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POSTER PRESENTATION

Open Access

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

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

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