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Gene leads to nearsightedness when kids read

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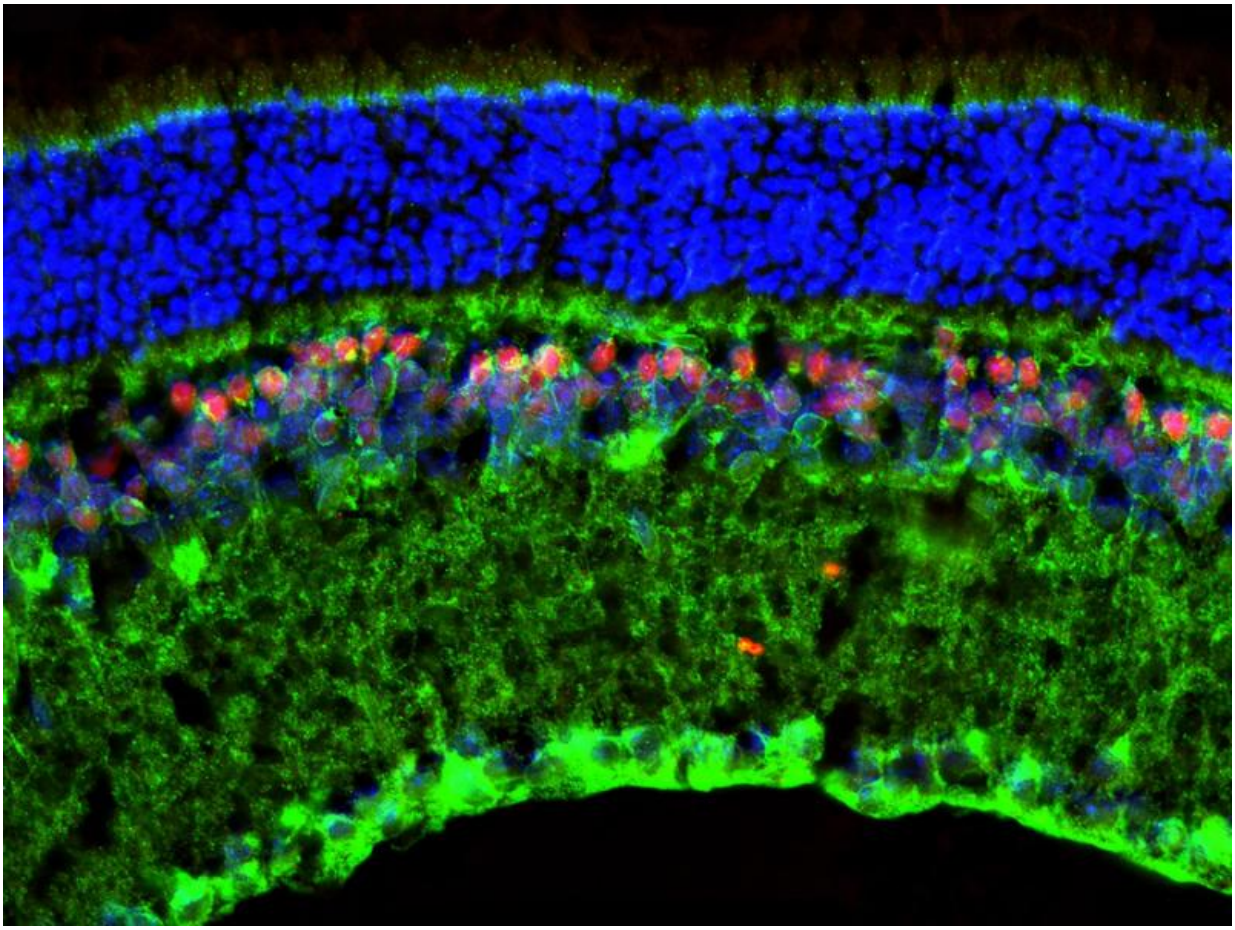

An antibody-stained cross section of a mouse retina. Credit: Andrei Tkachenko/Columbia University Medical Center

Vision researchers at Columbia University Medical Center have discovered a gene that causes myopia, but only in people who spend a lot

of time in childhood reading or doing other "nearwork."

Using a database of approximately 14,000 people, the researchers found that those with a certain variant of the gene - called APLP2 - were five times more likely to develop myopia in their teens if they had read an hour or more each day in their childhood. Those who carried the APLP2 risk variant but spent less time reading had no additional risk of developing myopia.

"We have known for decades that myopia is caused by genes and their interactions with environmental factors like reading and nearwork, but we have not had hard proof. This is the first known evidence of gene-environment interaction in myopia," says the study's lead investigator, Andrei Tkatchenko, MD, PhD, of CUMC. The research was published August 27 in *PLOS Genetics*.

Although it's not yet known how genetic variation at the APLP2 gene causes myopia, Dr. Tkatchenko and his colleagues think the risk variant may increase the amount of APLP2 protein produced in the eye, which in turn may cause the eye to undergo excessive elongation.

They found that mice exposed to a visual environment that mimics reading were less likely to develop myopia when little APLP2 protein was present in the eye.

"By reducing the level of APLP2 in the eye, you can reduce susceptibility to environmentally induced myopia. This gives us an opportunity to develop a therapy to prevent myopia in everyone, regardless of the APLP2 variant they carry," Dr. Tkatchenko says.

Developing such a therapy, however, could take years, as researchers don't yet know how APLP2 levels could be reduced in people. And the therapy would be most effective in young children, before the eye has

started to elongate and become myopic.

"Once the eye has elongated, you cannot shrink it, so we would need to identify kids with genetic risk factors as they enter school," Dr. Tkatchenko says. That's not yet possible because there are probably hundreds of genes that can cause myopia, and so far, only 25 candidates have been identified. The high-risk variant of APLP2 is relatively uncommon, occurring in about 1 percent of the population.

Though a drug or gene therapy to prevent myopia may be years away, Dr. Tkatchenko says spending time outdoors is the best way to reduce kids' risk of developing myopia.

"We pretty much know all the environmental risk factors: time spent reading increases the risk, while time spent outdoors reduces it," Dr. Tkatchenko says.

Myopia rates have shot up in recent years due to changes in these environmental factors. In the U.S., 44 percent of adults are now nearsighted, up from 25 percent 30 years ago. And in some parts of Asia, 80 percent of young adults are now myopic.

"The critical period for myopia development is during elementary and middle school, so when kids are in school, make sure they also spend at least two hours outdoors each day," Dr. Tkatchenko says.

Preventing myopia not only reduces the need for vision correction, but also prevents potentially blinding eye conditions later in life.

"People say, 'What's so terrible about myopia? You just have to get glasses,'" Dr. Tkatchenko says, "But myopia increases the risk of developing cataracts, glaucoma, and retinal detachment later in life. All of these can lead to blindness.

"Even in people with mild myopia there is a significant risk, but it's especially high in people who need five or more diopters of vision correction (about 20/400 vision). That's why it's important to study myopia."

More information: The paper is titled "APLP2 regulates refractive error and myopia development in mice and humans."

Provided by Columbia University Medical Center