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

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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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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 guidelines1 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 callosum2 but complete ACC has never been reported.

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### Appendix Authors

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