

## Hearing loss clue uncovered

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(Medical Xpress)—Researchers from the Department of Otolaryngology at the University of Melbourne and the Department of Biochemistry and Molecular Biology at Monash University have discovered how hearing loss in humans is caused by a certain genetic mutation.

A novel genetic mutation was first identified in 2010 as causing hearing loss in humans. Researchers have now discovered that this mutation induces malfunction of an inhibitor of an enzyme commonly found in our body that destroys proteins. Published today in the *American Journal of Pathology*, this inhibitor is known scientifically as SERPINB6.

Dr Justin Tan of the University of Melbourne and lead author of the study said individuals who lack both copies of this good gene were reported to lose their hearing from 20 years of age.

"This is unusual because most people show gradual signs of age-related hearing loss from 60 years of age onwards but mutations in SERPINB6 accelerate this process. It is not yet clear how this mutation causes hearing loss."

Working with animal models induced with the condition, mice started to lose their hearing at three weeks of age, which is comparable to teenage years in humans. Hearing loss continued to worsen as the mice aged, a trend that was also noticed in humans. When the inner ears of these mice were examined under the microscope, the Melbourne team uncovered that tiny, specialised cells in the inner ear, responsible for hearing, had died.



These cells include, not only the <u>sensory hair cells</u> that detect <u>sound vibrations</u>, but also neighbouring cells that belong to a group of cells called fibrocytes.

Both types of cells are required to transform sound into electrical signals in our hearing nerve. Mutations affecting the sensory hair cells have been known for decades to cause hearing loss in humans but mutations affecting the fibrocytes remain uncommon.

"This is an exciting discovery for our hearing because the role of SERPINB6 as an inhibitor is now being unraveled," said Dr Tan.

## Provided by University of Melbourne

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