New cataract gene discovered

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Same image as above, but simulated as seen through the lens of a cataract patient with a different clouding defect of the lens compared to picture 2. Glare, in addition to blurry images, disturb the visual perception. Photo: Stephan Labs

The international team of researchers was able to identify the location and defect in the coding region of the gene through analysis of genetic material (DNA) from members of a large Swiss family, the majority of whom suffered from autosomal dominant juvenile cataract. The corresponding protein belongs to a family of monocarboxylate transporters which move small molecules across cell mem-branes. Surprisingly, this genetic defect may also lead to the condition of renal glucosuria, a non-pathological kidney defect with elevated levels of glucose in the urine, but not in blood.

The search for the molecule that is carried by this new transporter across the membrane has now begun. The researchers suspect that this genetic

defect interferes with homeostasis in the lens as well as in the kidney.

Since the known environmental risk factors for age-related cataract point to physiological and oxidative damages accumulating over time within the lens, the researchers assume that defects in this newly discovered transporter may also be a cause of age-related cataract. Age-related cataract patients are now being screened to find mutations in this gene. Understanding the exact function of this transporter may open new venues for non-surgical treatment of cataract.

Cataract, the condition of altered refractive index in the lens of the eye, is the clouding of the eye's normally clear lens and results in blurry vision that frequently includes disturbing glare in the visual field. Successful treatment today involves surgical removal of the affected lens and replacement with an artificial one.

Age onset of cataract is the distinguishing characteristic between affliction in childhood or in later years. Whereas about 14% of blind children are affected, half of the adult population worldwide aged 60 and older suffers from age-related cataract. Various underlying defects cause its development. Research in molecular and genetic science has enabled identification of several genes that play an important role in structural or regulatory functions in lens cells. Although all cataract genes identified to date are known to affect children, genetic factors for age-related cataract continue to remain widely undiscovered.

Source: ETH Zurich