A loss-of-function mutation in the CFC domain of TDGF1 is associated with human forebrain defects

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Author(s) de la Cruz, June M.; Bamford, Richard N.; Burdine, Rebecca D.; Roessler, Erich; Barkovich, James A.; Donnai, Dian; Schier, Alexander F.; Muenke, Maximilian
Author(s) at UniBasel Schier, Alexander ;
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TDGF1 (CRIPTO) is an EGF-CFC family member and an obligate co-receptor involved in NODAL signaling, a developmental program implicated in midline, forebrain, and left-right axis development in model organisms. Previous studies of CFC1 (CRYPTIC), another member of the EGF-CFC family, demonstrated that normal function of this protein is required for proper laterality development in humans. Here we identify a mutation in the conserved CFC domain of TDGF1 in a patient with midline anomalies of the forebrain. The mutant protein is inactive in a zebrafish rescue assay, indicating a role for TDGF1 in human midline and forebrain development.
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