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Complete Agenesis of Corpus Callosum in *KCNQ2*-Related Neonatal Epileptic Encephalopathy

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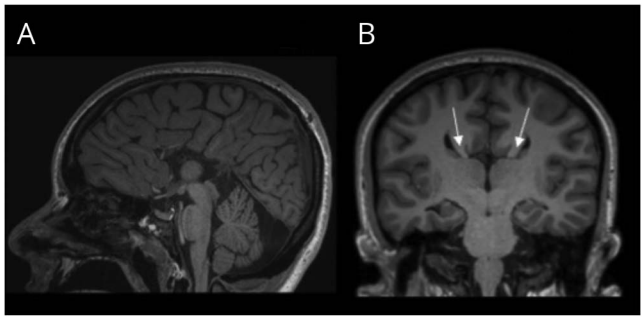
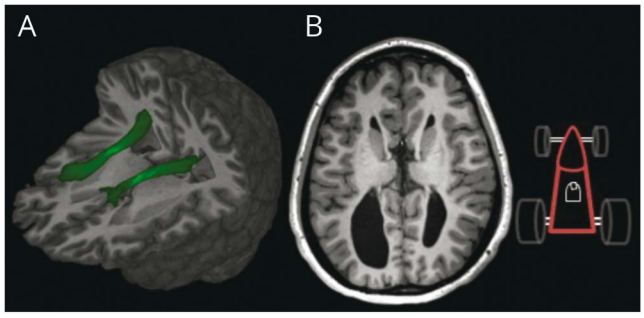
Neurol Genet 2022;8:e200042. doi:10.1212/NXG.0000000000200042

Figure 1 Complete Agenesis of Corpus Callosum and Probst Bundles



(A) Sagittal T1-weighted image: complete agenesis of corpus callosum. (B) Coronal T1-weighted image: probst bundles (arrows) (i.e., aberrant collections of axons that would normally form the corpus callosum but fail to cross the midline); hippocampal malrotation.

Figure 2 Tractography and Racing-Car Sign



(A) 3D-DTI tractography reconstruction: probst bundles running parallel to the interhemispheric fissure with anteroposterior orientation. (B) Axial T1-weighted image: asymmetric "racing-car sign." DTI = diffusion tensor imaging.

This 22-year-old female patient presented with tonic seizures on the second day of life. Psychomotor delay and intellectual disability then became evident. Seizures were initially controlled by phenobarbital, except for rare relapses. The patient experienced 1–2 seizures/yr between ages 12 and 20 years; since then, she has been seizure-free on oxcarbazepine and valproate.

She also had partial growth hormone deficiency.

EEG showed posterior epileptiform abnormalities. 3T-brain MRI disclosed complete agenesis of corpus callosum (ACC) (Figures 1 and 2).

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Go to Neurology.org/NG for full disclosures. Funding information is provided at the end of the article.

The Article Processing Charge was funded by the authors.

Karyotype and Array - Comparative Genomic Hybridization were normal. An in silico ACC multigene panel extracted from whole exome sequencing (WES; mean coverage: ×52; read length: 150 bp) in 2021 was negative.

WES analysis showed a heterozygous missense variant in *KCNQ2* (NM_172107.4), p.Arg353Cys, arising de novo. The ACMG guidelines[1] classify the variant as pathogenic (PM1, PM5, PM2, PP2, PP3, PP5, PS1, PS2).

The patient was diagnosed with *KCNQ2*-related neonatal epileptic encephalopathy.

KCNQ2-associated brain abnormalities include thinning of the corpus callosum[2] but complete ACC has never been reported.

Acknowledgment

The authors thank the patient and her family for participating. Thanks to the Epi25 consortium, supported by National Human Genome Research Institute Grant UM1 HG008895.

Study Funding

The authors report no targeted funding.

Disclosure

The authors report no disclosures relevant to the manuscript. Full disclosure form information provided by the authors is available with the full text of this article at Neurology.org/NG.

Publication History

Received by *Neurology: Genetics* June 29, 2022. Accepted in final form September 22, 2022. Submitted and externally peer reviewed. The handling editor was Stefan M. Pulst, MD, Dr med, FAAN.

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