

Gene mutation leads to impairment of 2 senses: Touch and hearing

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People with good hearing also have a keen sense of touch; people with impaired hearing generally have an impaired sense of touch. Extensive data supporting this hypothesis was presented by Dr. Henning Frenzel and Professor Gary R. Lewin of the Max Delbrück Center for Molecular Medicine (MDC) Berlin-Buch, Germany.

The two researchers showed that both senses – [hearing](#) and touch – have a common genetic basis. In patients with [Usher syndrome](#), a hereditary form of deafness accompanied by impaired vision, they discovered a gene mutation that is also causative for the patients' impaired touch sensitivity. The examination was preceded by various studies, including studies with healthy identical and non-identical human twins (*PLoS Biology*). In total, the researchers assessed sensory function in 518 volunteers.

In all vertebrates, and consequently also in humans, hearing and touch represent two distinct sensory systems that both rely on the transformation of mechanical force into electrical signals. When we hear, sound waves trigger vibrations that stimulate the hair-like nerve endings in the cochlea in the inner ear. These then transform the mechanical stimuli into electrical signals, which are transmitted to the brain via the auditory nerve. When we touch something a similar process takes place: The mechanical stimulus - sliding the fingers over a rough or smooth surface, the perception of vibrations - is taken up via sensors in the skin, converted into an electrical stimulus and transmitted to the brain.

Twin study with 100 pairs of twins

In recent years about 70 genes have been identified in humans, mutations in which trigger hearing loss or deafness. "Surprisingly, no genes have been found that negatively influence the [sense of touch](#)," Professor Lewin said. To see whether the sense of touch also has a hereditary component, the researchers first studied 100 pairs of twins - 66 pairs of monozygotic twins and 34 dizygotic pairs of twins. Monozygotic twins are genetically completely identical; dizygotic twins are genetically identical to 50 percent. The tests showed that the touch sensitivity of the subjects was determined to more than 50 percent by genes. Furthermore, hearing and touch tests showed that there is a correlation between the sense of hearing and touch.

The researchers therefore suspected that genes that influence the sense of hearing may also have an influence on the sense of touch. In a next step, they recruited test subjects at a school in Berlin for students with hearing impairments. There they assessed the touch sensitivity in a cohort of 39 young people who suffered from severe congenital hearing impairment. The researchers compared these findings with the data from their twin study and discovered that not all of the young people with hearing loss had impaired tactile acuity. "Strikingly, however, many of these young people did indeed have poor tactile acuity," Professor Lewin explained.

The researchers decided it would take too much time to analyze which of the approximately 70 genes that adversely affect the sense of hearing may also negatively affect the sense of touch. Therefore, the researchers focused specifically on patients with the Usher syndrome, a hereditary form of hearing impairment, in which the patients progressively become blind. Usher syndrome patients have varying degrees of hearing impairment, and the disease is genetically very well studied. There are nine known Usher genes carrying mutations which cause the disease.

The researchers examined one cohort of patients in a special consultation at the Charité - Universitätsmedizin Berlin for Usher patients from all over Germany. A second cohort was recruited at the university hospital La Fe in Valencia, Spain. The studies revealed that not all patients with Usher-syndrome have poor tactile acuity and touch sensitivity. The researchers showed that only patients with Usher syndrome who have a mutation in the gene USH2A have poor touch sensitivity. This mutation is also responsible for the impaired hearing of 19 patients. The 29 Usher-syndrome patients in whom the mutation could not be detected had a normal sense of touch. The researchers thus demonstrated that there is a common genetic basis for the sense of hearing and touch. They suspect that even more genes will be discovered in the future that influence both mechanosensory traits.

Women hear better than men and have a finer sense of touch

The researchers discovered another interesting detail during their five-year study. "When women complain that their men are not really listening to them, there is some truth in that," Professor Lewin said. "The studies with a total of 518 individuals including 295 women have actually shown that women hear better and they also have a finer [sense](#) of touch than men; in short woman hear and feel more than men!"

More information: *PLoS Biology* [doi:10.1371/journal.pbio.1001318](https://doi.org/10.1371/journal.pbio.1001318)

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