Characterization of a gene of unknown function involved in a form of familial epilepsy

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Research question and background

By exome sequencing, we identified in seven of eight reported families with FFEVF a mutations in a novel unknown gene, called DEPDC5. In addition, we have shown that the mutations in this gene account for approximately 12% of non-lesional focal epilepsy in small families. Shared homology with G protein signalling molecules and localization in human neurons suggest a role for this gene in neuronal signal transduction. Our study reveals that the mutations identified in this gene make an important contribution to familial focal epilepsies and understanding its function will provide critical insights into these disorders.

Methods and tissues used

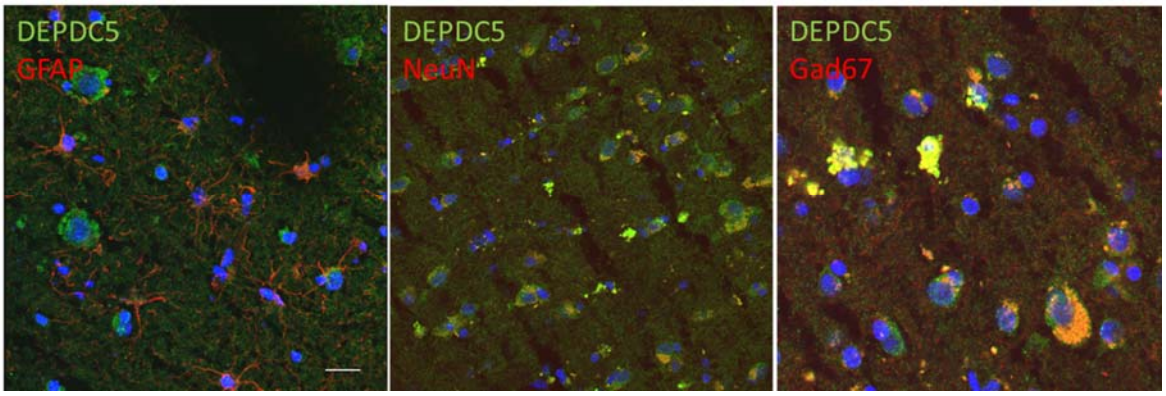
So far, we were able to provide information regarding localization and expression in mouse brain tissue. Unfortunately we do not have any human brain tissue available and for this reason we requested some tissues to the Netherlands Brain Bank.

Methods**:** By using a cryostat, we cut sections of about 8-10µm of human medial frontal gyrus provided by the Netherlands Brain Bank. We fixed them in 4% formalin and stained them for NeuN, GAD67, GFAP and DEPDC5.

Results and conclusion

We assessed the localization of DEPDC5 in tissue and in which cell types it was enriched. We observed no co-localization of DEPDC5 with GFAP (glial marker), but some co-localization with NeuN (neuronal marker) and gad67 (GABAergic marker). The meaning of these findings is still under investigation. However the staining protocol has still to be improved in order to obtain more convincing staining for the localization of DEPDC5.

Figure 1. Co-localization of DEPDC5 with GFAP, NeuN, GAD67.



No papers have been published with these data yet.