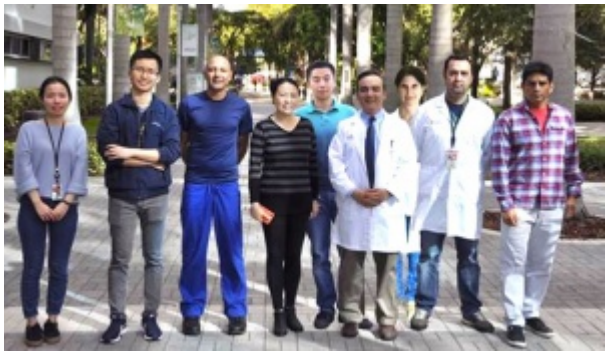
Miller School Researchers Discover New Genetic Cause of Deafness

Hearing loss is the most common sensory deficit worldwide. There is, however, a great deal that is still unknown about its causes.



From left, researchers in the study included Jiaqi Liu, Yi Zhu, M.D., Amjad Farooq, Ph.D., Grace Zhai, Ph.D., Chong Li, Ph.D., Mustafa Tekin, M.D., Katherina Walz, Ph.D., Guney Bademci, M.D., and Clemer Abad, D.V.M.

When parents are told their children are deaf or hard of hearing, they often have many questions, and one of the most common is: "Why?"

"In more than half of infants affected by hearing loss, genetic factors are involved," said Dr. Mustafa Tekin, M.D., a professor in the Dr. John T. Macdonald Foundation Department of Human Genetics and a member of the John P. Hussman Institute for Human Genomics at the University of Miami Miller School of Medicine, where he also serves as chief of the Clinical and Translational Genetics Division. "To date, we know of more than 100 genes that are linked to deafness, but they can explain only about half of cases with hearing loss."

A study by Miller School researchers and scientists at institutions in Turkey and China, led by Dr. Tekin, recently revealed that a mutation in the Grb-2-related adaptor protein (*GRAP*) gene is linked to hearing loss in people and fruit flies. An article on the *GRAP* gene's role as a genetic cause of deafness was published in the *Proceedings of the National Academy of Sciences* on Jan. 4.

The defect in the *GRAP* gene has been found in two unrelated Turkish families. Grace Zhai, Ph.D., an associate professor in the Department of Molecular and Cellular Pharmacology, conducted studies on the fruit fly counterpart of *GRAP* called *drk*. Flies with mutant *drk* were unable to detect gravity and keep their balance, which was due to defects in their hearing organ. Importantly, the normal human *GRAP* (but not the mutant form) can restore the sensory function of the *drk* mutant flies. The findings support the deleterious nature of the mutant *GRAP* identified in patients with hearing loss.

"We have not just found a new genetic cause of deafness," said Dr. Tekin. "This study also unveils a gene playing a fundamental role in hearing in different species, which will inform future research on therapeutic targets."

Currently, there are no cures for hearing loss. Symptoms are often treated with hearing aids or cochlear implants.

Other collaborators on this study include Chong Li, Ph.D., and Guney Bademci, M.D. (co-first authors), Oscar Diaz-Horta, Ph.D., Yi Zhu, M.D., Timothy Gavin Mitchell, Clemer Abad, D.V.M., Serhat Seyhan, M.D., Filiz Basak Cengiz, Ph.D., Susan H. Blanton, Ph.D., Amjad Farooq, Ph.D., and Katherina Walz, Ph.D.