Medical Xpress

Electronic records help link genes to age-related hearing loss

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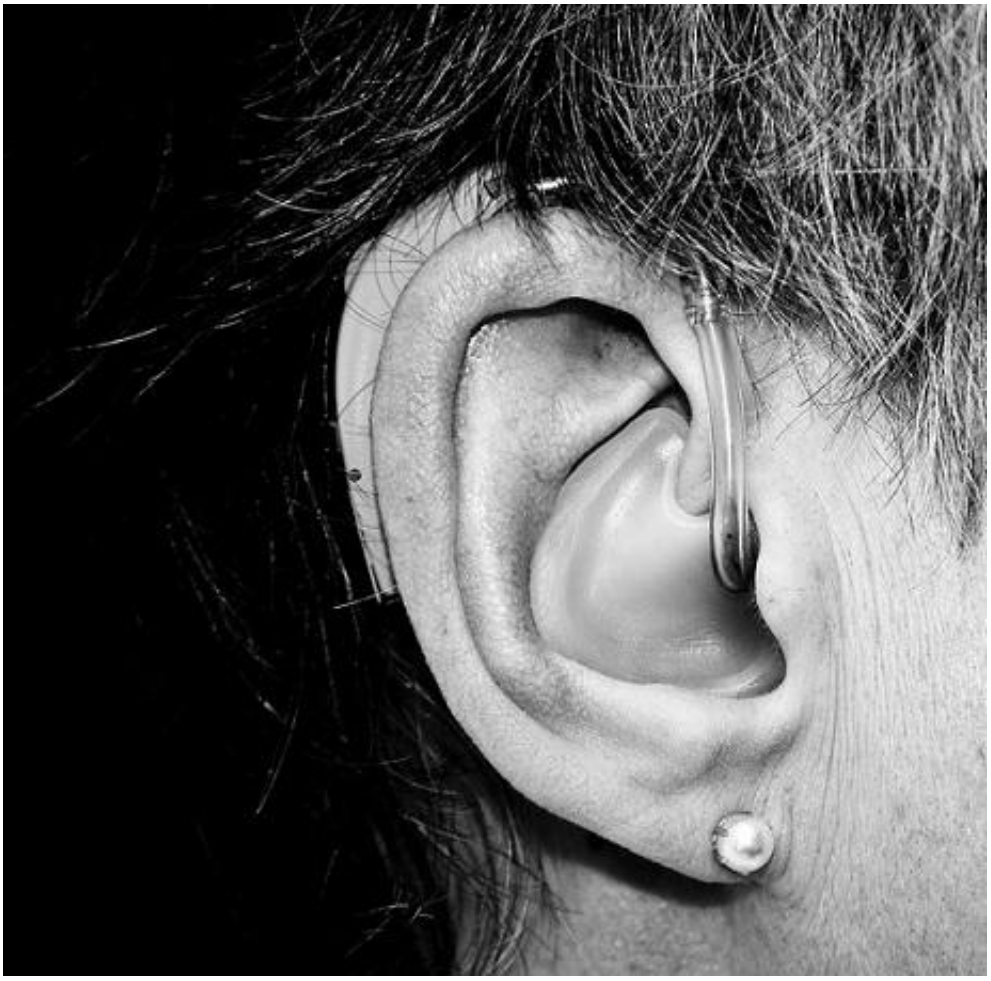

Age-related hearing impairment (ARHI) affects one quarter of individuals aged over 65 and half of individuals who are 80 and older. Credit: Steve Johnson, Flickr, CC BY

A study of patient electronic medical records and genome sequences from adults with age-related hearing impairment by researchers at UC San Francisco and Kaiser Permanente Northern California, identified two genetic variations linked to the hearing disorder.

The research—led by Thomas Hoffmann, PhD, a professor of epidemiology and biostatistics and member of the Institute for Human Genetics at UCSF—was published October 20, 2016 in *PLOS Genetics*.

"This is one of the first studies that has found and replicated genes linked with age-related hearing impairment," Hoffmann says. "And it provides some of the best evidence to date that age-related hearing loss may share a common biological basis with other forms of hearing impairment."

Age-related hearing impairment (ARHI) is a common condition, affecting one quarter of individuals aged 65, and half of individuals aged 80 and older. While hearing aids and other technologies offer treatments, scientists hope that a better understanding of the underlying genetics of the disorder may one day yield a cure.

To find genetic variations linked to ARHI, the scientists conducted a genome-wide association study using 6,527 age-related hearing impairment cases and 45,882 controls among white participants in the Genetic Epidemiology Research on Adult Health and Aging (GERA) cohort, a collaboration between UCSF and the Kaiser Permanente Research Program on Genes, Environment, and Health (RPGEH).

"Our collaboration with Kaiser is incredibly powerful," Hoffmann said. "They have a very long history of electronic health records on all of their patients in the system, so as a researcher you have this rich set of information that you can look at to understand the factors that drive health and disease."

The researchers discovered two genome variants that contribute to the disorder: a novel variation near the ISG20 gene, and a second variant within TRIOBP, a gene previously associated with another type of hearing loss.

To verify their findings, the scientists replicated the experiment in Latino, East Asian, and African American cohorts within GERA and in an independent cohort of individuals from the UK Biobank. They also looked at genes known to play a role in hearing loss and identified two additional genetic variations linked to ARHI.

This study provides novel insights into the genetic factors involved in ARHI. In particular, the discovery of the two additional variations in known hearing loss genes suggests that these may be regions of the genome to focus on in future studies of ARHI. The findings also highlight the utility of large cohorts with matched genomic data and electronic health records for revealing the underlying genetic basis of ARHI and other disorders.

"This has been a long process," Hoffmann says of the project to collect genomic data and link it with GERA health records. "It's really nice to see at the end that all the collaborative work that we've done is producing interesting results that help advance the field."

More information: *PLOS Genetics*, dx.doi.org/10.1371/journal.pgen.1006371

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