Novel genetic loci identified for high-frequency hearing loss

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The genetics responsible for frequency-specific hearing loss have remained elusive until recently, when genetic loci were found that affected high-frequency hearing. Now, a study published today in the open access journal *BMC Genetics* reports, for the first time, genetic loci with effects that are limited to specific portions of the hearing frequency map, particularly those that are most affected in ageing-related hearing loss.

Presbycusis is the loss of hearing for high-pitched sounds that gradually occurs in most individuals as they grow older. Although many genetic loci have been linked to hearing deficits in humans, many loci that contribute to tonotopy, i.e. the organization of the auditory system that permits detection and discrimination of sounds of different frequency, remain undiscovered.

A group from the National Institute on Deafness and Other Communication Disorders (NIDCD) at the National Institutes of Health (NIH), used genome-wide linkage analysis in NIH Swiss mice to successfully identify two quantitative trait loci that affect hearing at high frequencies – Hfhl1 and Hfhl3. Specifically the effect of the locus Hfhl1 is thought to be confined to hearing frequencies from 25-44kHz of the tonotopic map, whilst Hfhl3 is restricted to the 35-44kHz region.

Lead author James M Keller commented, "Our results support the hypothesis that frequency-specific hearing loss results from variation in gene activity along the cochlear partition and suggest a strategy for

creating a map of genes that influence differences in hearing sensitivity and or vulnerability in restricted portions of the cochlea."

He continued, "The high-frequency hearing loss loci, Hfhl1 and Hfhl3, explain only a portion of the variation in high-frequency hearing loss observed in these mice. Other loci, and cross talk between genes at different loci, probably account for much of the remainder - in fact we detected a number of additional loci that could account for some of the residual variation. Additional genotyping and analysis could greatly increase our understanding of the genetic architecture of the HFHL phenotype."

More information: Genome-wide linkage analyses identify Hfhl1 and Hfhl3 with frequency-specific effects on the hearing spectrum of NIH Swiss mice James M Keller and Konrad Noben-Trauth *BMC Genetics* (in press)

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