

Researchers identify gene that leads to myopia (nearsightedness)

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A Ben-Gurion University of the Negev research group led by Prof. Ohad Birk has identified a gene whose defect specifically causes myopia or nearsightedness.

In an article appearing online in the [American Journal of Human Genetics](#) today, Birk and his team reveal that a mutation in LEPREL1 has been shown to cause myopia.

"We are finally beginning to understand at a molecular level why [nearsightedness](#) occurs," Prof. Birk says. The discovery was a group effort at BGU's Morris Kahn Laboratory of Human Genetics at the National Institute for Biotechnology in the Negev and the Dayan [Clinical Genetics](#) Wing at Soroka University Medical Center.

Nearsightedness is the most common human eye disorder and is mostly a hereditary trait. Aside from being a significant public health concern, nearsightedness also leads to a higher incidence of other secondary eye disorders, such as [retinal detachment](#), macular degeneration, as well as early onset glaucoma and cataracts. Despite decades of intensive research, the specific genes whose defects lead to nearsightedness have remained elusive.

The defective gene was identified in a thorough study of severe early-onset myopia that is common in a specific Bedouin tribe in southern Israel. As part of the research and in collaboration with a Finnish group, studies in a model system using [insect cells](#) demonstrated that the

mutation is detrimental to the enzymatic activity of the gene.

The gene, LEPREL1, encodes an enzyme that is essential for the final modification of collagen in the eye. In the absence of the active form of this enzyme, aberrant collagen is formed, causing the human eyeball to be longer than normal. As a consequence, [light beams](#) entering the eyeball focus in front of the retina rather than on the retina itself and myopia emerges.

Future studies will determine whether LEPREL1 or its related genes play a significant role in the causation of myopia in the population at large as well.

Prof. Birk's group has thus far elucidated the molecular mechanisms leading to more than 15 human diseases, and the research findings are effectively implemented in massive screening tests and prevention programs.

Provided by American Associates, Ben-Gurion University of the Negev

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