

CG31320 (*Heatr2*) - ciliopathy candidate gene, functional analysis in fly and mouse models

G Mali^{1*}, P Mill¹, PI zur Lage², EA Hall¹, AP Jarman², IJ Jackson¹

From First International Cilia in Development and Disease Scientific Conference (2012)
London, UK. 16-18 May 2012

The structural and functional roles of many of the 800-1000 proteins that make up the microtubule core and specialized membranes of cilia and flagella are poorly understood. Following from our recent expression study to identify putative ciliary candidates in *Drosophila* sensory neurons, we focused on a subset that were targets of the transcription factor Fd3f, which regulates functional specialization of mechanosensory cilia. Bioinformatic enrichment for known ciliary domains as well as orthologous protein-protein interaction network modelling provided a list of putative ciliary genes for further functional characterization. One such candidate, *CG31320*, has been initially characterized in *Drosophila*. Little is known about this gene, except the encoded protein contains HEAT repeats – belonging to an armadillo-like fold family associated with intracellular transport. In situ analysis confirms that *CG31320* mRNA is highly expressed in the ciliated chordotonal neurons. RNAi-mediated knock-down resulted in abnormal chordotonal ciliary morphology and locomotory defects, consistent with impaired mechanosensory cilium function. Currently, we are studying whether the ortholog *Heatr2* is also required for mammalian cilia. Protein localization studies suggest that *Heatr2* plays a role in trafficking to primary cilia. RNAi knock-down and protein interaction studies using mammalian cells are underway to functionally dissect *Heatr2* roles; results will be presented. We are generating a *Heatr2* conditional mouse mutant to investigate its function in different types of cilia and sperm flagella. We present a multisystem experimental pipeline for functional characterization of novel genes expressed in cilia as well as putative ciliopathy candidates.

Author details

¹MRC Human Genetics Unit, MRC IGMM, University of Edinburgh, UK.
²University of Edinburgh, UK.

Published: 16 November 2012

doi:10.1186/2046-2530-1-S1-P92

Cite this article as: Mali et al.: *CG31320 (Heatr2)* - ciliopathy candidate gene, functional analysis in fly and mouse models. *Cilia* 2012 1(Suppl 1): P92.

Submit your next manuscript to BioMed Central and take full advantage of:

- Convenient online submission
- Thorough peer review
- No space constraints or color figure charges
- Immediate publication on acceptance
- Inclusion in PubMed, CAS, Scopus and Google Scholar
- Research which is freely available for redistribution

Submit your manuscript at
www.biomedcentral.com/submit



* Correspondence: Girish.Mali@hgu.mrc.ac.uk

¹MRC Human Genetics Unit, MRC IGMM, University of Edinburgh, UK
Full list of author information is available at the end of the article