

POSTER PRESENTATION

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Telencephalic morphogenesis is impaired in *Ftm/Rpgrip1l* KO mice

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Primary cilia have essential functions in vertebrate development and signaling. However, little is known about cilia function in brain morphogenesis, a process that is severely affected in human ciliopathies. Here, we study telencephalic morphogenesis in a mouse mutant for the ciliopathy gene *Ftm/Rpgrip1l* (also called *NPHP8*, *JBTS7* or *MKS5*). We have previously shown that E12.5 *Ftm*^{-/-} telencephalic neuroepithelial cells lack primary cilia and that the telencephalon is ventralized at this stage. This dorso-ventral (DV) patterning defect leads to an ectopic localization of the olfactory bulb primordium, which consequently does not display morphological outgrowth at the end of gestation. This phenotype is reminiscent of *Gli3* mutant and indeed correlates with a decreased production of the short, repressor form of *Gli3* (*Gli3R*). We then demonstrate that the main function of primary cilia in the developing brain is to permit *Gli3* processing, since introduction of *Gli3R* in *Ftm*^{-/-} background is sufficient to rescue DV patterning and OB morphological defects, despite the persistence of the cilia defects. In addition, we observed an impairment of the neocortex in *Ftm* mutant at the end of gestation. Cortical lamination is impaired, showing patches of very abnormal regions lying between regions that are less severely affected. This heterogeneity correlates with a disorganization of radial glial fibres and a reduction of the thickness of the ventricular layers and of the cortical plate. Investigating the developmental origin of these defects should shed light on aspects of physiopathology in human diseases.

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