# IMI – Myopia Genetics Report 2025

**Dr. Mats Voogelaar**
MD

Department of Ophthalmology, Erasmus MC University Medical Center, Rotterdam, the Netherlands
The Rotterdam Eye Hospital, Rotterdam, the Netherlands

Since the last IMI Genetics Report in 2019, research into the genetics of myopia has expanded rapidly. Thousands of genetic variants are now linked with refractive error, offering new insight into how the eye grows and why some people develop high myopia while others do not.

Recent studies confirm that myopia has a strong genetic component, but genes act together with environment and lifestyle. High levels of education, extensive near work, and limited outdoor time can amplify genetic risk. New "polygenic risk scores" show potential for predicting which children are most at risk when combined with environmental factors, although they are not yet ready for clinical use.

Large international studies have uncovered genes involved in the biological processes that shape the eye including genes for eye size regulation, retinal signaling, neurodevelopment, and extracellular matrix remodeling. Research into rare genetic variants has also identified mutations that overlap with retinal and connective-tissue disorders, highlighting when genetic testing can assist in identifying syndromic or high-risk cases of myopia.

Emerging work on epigenetics (how environment alters gene expression) is revealing how factors such as light exposure, circadian rhythm, and inflammation may "switch on" or "switch off" certain genes that influence ocular growth.

In summary, the field is moving from discovering genes to understanding how they function and interact with environment. This knowledge will eventually support personalized myopia prevention and treatment. Clinicians should remain aware that while genetic tools are not yet part of routine care, they are shaping the future direction of early-risk identification and targeted intervention.

**Key Takeaways for Clinicians**

**Genes and environment act together:** Genetic risk is amplified by environmental and lifestyle factors such as near work, education intensity, and limited outdoor time.

**Polygenic risk scores show promise** for early identification of children at higher risk, but they are not yet ready for clinical application.

**Rare variant testing can help** identify syndromic or severe myopia cases linked with retinal or connective tissue disorders.

**Epigenetic and molecular research** is revealing how factors like light exposure, sleep, and inflammation can influence eye growth through gene regulation.

**Clinical translation is coming:** Understanding genetic mechanisms will enable more personalized prevention and treatment strategies in the future.

## ACKNOWLEDGMENTS

A full list of the IMI taskforce members and the complete IMI white papers can be found at myopiainstitute.org. The publication and translation costs of the clinical summary was supported by donations from the BHVI, ZEISS, Essilorluxottica, CooperVision, Alcon, HOYA, Théa, and Oculus.

## REFERENCE

Mats Voogelaar, Milly S. Tedja, Jeremy A. Guggenheim, Seang-Mei Saw, Martha Tjon-Fo-Sang, David A. Mackey, Christopher J. Hammond, Caroline C. W. Klaver, Virginie J. M. Verhoeven; IMI—Myopia Genetics Report. *Invest. Ophthalmol. Vis. Sci.* 2025;66(13):22. https://doi.org/10.1167/iovs.66.13.22.

## CORRESPONDENCE

Brien Holden Vision Institute Ltd
Level 4, North Wing, Rupert Myers Building, Gate 14 Barker Street,
University of New South Wales, UNSW NSW 2052