Researchers identify mutation that causes short-sightedness and hearing loss

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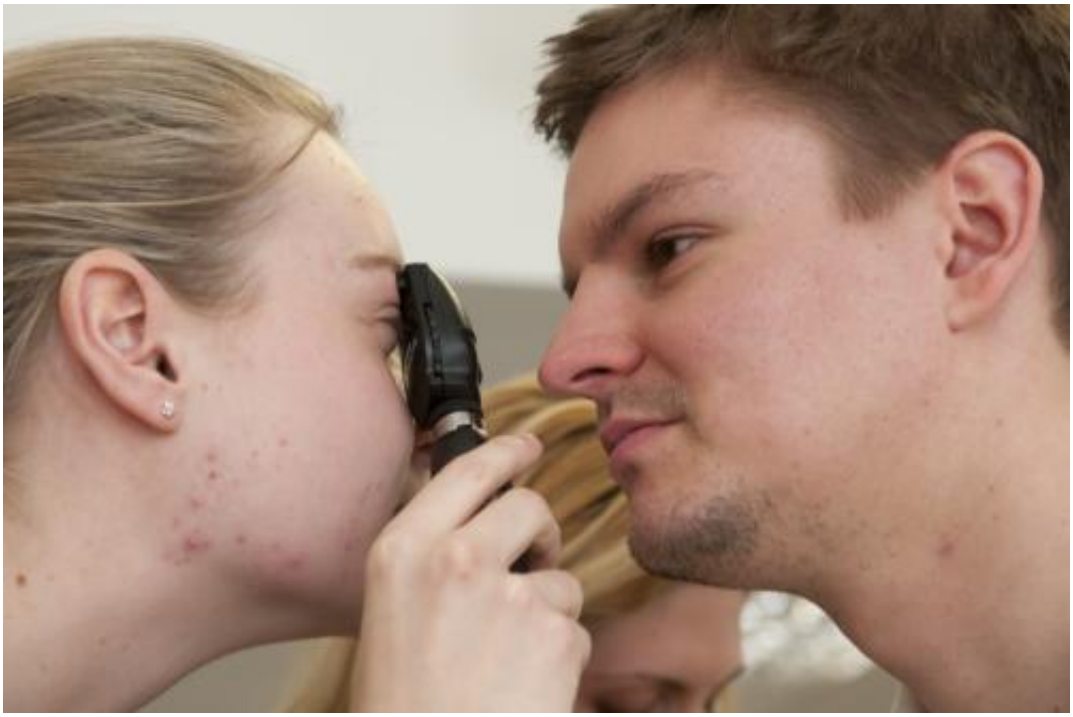

(Medical Xpress)—Researchers have identified a new disorder caused by a genetic mutation that leads to short sightedness and deafness. They say the new link between the two sensory problems could lead to better understanding of the disease mechanism of each.

The researchers – led by St George's, University of London and the

Miller School of Medicine in Miami – discovered that a mutation in the gene SLITRK6 causes a syndrome symptomised by myopia (short sightedness) and deafness.

Myopia – the most common eye disorder – and hearing loss have already been linked by more than 150 other genetic syndromes, but this is the first one not to involve abnormalities in other systems or other eye problems.

"We hope that identifying a genetic cause of these common sensory problems and a causative link between myopia and hearing loss will lead to better understanding of the mechanism of these disorders and eventually how to better treat them," said study co-lead author Dr Mustafa Tekin, associate professor at the Dr. John T. Macdonald Foundation Department of Human Genetics at the Miller School of Medicine.

Myopia affects nearly a billion people worldwide. Previous research has uncovered myopia's genetic component, but very few studies have demonstrated clear examples of when a single gene defect can cause myopia.

Co-lead author Professor Andrew Crosby, professor of human genetics at St George's, University of London, said: "By studying the genetic make-up of three families with multiple members affected by deafness and short-sightedness, we discovered that the SLITRK6 gene was mutated in all those who were affected by the condition."

The researchers also found that the mutation caused the same symptoms in mice.

The study has been published online in the *Journal of Clinical Investigation*. It was a collaborative effort with investigators from 11

universities and research institutions in the United States, the United Kingdom, Japan, Turkey, Greece, and Denmark.

The full paper can be found here.

Provided by St. George's University of London