CDB SEMINAR

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16:00~17:00 A7F Seminar Room

The human ciliopathy protein JBTS17 is required for basal body docking and intraflagellar transport for ciliogenesis

Summary

Ciliopathies are a broad class of human disease which share an etiology of defective cilia structure or function. Jbts17 (aka C5orf42) is a recently identified gene which is mutated in human patients with Joubert, Meckel-Gruber, and Oral-Facial-Digital Type VI syndromes, but it's cellular or molecular function is not understood. Here, we report the expression and functional characterization of Jbts17 in *Xenopus* embryos and elucidate the pathogenic mechanism of human ciliopathy disease. Jbts17 is highly expressed in neural tissues and multiciliated cells (MCCs). The expression of sonic hedgehog target genes, which is mediated by primary cilia function, is attenuated in Jbts17 knockdown embryos and resulted in showing several developmental abnormalities relating to neural tube closure results in eye formation, and embryonic growth. Jbts17 is strongly localized at basal body in MCCs, and Jbts17 knockdown cells fail to dock basal bodies to the apical membrane. We also identified the functionally important domain for ciliogenesis, which is truncated in some human ciliopathy patients. The normal expression and localization of Jbts17 at basal bodies is required for recruitment of Inturned and Rsg1, which are also involved in basal body docking. Moreover, Intraflagellar transport protein localization at basal bodies and trafficking in axonemes, are essential for protein trafficking in cilia which is disturbed in Jbts17 knockdown cells. We propose that Jbts17 has an essential function for ciliogenesis by controlling basal body docking and IFT protein trafficking in cilia.

This research was done at the laboratory of Dr. Wallingford, University of Texas at Austin.

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