MedicalXpress

Researchers identify gene mutation that causes rare form of deafness

July 12 2010

Researchers have identified a gene mutation that causes a rare form of hearing loss known as auditory neuropathy, according to U-M Medical School scientists.

In the study published online today in the Proceedings of the National Academy of Sciences USA, U-M's Marci Lesperance, M.D., and Margit Burmeister, Ph.D. led a team of researchers who examined the DNA of individuals from the same large family afflicted with the disorder.

The researchers identified a mutation in the DIAPH3 gene that causes over-production of a compound known as a diaphanous protein. In previous studies, hearing loss has been linked to a related gene that also affects a diaphanous protein.

Currently, diagnosing auditory neuropathy requires specific testing. Auditory neuropathy may be unrecognized if testing is not performed early in life.

"Since we previously knew of only two genes associated with auditory neuropathy, finding this gene mutation is significant," says Lesperance, professor in U-M's Department of Otolaryngology and chief of the Division of Pediatric Otolaryngology.

"This discovery will be helpful in developing genetic tests in the future, which will be useful not only for this family, but for all patients with auditory neuropathy," Lesperance says.

To investigate the role of these compounds in auditory function, the authors engineered a line of fruit flies that expressed an overactive diaphanous protein in the insects' auditory organ. Using sound to induce measurable voltage changes, Frances Hannan of New York Medical College determined that the flies' hearing was significantly degraded compared to normal flies.

Burmeister says finding the genes causing such rare disorders is very difficult because researchers cannot look at many different families, and instead have to rely on a single family that is often not large enough. But in this study, the researchers used a multi-pronged approach. Rather than relying purely on genetic inheritance information, they combined this information with biological function regarding gene activity.

"The approach we used here of combining genetic inheritance with functional information can be applied to identify the culprit genes in many other rare genetic diseases that have so far been impossible to nail down," says Burmeister, professor of Psychiatry and Human Genetics.

"We can now say we have a tool by combining several genomic approaches to find these genes."

More information: Journal reference: www.pnas.org , Article number 201003027

Provided by University of Michigan Health System