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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

DA Mans^{1*}, KLM Coene¹, K Boldt², IJC Lamers¹, J van Reeuwijk¹, NT Loges³, E Bolat¹, L Franke⁴, L Hetterschijt¹, SJF Letteboer¹, TA Peters^{5,6}, H Omran³, FPM Cremers¹, M Ueffing^{2,7}, R Roepman¹

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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Author details

¹Department of Human Genetics, Nijmegen Centre for Molecular Life Sciences, Radboud University Nijmegen Medical Centre, the Netherlands.

²Division of Experimental Ophthalmology and Medical Proteome Center, Center of Ophthalmology, University of Tübingen, Germany.

³Children's Department, University Hospital Münster, Germany.

⁴Department of Genetics, University Medical Center Groningen and University of Groningen, Germany. ⁵Department of Otorhinolaryngology, Nijmegen Centre for Molecular Life Sciences, Radboud University Nijmegen Medical Centre, the Netherlands. ⁶Donders Institute for Brain, Cognition and Behaviour, the Netherlands. ⁷Department of Protein Science, Helmholtz

* Correspondence: D.Mans@antrg.umcn.nl

¹Department of Human Genetics, Nijmegen Centre for Molecular Life Sciences, Radboud University Nijmegen Medical Centre, the Netherlands
Full list of author information is available at the end of the article

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