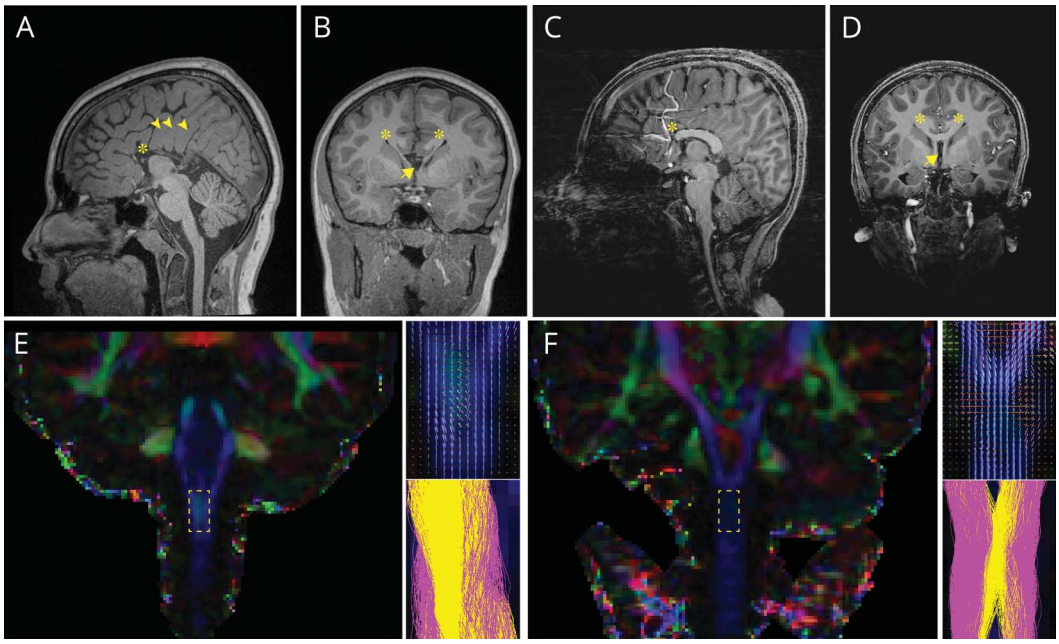
Teaching NeuroImages: Imaging features of *DCC*-mediated mirror movements and isolated agenesis of the corpus callosum

Timothy J. Edwards, MBBS,* Ashley P.L. Marsh, PhD,* Paul J. Lockhart, PhD, Linda J. Richards, PhD, and Richard J. Leventer, MBBS, PhD

Correspondence
Dr. Leventer
Richard.Leventer@rch.org.au

Neurology® 2018;91:e886-e887. doi:10.1212/WNL.0000000000006085

Figure T1-weighted MRI and color fractional anisotropy (FA) maps



MRI shows complete (A, B) and partial (C, D) isolated agenesis of the corpus callosum with radial gyri and absence of the cingulate gyrus (arrowheads, A), unfused and thickened septum pellucidum (arrows, B and D) and Probst bundles (asterisks, B and D). Neurotypical (E) and *DCC*+/− (F) color FA maps show decreased corticospinal decussation (bottom inset: yellow/crossed, purple/uncrossed) and corresponding fiber orientation distribution (top insets).

Two unrelated children were prenatally diagnosed with isolated agenesis of the corpus callosum (iACC) in otherwise uneventful pregnancies. Postnatal clinical assessments identified mirror movements in these offspring, their siblings, and their respective mothers. MRI (figure) showed characteristic features of complete (A, B) and partial (C, D) iACC, and abnormal crossing of the corticospinal tracts (E, F) on diffusion imaging. Sequencing revealed monoallelic missense mutations in the axon guidance receptor *DCC*.[1] The association of iACC and abnormal corticospinal decussation is unique to only a handful of genes known to cause agenesis of the corpus callosum,[2] and can provide a clinical clue towards a genetic diagnosis.

MORE ONLINE

→**Teaching slides**
links.lww.com/WNL/A645

*These authors contributed equally to this work.

From the Queensland Brain Institute (T.J.E., L.J.R.), Faculty of Medicine (T.J.E.), and School of Biomedical Sciences (L.J.R.), The University of Queensland, Brisbane; Bruce Lefroy Centre for Genetic Health Research (A.P.L.M., P.J.L.) and Neuroscience Research Group (R.J.L.), Murdoch Children's Research Institute, and Department of Neurology (R.J.L.), Royal Children's Hospital; and Department of Paediatrics (A.P.L.M., P.J.L., R.J.L.) University of Melbourne, Parkville, Victoria, Australia.

Go to Neurology.org/N for full disclosures. Funding information and disclosures deemed relevant by the authors, if any, are provided at the end of the article.

Author contributions

Dr. Edwards: study concept and design, analysis and interpretation. Dr. Marsh: study concept and design, analysis and interpretation. Dr. Lockhart: critical revision of the manuscript for important intellectual content. Dr. Richards: critical revision of the manuscript for important intellectual content, study supervision. Dr. Leventer: study concept and design, acquisition of data, critical revision of the manuscript for important intellectual content, study supervision.

Study funding

No targeted funding reported.

Disclosure

The authors report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

References

1. Marsh APL, Heron D, Edwards TJ, et al. Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. Nat Genet 2017;49:511–514.
2. Edwards TJ, Sherr EH, Barkovich AJ, Richards LJ. Clinical, genetic and imaging findings identify new causes for corpus callosum development syndromes. Brain 2014;137:1579–1613.

Teaching NeuroImages: Imaging features of *DCC*-mediated mirror movements and isolated agenesis of the corpus callosum

Timothy J. Edwards, Ashley P.L. Marsh, Paul J. Lockhart, et al.

Neurology 2018;91;e886-e887
DOI 10.1212/WNL.0000000000006085