Blind mice shed light on human sight loss

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Mutant mice could provide genetic clues to understanding incurable human sight loss resulting from retinal degeneration. Research published in the online open access journal *Genome Biology* uncovers a role for microRNA in retinal disease, and may point the way to future therapies.

A team from the Trinity College Dublin and the Sanger Institute, Cambridge (UK), led by Dr Arpad Palfi and Dr Jane Farrar of the Smurfit Institute of Genetics, Trinity College Dublin used mutant mice that model the human eye disease retinitis pigmentosa (RP). The researchers compared these mice with wild-type mice, testing their hypothesis that changes in microRNA expression may be evident in retinal degeneration.

Retinitis pigmentosa is the most common form of inherited retinal degeneration affecting more than one million individuals worldwide. Progressive photoreceptor cell death eventually leads to blindness. Mutations in more than 40 genes have been linked to the disease and no therapy is currently available.

The team found very similar patterns of microRNA expression in retinas of two wild-type mouse strains, but, microarray profiling revealed that in these wildtype mice the patterns of microRNA expression differed between the brain and retina. Furthermore, there were clear differences in the microRNA expression patterns between wild type and mutant mice. The researchers found alterations greater than two-fold in the expression of 9 microRNAs in mutant mouse retinas compared with those of the wild-type mice. These microRNAs potentially regulate

genes implicated in retinal diseases and genes encoding components involved in cell death and intracellular trafficking.

"The results from the study suggest that miRNA expression is perturbed during retinal degeneration" says Dr Jane Farrar of Trinity College Dublin. "Modulation of expression of retinal microRNAs may possibly represent a future therapeutic strategy for retinopathies such as retinitis pigmentosa."

Source: BioMed Central