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The molecular basis of touch sensation: New function of a well-known gene identified

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A gene known to control lens development in mice and humans is also crucial for the development of neurons responsible for mechanosensory function, as neurobiologists of the Max Delbrück Center for Molecular Medicine (MDC) Berlin-Buch have now discovered.

They found that in mice in which they had removed the c-Maf gene in the nerve cells, touch sensation is impaired. This similarly applies to human carriers of a mutant c-Maf gene. People with such a mutation suffer already at a young age from cataracts, a clouding of the lens which typically affects the elderly. The patients, as demonstrated by Professor Carmen Birchmeier and Dr. Hagen Wende in collaboration with Professor Gary Lewin and Dr. Stefan Lechner, have difficulty holding objects such as a sheet of paper as a consequence of this mutation.

Professor Birchmeier, a developmental biologist, commented on the findings of her research group: "c-Maf is an important gene for the development of the peripheral nerve cells." The gene controls the development of neurons that detect touch, the mechanosensory neurons. Previously, c-Maf was known as a key regulator of lens development.

Furthermore, the gene is also active in the dorsal root ganglia, an aggregate of nerve cells next to the spinal cord in which the cell bodies of mechanosensory neurons are localized. The nerve cells form long axons, which terminate in the skin in touch corpuscles or at hair shafts. These axons detect mechanical stimuli, which in turn are converted into

electrical signals and transmitted to the brain. When you stroke your fingers over a surface, its structure triggers high-frequency vibrations in the finger, to which specific touch receptors, the Pacinian corpuscles, respond.

In mice with deactivated c-Maf gene only few Pacinian corpuscles are formed, and moreover these few are not intact. The mice are therefore unable to recognize high-frequency vibrations. The same is true for a Swiss family with an inherited mutant c-Maf gene. The consequence is that the affected patients develop cataracts at an early age and have an impaired sense of touch.

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