

New mouse mutant contains clue to progressive hearing loss

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Researchers have defined a mutation in the mouse genome that mimics progressive hearing loss in humans. A team from the Wellcome Trust Sanger Institute in Cambridge, UK, working with colleagues in Munich and Padua, found that mice carrying a mutation called Oblivion displayed problems with the function of hair cells in the inner ear, occurring before clear physical effects are seen. The study is published October 31 in the open-access journal *PLoS Genetics*.

Progressive loss of hearing affects around six out of ten people over the age of 70. Whilst environmental causes can contribute, genetic influences also play a major role. Although we know of many genes involved in deafness in childhood, most of these genes contribute only rarely to progressive hearing loss in humans and their role is poorly understood.

The team found that the Oblivion mutation showed features in common with forms of human deafness. In mice with one mutant copy of the Oblivion gene, the hair cells showed some function at first but later degenerated; in mice with two mutant copies, the hair cells were already damaged at birth.

"When we mapped the mutation to the mouse genome, we quickly found a probable cause for hearing loss," explains senior author Professor Karen Steel. "We showed that the mutant mice carried a change in one letter of their genetic code in a gene called *Atp2b2*. Changing a specific C to a T in this gene stops it from producing a normal molecular pump

that is needed to keep hair cells in the ear working efficiently by pumping excess calcium out of the cell."

Although other mutations have been described, Oblivion is unique in the way it leads to hearing loss due to mutations in *Atp2b2*, a gene which has previously been implicated in hearing loss in humans. "One aim of identifying and characterizing mice with impaired hearing is to help us to understand the biology of this remarkable sense," says Professor Steel. "Improving our understanding of the molecular and cellular action of genetic variants will help us to develop improved diagnostics and improved treatments for humans."

Citation: Spiden SL, Bortolozzi M, Di Leva F, de Angelis MH, Fuchs H, et al. (2008) The Novel Mouse Mutation Oblivion Inactivates the PMCA2 Pump and Causes Progressive Hearing Loss. *PLoS Genet* 4(10): e1000238. doi:10.1371/journal.pgen.1000238
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