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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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From First International Cilia in Development and Disease Scientific Conference (2012)
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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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