

POSTER PRESENTATION

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The lebercilin-like protein is embedded in a ciliary protein network and is preferentially expressed in motile cilia

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Mutations in LCA5 are causative for Leber congenital amaurosis, a severe hereditary retinal dystrophy in humans. Lebercilin, encoded by LCA5, localizes to connecting cilia of photoreceptor cells in the retina and specifically interacts with the intraflagellar transport (IFT) machinery. Bioinformatic analysis has identified lebercilin-like protein, previously known as C21orf13, as a lebercilin homolog in humans. In this study, we have characterized the molecular properties of lebercilin-like protein by defining the lebercilin-like interactome and assessing its (sub)cellular localization in ciliated cells. We show that lebercilin-like protein is embedded in a ciliary protein network and specifically localizes at the basal body and ciliary axoneme of ciliated cells, like lebercilin. mRNA expression studies indicate that lebercilin-like protein is preferentially expressed in tissues featuring motile cilia and/or flagella. Based on these data and bioinformatic co-expression profiling, we suggest that LCA5L is a likely candidate gene for motile ciliopathies such as Primary Ciliary Dyskinesia (PCD).

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