

# Ophthalmology Update

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## In this issue

- 2 Global Track for Residents: International Education, Bidirectional Impact
- 3 Pediatric Myopia Clinic Prioritizes Early Intervention and Research
- 4 Valuable Addition to the Team Brings Shared Dedication to Patients, Passion for Research and Education



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## First FDA-Approved Retinal Gene Therapy Available at Mayo Clinic

Gene therapy has the potential to treat previously incurable complex diseases. The first prescription retinal gene therapy product is now available at Mayo Clinic. For eligible patients who have a mutation in both copies of the *RPE65* gene, voretigene neparvovec-rzyl is a one-time gene therapy that has the potential to restore the visual cycle and to improve functional vision.

With credentialed physicians to administer voretigene neparvovec-rzyl, Mayo Clinic now provides the first FDA-approved gene therapy for people with an inherited retinal disease (IRD). “This is a game changer, not only for ophthalmology but also for all of medicine,” says Brittini A. Scruggs, M.D., Ph.D., an ophthalmologist at Mayo Clinic in Rochester, Minnesota, who is trained in administering the therapy. “This treatment sets the stage for other gene therapies, and it’s an exciting innovation to be a part of.”

Voretigene neparvovec-rzyl is used to treat patients who have both of the following:

- Leber congenital amaurosis due to mutations in both copies of the *RPE65* gene.
- Sufficient viable cells remaining in the outer retina, as determined by an IRD specialist.

Within the United States, a phase 3 clinical trial, published in *The Lancet* in 2017, showed that voretigene neparvovec-rzyl improved functional

vision, which increased patients’ ability to perform daily life activities.

“We inject voretigene neparvovec-rzyl under the retina, making a localized retinal detachment. This adeno-associated virus solution infects cells — specifically, the retinal pigment epithelium — that are still present but are not functioning,” says Dr. Scruggs. “The virus works quickly, allowing the visual cycle to continue.”

*RPE65* is one of more than 300 genes that cause IRDs. Confirming a patient’s specific gene mutation or mutations with a genetic test is the first step to determining whether this gene therapy is a relevant treatment option.

“Determining the exact genetic cause of a patient’s disease allows our team to accurately make a diagnosis and counsel on prognosis, clinical trial opportunities and vision rehabilitation strategies,” says Raymond Iezzi Jr., M.D., an ophthalmologist at Mayo Clinic in Minnesota who treats patients with IRDs.

At Mayo Clinic, highly skilled medical geneticists and board-certified genetic counselors meet with patients of all ages. Lisa A. Schimmenti, M.D., a board-certified clinical geneticist at Mayo Clinic, along with Mayo Clinic genetic counselors Kahlen R. Darr, M.S., CGC, and Lea M. Coon, M.S., CGC, provide timely, detailed and compassionate care.

“In a shared decision-making model, genetic testing is offered to individuals with suspected IRDs,” says Dr. Schimmenti. “Our team gives individuals and families the knowledge needed to make informed decisions regarding genetic testing. Results are returned to individuals and families in the context of genetic counseling to understand the impact of their results and their eligibility for gene therapy.”

Using state-of-the-art imaging modalities and collaborative relationships across Mayo Clinic, the ophthalmology team has a breadth of resources available for each patient’s unique needs.

Currently, clinical trials are ongoing in the following research areas:

- Evaluating treatment for Stargardt disease.
- Retinal dystrophies associated with rare disease-causing genetic variants.
- Evaluating oral treatments in patients with retinitis pigmentosa.
- Evaluating treatments for male participants with X-linked retinitis pigmentosa.

“Some patients with IRDs might not have a clinical trial or treatment as an option yet, and our priority is to optimize our patients’ quality of vision and quality of life with the

resources that we have,” says Dr. Scruggs. “For patients who are candidates for voretigene neparvovec-rzyl, we’re excited to be at the forefront of providing cutting-edge gene therapy — providing our patients a more vibrant view of the world.”

#### FOR MORE INFORMATION

- Russell S, et al. Efficacy and safety of voretigene neparvovec (AAV2-hRPE65v2) in patients with RPE65-mediated inherited retinal dystrophy: A randomised, controlled, open-label, phase 3 trial. *The Lancet*. 390;10097:849.

## Global Track for Residents: International Education, Bidirectional Impact

Mayo Clinic’s Ophthalmology Residency training program in Rochester, Minnesota, aims to educate and inspire residents within a stimulating environment to learn the science and art of ophthalmology. This year, Mayo Clinic launched a new global ophthalmology and health equities track that thoughtfully prepares residents for local and international experiences.

The program provides flexible opportunities for residents to contribute in meaningful, bidirectional ways, which helps equip them with the necessary tools and experiences for providing equitable eye care.

“It’s a four-year track, and it has integrated lectures from myself and other faculty,” says Ashlie A. Bernhisel, M.D., an ophthalmologist at Mayo Clinic in Rochester, Minnesota, and an assistant professor of ophthalmology at Mayo Clinic Alix School of Medicine (Figure). Furthermore, Mayo Clinic launched a six-month-long health equities curriculum, including a book club, that those participating in the track will complete during their PGY3 year.

Combining didactics, online courses, surgical experience and fieldwork, residents cover a variety of topics, including social justice, advocacy, inclusion, epidemiology and social determinants of health. The track also includes volunteer requirements within the local area and an international experience.

“In addition to the typical benefits of an international trip such as experiencing a new culture or healthcare system, one very important element of this curriculum is bidirectionality,” says Dr. Bernhisel. “We want to ensure that residents are proportionately giving back to the community that they are learning from.”

This track is also part of Mayo Clinic Ophthalmology efforts to broaden involvement in global ophthalmology, which includes joining the Global Ophthalmology Consortium. The consortium hosts an annual Global Ophthalmology Summit, which brings together like-minded local and international ophthalmologists,



Figure. Dr. Bernhisel mentors a trainee during practice lab.

leaders from American Academy of Ophthalmology, and nongovernmental organizations to ease some of the burden of global and local blindness.

“Mayo Clinic’s unique patient-centered and highly integrated practice offers an ideal environment for clinical, surgical and academic training in ophthalmology,” says Andrea A. Tooley, M.D., an ophthalmologist at Mayo Clinic in Minnesota, and Ophthalmology Residency director at Mayo Clinic Alix School of Medicine. “In addition to the international opportunities through the global track, the clinical and academic environment encourages intellectual curiosity with an abundance of cutting-edge research opportunities and institutional resources.”

Whether residents aim to practice comprehensive or subspecialty ophthalmology, want to embark on a career in academic medicine, or plan to focus on global health initiatives, Mayo Clinic's dedicated faculty members mentor and encourage residents to maximize their potential and achieve individual goals.

"The global ophthalmology track helps residents understand health disparities — not only worldwide but also within the United States," says Dr. Bernhisel. "In addition to its academic benefits, it

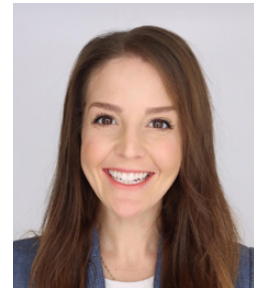
teaches important life skills and provides experiences that can contribute to a more compassionate, well-rounded resident and future ophthalmologist."

#### FOR MORE INFORMATION

Ophthalmology Residency (Minnesota). Mayo Clinic.



Ashlie A. Bernhisel, M.D.



Andrea A. Tooley, M.D.

## Pediatric Myopia Clinic Prioritizes Early Intervention and Research

Myopia is a common cause of correctable vision loss in children, and it is the most common source of distance vision impairment in the world. According to an article published in the December 2021 issue of *Beyoglu Eye Journal*, the global prevalence of myopia is predicted to increase from 27% of the world's population in 2010 to 52% by 2050.

"Because there is a strong genetic component to myopia, if either parent has a high degree of myopia, it can be beneficial to check whether a child is developing early or mild myopia," says Sean P. Templeman, O.D., a pediatric optometrist at Mayo Clinic in Rochester, Minnesota. "Early intervention and detection in children with myopia is the key to reducing the long-term impact on their ocular health and improving their future."

"Very high myopia may also be associated with other systemic, genetic or ocular comorbidities," says Eric J. Kim, M.D., a pediatric ophthalmologist at Mayo Clinic in Minnesota. "Early identification of myopia can allow for an opportunity for interventions and treatments that can help slow the rate of progression."

Mayo Clinic's pediatric myopia clinic offers coordinated care within a multidisciplinary team that has extensive experience managing the care of children with myopia. Healthcare professionals in specialties including pediatric optometry, pediatric ophthalmology and pediatric retina work closely together to come up with a comprehensive, individualized treatment plan.

"One of the benefits of our pediatric clinic is that we have access to specialized testing such as optical biometry to measure the axial length of the eye to monitor for progression," says Dr. Kim. "We're also able to work closely with our clinical genomics colleagues and refer for genetic testing when indicated."

#### Current therapies that show the most promise include:

- Low-dose atropine.
- Increased time outside.
- Multifocal contact lenses.
- Orthokeratology.

Researchers and clinical practitioners continue to seek more-effective approaches to slow the progression of nearsightedness in children and adolescents. "In our clinic, we have the opportunity to stay connected with current research that is going on with myopia," says Dr. Templeman. "We are constantly gathering data, which aids in the development of protocols and treatment strategies. We have the unique ability to be at the forefront and stay up to date on relevant changes in the field."

Future research studies may include the use of multifocal spectacles, which have shown promise in slowing the rate of myopia progression but are still pending approval for use in the U.S.

Prevention is key in slowing the progression of myopia in children, and it helps address an increasing global incidence. "Even if children have passed

their vision screenings at school, and nothing is apparently wrong, sometimes vision issues can slip through the cracks," says Dr. Templeman. "Something we've learned is that we need to be as preventive as possible and follow children a bit more closely than we have historically."

#### FOR MORE INFORMATION

Nouraeinejad A. More than fifty percent of the world population will be myopic by 2050. *Beyoglu Eye Journal*. 2021;6:255.



Sean P. Templeman, O.D.



Eric J. Kim, M.D.



# Valuable Addition to the Team Brings Shared Dedication to Patients, Passion for Research and Education

Patients faced with life-changing illnesses find a path to healing at Mayo Clinic. Mayo's multidisciplinary, integrated team is committed to solving the most serious or complex challenges through clinical practice, education and research.

Greg J. Griepentrog, M.D., has joined Ophthalmology at Mayo Clinic in Rochester, Minnesota, bringing his own unique experience and knowledge. "Direct care for patients has been my greatest joy in this profession, and I'm looking forward to continuing the core value of my Mayo Clinic training — 'the needs of the patient come first,'" says Dr. Griepentrog. "A distinct advantage in joining Mayo Clinic is the existing world-class research infrastructure. I'm also looking forward to the opportunity to continue pediatric oculoplastic research in the realms of craniofacial malformations, the treatment of vascular anomalies and childhood ptosis."

"We are pleased to welcome Dr. Griepentrog back to Mayo Clinic," says Sophie J. Bakri, M.D., chair of Ophthalmology at Mayo Clinic in Minnesota and professor of ophthalmology at Mayo Clinic College of Medicine and Science. "His dedication to patients, along with his passion for research and education, aligns with our values and mission."

"I have been privileged to participate in the training of numerous residents, along with 14 American Society of Ophthalmic Plastic and Reconstructive Surgery fellows," says Dr. Griepentrog. "As much as I have helped novice ophthalmic surgeons develop their clinical and surgical skills, mentees have taught me a great deal more, and their enthusiasm continually recharges my own desire to learn and improve."

## ABOUT DR. GRIEPENTROG

Dr. Griepentrog completed medical school at the University of Wisconsin School of Medicine and Public Health in Madison, Wisconsin. He completed a transitional year internship at Aurora Health Care, St. Luke's Hospital, in Milwaukee, Wisconsin, and then completed his residency in ophthalmology at Mayo Clinic in Rochester, Minnesota. He then completed a fellowship in ophthalmic plastic and

reconstructive surgery at the University of Wisconsin-Madison.

Prior to joining Mayo Clinic, he was an associate professor of ophthalmology for the Department of Ophthalmology and Visual Sciences at the Medical College of Wisconsin Eye Institute in Milwaukee, and he was a senior medical director for surgical and medical specialties within the Froedtert Health system.

## SPECIALTY AREAS

"My clinical practice spans a broad range of upper facial, lacrimal and orbital surgery, both adult and pediatric," says Dr. Griepentrog. "While the bulk of the practice is centered on functional ocular adnexal surgery and post-Mohs reconstruction, I have greatly enjoyed the opportunity to collaborate with a variety of medical and surgical specialists to manage complex pediatric care, orbital malignancy and reconstruction following trauma."

## RESEARCH HIGHLIGHTS

- Incidence and demographics of childhood ptosis.
- Anatomical position of hyaluronic acid gel following injection to the infraorbital hollows.
- Assault-related orbital trauma at an urban Level I Trauma Center: Racial segregation and other neighborhood-level social determinants.
- Prevalence and clinical features of orbital vascular anomalies in children.
- Eyelid blastomycosis with concurrent medial malleolus osteolysis in an urban region.



Greg J. Griepentrog, M.D.



Sophie J. Bakri, M.D.



## CONTACT US

Mayo Clinic welcomes inquiries and referrals, and a request to a specific physician is not required to refer a patient.

**Phoenix/Scottsdale, Arizona**  
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**Rochester, Minnesota**  
800-533-1564



## RESOURCES

[mayoclinic.org/medical-professionals](https://www.mayoclinic.org/medical-professionals)

Clinical trials, CME, Grand Rounds, scientific videos and online referrals



## EDUCATION OPPORTUNITIES

Visit [ce.mayo.edu/ophthalmology](https://ce.mayo.edu/ophthalmology)  
Call 800-323-2688  
or email [cme@mayo.edu](mailto:cme@mayo.edu)

## Ophthalmology Update

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### Cover Image

Human multicolored iris of the eye animation concept. Credit: CG Alex

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