

## When poor communication pokes you in the eye

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The ocular lens belongs to the optical apparatus and focuses incidental beams of light onto the retina. Now, a research team led by Dr. Jochen Graw of the Institute of Developmental Genetics, of the Helmholtz Zentrum Munchen, has been able to decipher a genetic defect responsible for small eyes and an incomplete, clouded lens in the socalled Aey12 mouse mutants. These results lead to conclusions concerning cataracts in humans, because, in this case too, the lens loses its transparency.

The development of the eye in mammals (and this naturally includes humans) is an extraordinarily complex process beginning in an early embryonic phase. The same applies also to the formation in healthy eyes of elastic and transparent lenses, which focus light beams. With the aid of the ciliary muscles, the lens can change its degree of curvature and thus set itself on varied, far distant objects. As a result, a pin sharp image is created on the retina.

"As with humans, with mice too, the development of the lens starts with the formation of a spherical, hollow sac," Graw says. "That is the lens vesicle, the cover of which is surrounded by the lens epithelium, composed of a layer of cells. The vesicle is then filled in with fiber cells. In the following course of development, additional fibers originate in the equator of the lens. These scale up the diameter of the lens: a process that lasts a lifetime."

But not so with the Aey12 mouse mutants, which Graw's eye researchers



in the study group "Molecular Eye Development" have iknvestigated in detail. The animals of this line are distinguished by their unusually small eyes, a microphthalmia. Combined with cataracts, this dosorder is also known in humans and leads nearly always to blindness. With Aey12 mice, the early development of the eye lens is strongly affected.

As the scientists in the current issue of the well-known, American journal in the field of ophthalmology, "Investigative Ophthalmology & Visual Sciences", report, with the mutant mice, the growth of the fibers that fill up the body of the lenses, is completely blocked. "What remains is then a cloudy and functionless small lens sac," Oliver Puk adds, the first author of the study. "The animals thereby lose their sight almost completely."

As the scientists in the current study were able to show, the basis of the disease is a defect in a hitherto unknown gene. Genes are units of the hereditary DNA molecule that contains blueprints for proteins. Errors in the sequence of the gene building blocks can lead to proteins with limited or completely lost functions: and in this way, in the worst case, to serious diseases or development disorders.

The Neuherberg eye researchers gave the name Gjf1 to the gene responsible for the alterations of the Aey12 mutant mice. It is a member of the Connexin family. "The genes belonging to this group get the information for the construction of channel proteins, which build the cell to cell connections," Graw declares. "Such channels are of great importance for the interchange between cells: also, among other things, between the fiber cells of the developing eye lens itself."

The scientists now speculate that through the newly discovered mutation, the structure of the Gjf1 channel protein is changed, and in this way the formation of the channel is hindered. But through this, the communication between the developing lens fibers would break down.



Thus, it would be conceivable that signal molecules essential for the development of the lens are no longer exchanged, or only to a limited extent. In this scenario, faulty cell communication would be the cause of the development of the fibers stopping: and ultimately, of the cloudiness of the eye lens, which would otherwise be transparent.

This very phenomenon is also observed in cataracts in humans, a common disease, which appears mostly in the elderly: in Germany alone more than half a million operations are carried out annually, in which the cloudy and opaque lens is replaced with an implant. "Our results will surely also provide an insight into the origin of cataracts," Graw adds. "Moreover, so far, there has not yet been any mutation in humans equivalent to Gjf1 gene of mice. But this will now change soon, for sure."

Source: Helmholtz Zentrum München

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