugs and without interdisciplinary consensus for epilepsy surgery, at 18 months he started modified CT (1.5:1) with a 50% reduction in seizures at 2 months and more than 90% at 22 months.).

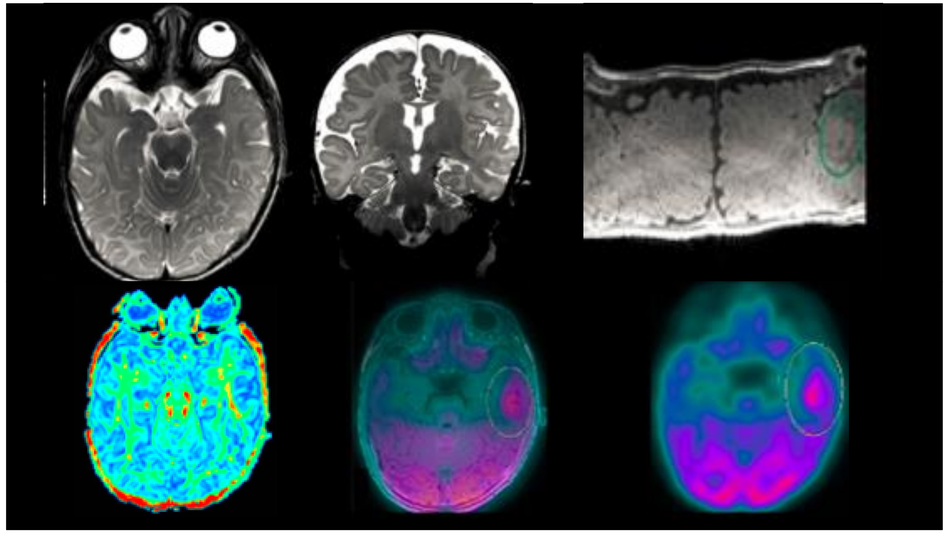

Figure A: Axial section T2. **Figure B**: T2 coronal section. In the left temporal lobe, an area of alteration in gray-white matter differentiation is observed in the middle temporal gyrus that extends to the superior temporal gyrus, compatible with cortical dysplasia. **Figure C:** Cortical deployment. The previously mentioned cortical dysplasia can be observed. **Figure D:** Positron emission tomography. A hypermetabolic area is observed at the level of the middle temporal gyrus.

PATIENT 2

A 4 y-old boy, begins at 13 months with focal motor seizures with partial compromise of consciousness.

EEG: Right central-parietal dicharges.

Neuroimaging: Right postcentral parietal DCF in the brain MRI.

Genetic Test: heterozygous pathogenic mutation in the exome sequencing panel in DEPDC5-gene.

Evolution: No response to first and second-line drugs. Start modified TC (2:1) with a 90% reduction in seizures per month. Epilepsy surgery was scheduled with a good outcome.

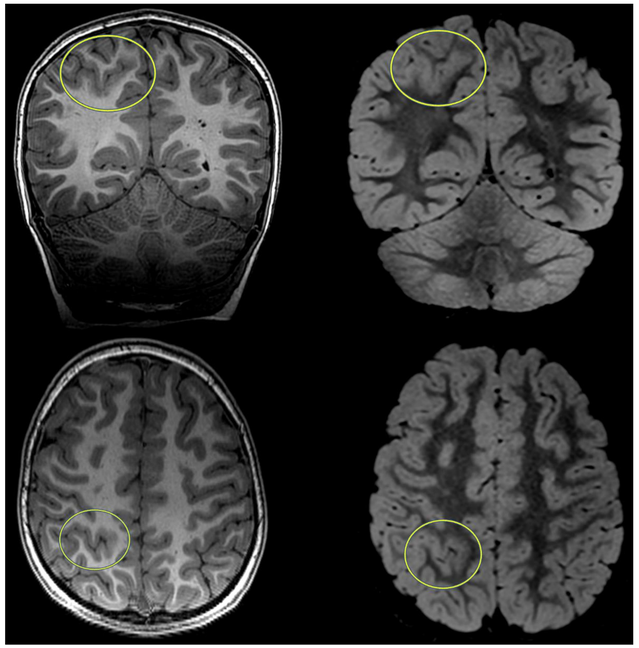

Figure E and F: T1 and T2 coronal section. **Figure G and H:** Axial section in T1 and T2/FLAIR. Deepening of a right precentral frontal cortical sulcus, associated with verticalization of the ipsilateral central sulcus, compared to its contralateral. Alteration in gray/white matter differentiation in right postcentral parietal topography.

Contact: angeles@fleni.org.ar