Highlight: Researchers uncover new data about Arl13b function in Joubert syndrome

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Researchers in Ireland have gained new understanding of the role played by the cilial protein Arl13b in Joubert syndrome (JS), a rare disorder characterized by developmental delay, mental retardation, and low muscle tone, among other symptoms.

The findings will be published online March 15 in the Journal of Cell Biology.

Although Arl13b—which is required for cilium biogenesis and embryo development—is known to be mutated in patients with JS, the specific cilial and molecular basis of Arl13b function has been poorly understood. Oliver Blacque and colleagues (University College Dublin) used C. elegans and mammalian cell culture systems to investigate Arl13b function.

Their findings show that JS-associated Arl13b works at ciliary membranes, where it regulates ciliary transmembrane protein localization and transport of proteins to the tip of the cilium.

More information: Cevik, S., et al. 2010. J. Cell Biol. doi:10.1083/jcb.200908133

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