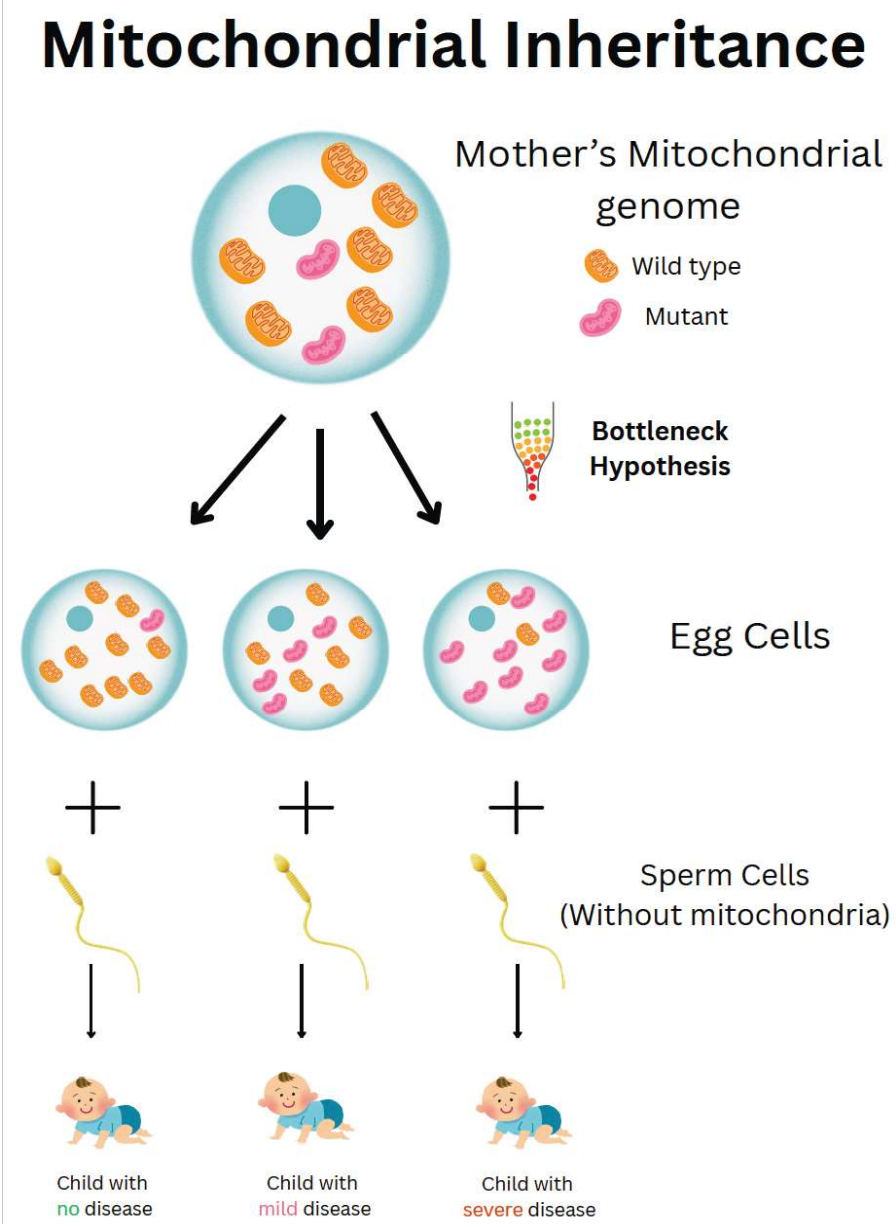
Global Eye Genetics Consortium

Newsletter #10

THE GENETICS 101



Mitochondria, the "powerhouses" of the cell, have their own DNA (mtDNA) and follow a unique inheritance pattern—strictly maternal. Unlike nuclear genes, which come from both parents, mtDNA is passed down only through the egg, meaning all children of an affected mother can inherit the mutation, while fathers do not transmit it further. This maternal lineage of transmission underlies several multisystem disorders, particularly those affecting high-energy tissues such as the brain, muscles, and eyes.

A key concept in mitochondrial genetics is the bottleneck hypothesis. During egg development, only a small, random subset of mitochondria is passed to the next generation. This results in considerable variability in the proportion of mutant versus normal mtDNA—called heteroplasmy—among siblings. The clinical expression of mitochondrial disease depends on this proportion: higher levels of mutant mtDNA generally cause more severe disease, while lower levels may result in milder or even subclinical presentations. This threshold effect explains the wide spectrum of symptoms seen in families carrying the same mitochondrial mutation.

Ocular involvement is a hallmark of many mitochondrial disorders due to the high energy demands of retinal and optic nerve tissues. Leber hereditary optic neuropathy (LHON), one of the most well-known conditions, causes acute or subacute painless vision loss due to degeneration of retinal ganglion cells. Other mitochondrial disorders such as chronic progressive external ophthalmoplegia (CPEO) present with ptosis and ophthalmoplegia, while some syndromic forms can also involve pigmentary retinopathy. Recognizing these ocular features is critical, as they often represent the first clue to an underlying mitochondrial disorder.

Ophthalmic Genetics News

Around the World

Raising hopes for future human eye regeneration?

In a study funded by the Howard Hughes Medical Institute, the Society for Developmental Biology, the American Association for Anatomy and the Stowers Institute for Medical Research and published August 6,2025 in Nature Communications, a team of researchers led by University of California-Davis biologist, Alice Accorsi, has shown that apple snail and human eyes share many anatomical and genetic features. Most of the work was done prior to Accorsi joining UC Davis in 2024. Using a combination of dissections, microscopy and genomic analysis, Accorsi's team was able to demonstrate these similarities. In addition to being easy to grow in the lab, apple snails have "camera-type" eyes -- the same type as humans that are known for producing particularly high-resolution images. Unlike humans, however, the snail can regenerate a missing or damaged eye. The assistant professor of molecular and cellular biology at UC Davis is studying how these snails regrow their eyes -- with the goal of eventually helping to restore vision in people with eye injuries. In the study, the researchers mapped the regeneration process and used CRISPR-Cas9 to identify genes, including pax6, as essential to eye development, raising hopes for future human vision restoration. According to Accorsi, "Apple snails are extraordinary organisms. They provide a unique opportunity to study regeneration of complex sensory organs. Before this, we were missing a system for studying full eye regeneration. If we find a set of genes that are important for eye regeneration, and these genes are also present in vertebrates, in theory we could activate them to enable eye regeneration in humans"

As a first test, the team used CRISPR/Cas9 to mutate a gene called pax6 in snail embryos. Pax6 is known to control the development and organization of brain and eye in humans, mice and fruit flies. Like humans, snails have two copies of each gene - one from each parent. The researchers showed that when apple snails have two non-functional versions of pax6, they develop without eyes, which shows that pax6 is also essential for initial eye development in apple snails. The team is working on the next step, which will be to test whether pax6 also plays a role in eye regeneration. To determine this, researchers will need to mutate or turn off pax6 in adult snails and then test their regenerative ability. In addition, Accorsi is investigating other eye-related genes, including genes that encode specific parts of the eye, like the lens or retina, and genes that control pax6.

Sources:

1. University of California - Davis. "Can humans regrow eyes? These snails already do." ScienceDaily. ScienceDaily, 6 August 2025. <www.sciencedaily.com/releases/2025/08/250806094112.htm>.
2. Alice Accorsi, Brenda Pardo, Eric Ross, Timothy J. Corbin, Melainia McClain, Kyle Weaver, Kym Delventhal, Asmita Gattamraju, Jason A. Morrison, Mary Cathleen McKinney, Sean A. McKinney, Alejandro Sánchez Alvarado. A genetically tractable non-vertebrate system to study complete camera-type eye regeneration. Nature Communications, 2025; 16 (1) DOI: 10.1038/s41467-025-61681-6

Eye2Gene predicts causes of genetic eye diseases

An international study involving researchers from University College London and the NIHR Moorfields Biomedical Research Centre has developed Eye2Gene, an AI system that determines the genetic causes of inherited retinal diseases (IRDs) using standard eye scans. Eye2Gene, a deep learning algorithm detailed in Nature Machine Intelligence, represents the largest AI study of IRDs to date. It was trained on over 58,000 retinal scans from patients at centers in London, Oxford, Liverpool, Tokyo, Bonn and São Paulo. This tool is more accurate than human experts, which helps speed up diagnoses and prioritize genetic testing. It can also identify ideal candidates for clinical trials and new gene therapies.

Inherited retinal diseases (IRDs) are uncommon genetic conditions, impacting roughly 1 in 3,000 individuals. These disorders stem from mutations in over 270 different genes and cause the retina to degenerate. This degeneration can lead to vision problems from birth or a gradual decline in sight as a person ages. For patients to get the newest treatments for inherited retinal diseases (IRD), it is crucial to get a genetic diagnosis as soon as possible, especially since more of these targeted therapies are becoming available and approved.

Associate Professor Nikolas Pontikos, lead investigator for Eye2Gene at the UCL Institute of Ophthalmology, explains that even with better genetic testing, inherited retinal diseases (IRDs) are frequently misdiagnosed or missed entirely. His team hopes to shorten the diagnostic journey for patients worldwide by integrating specialized knowledge—derived from carefully analyzed and genetically confirmed retinal scans—into their AI. This approach has significantly advanced the use of machine learning in the field of genetic eye diseases.

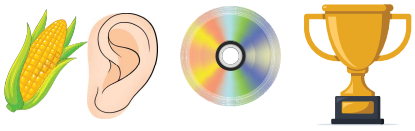
Sources:
1) UCL Translational Research Office(TRO).AI tool pinpoints genetic eye diseases from scans in international study led by UCL researchers. 19 June,2025. https://www.ucl.ac.uk/translational-research/news/2025/jun/ AI -tool- pinpoints- genetic -eye- diseases -scans -international -study led-UCL- researchers | Translational Research Office (TRO) - UCL – University College London. Accessed August 15,2025
2) Pontikos, N., Woof, W.A., Lin, S. et al. Next-generation phenotyping of inherited retinal diseases from multimodal imaging with Eye2Gene. Nat Mach Intell 7, 967–978 (2025). https://doi.org/10.1038/s42256-025-01040-8

TRIVIA

Guess the name of the condition depicted by the combination of the pictures

1) 

2) 

3) 

4) A  A

KNOW OUR MEMBERS

Dr. Andrea Vincent is a highly accomplished Ophthalmologist Clinician-Scientist from New Zealand, specializing in inherited retinal diseases, ocular genetics, and pediatric ophthalmology. She holds several qualifications, including MBChB, MD, and FRANZCO, which underscore her expertise and commitment to the field.

Following her ophthalmology training, Dr. Vincent enhanced her skills at the Royal Victorian Eye and Ear Hospital in Melbourne, Australia, and served as the inaugural Ocular Genetic Fellow at The Hospital for Sick Kids in Toronto, Canada, under the mentorship of Dr. Elise Héon and Dr. Alex Levin. Upon returning to New Zealand, she established a Genetic Eye Service at the Auckland District Health Board and created the Genetic Eye Disease Investigation Unit at the University of Auckland's Department of Ophthalmology. This unit is dedicated to identifying the genetic causes of inherited eye diseases and characterizing genotype-phenotype correlations.

Dr. Vincent's research encompasses various conditions, including blepharophimosis-ptosis-epicanthus inversus syndrome, corneal dystrophies, keratoconus, glaucoma, and inherited retinal and optic nerve diseases. Her groundbreaking work has resulted in the establishment of a Database of Inherited Retinal and Optic Nerve Disease in New Zealand, which now includes over 1,500 participants and family members. In addition to her research, Dr. Vincent practices as a pediatric ophthalmologist and has a keen interest in non-accidental injury in infants. She also possesses expertise in medical retina, particularly in electrophysiology testing.

Dr. Vincent currently holds the position of Associate Professor at the Faculty of Medical and Health Sciences at the University of Auckland. She is also a Consultant Ophthalmologist at the Auckland District Health Board and serves as the Director at Retina Specialists in Parnell.

Dr. Vincent is available for media inquiries and continues to make significant contributions to the field of ophthalmology, particularly in New Zealand and beyond.

Dr. Manuel Benjamin Ibanez, is one of the pioneers in the field of Ocular Genetics in the Philippines. Dr. Ibanez completed his residency in ophthalmology at Makati Medical Center and pursued fellowships in Pediatric Ophthalmology and Strabismus at Kims Eye Hospital in Seoul and Hospital Sant Joan De Deu in Barcelona. He further specialized with an Ocular Genetics fellowship under Dr. Alex Levin at Wills Eye Hospital.

As one of only two clinically trained Ocular Geneticists in the Philippines, Dr. Ibanez is a pioneer in establishing the first dedicated ocular genetics clinic at the DOH Eye Center, the designated national referral center for eye care. He also serves as the section head of Pediatric Ophthalmology and Ocular Genetics at Makati Medical Center.

Dr. Ibanez's expertise and leadership are instrumental in advancing the understanding and treatment of ocular genetic conditions in the country. We look forward to seeing the continued impact of his work in improving eye care for patients across the Philippines.

Adj Asst Prof Ngo Wei Kiong serves as Consultant and Deputy Head of Research at the National Healthcare Group Eye Institute (NHGEI), and is based at Tan Tock Seng Hospital.

Adj Asst Prof Ngo graduated from the Yong Loo Lin School of Medicine, National University of Singapore in 2009, and obtained his Master of Medicine (Ophthalmology) in 2016. During his ophthalmology residency, he served as Chief Resident and completed the Singapore Chief Residency Programme. He was awarded the Ministry of Health's Health Manpower Development Plan (HMDP) scholarship, under which he pursued fellowships in medical retina and inherited retinal diseases with Dr Richard Spaide at the Vitreous Retina Macula Consultants of New York, and Dr Stephen Tsang at Columbia University. Adj Asst Prof Ngo is currently a Clinical Senior Lecturer with the Yong Loo Lin School of Medicine, NUS, and an Adjunct Assistant Professor with the Lee Kong Chian School of Medicine, Nanyang Technological University.

Adj Asst Prof Ngo is actively engaged in retina research, with particular interests in age-related macular degeneration, diabetic retinopathy, and inherited retinal diseases. He has authored and co-authored over 50 publications, including book chapters and articles in peer-reviewed journals. His work has been recognized with multiple research awards both locally and internationally. He currently holds research funding exceeding half a million dollars from agencies such as the National Medical Research Council (NMRC).

Adj Asst Prof Ngo Wei Kiong's clinical interests span medical retina, inherited retinal diseases, cataract surgery, and general ophthalmology.

Dr. Sandeep Grover is a Professor of Ophthalmology at the University of Florida, where he practices at UF Health Jacksonville. A highly respected medical retina specialist, he has developed deep expertise in both common and complex retinal diseases, including diabetic retinopathy, age-related macular degeneration, and diverse retinal vascular occlusions. His outstanding contributions to the field have been recognized with the prestigious "Achievement Award" and "Senior Achievement Award" from the American Academy of Ophthalmology (AAO), where he also serves as a Council member.Beyond clinical excellence, Dr. Grover has assumed major academic and leadership responsibilities at the University of Florida. He currently serves as Medical Director of the Ophthalmology Trauma Service, Research Director, and Associate Chair of the Department of Ophthalmology. In these roles, he has been instrumental in shaping departmental strategy, strengthening research programs, and providing advanced training opportunities for future ophthalmologists.

Teaching and mentorship have been consistent hallmarks of Dr. Grover's career. His dedication to medical education has been recognized multiple times, including being honored as "Exemplary Teacher Award" on several occasions at the University of Florida. In 2025, he received the University of Florida College of Medicine's 20-Year Service Pin, a testament to his enduring commitment to the institution. He continues to play an integral role in course instruction, particularly in ophthalmology, significantly influencing the professional growth of medical students and residents.

Dr. Grover's research interests span a broad spectrum of retinal diseases, with a special focus on inherited retinal disorders such as retinitis pigmentosa, Stargardt disease, and cone-rod dystrophy.

His work also addresses retinal detachment, macular degeneration, and diabetic retinopathy. Driven by a commitment to improving patient outcomes, his research seeks to deepen the understanding of disease mechanisms while advancing innovative therapeutic approaches.

At UF Jacksonville, Dr. Grover leads an active research team and currently serves as the Principal Investigator for 18 ongoing clinical trials. These include studies on diabetic retinopathy, macular degeneration, and both X-linked and autosomal dominant retinitis pigmentosa, as well as Stargardt disease. Through this body of work, he is advancing the frontiers of knowledge in retinal disease while bridging the gap between clinical practice and translational research.

In sum, Dr. Grover's career exemplifies a rare combination of clinical expertise, academic leadership, dedicated teaching, and impactful research, making him a highly influential figure in ophthalmology and a leader in the advancement of retinal care.

Dr. Se Joon Woo is an ophthalmologist and a leading expert in retinal diseases and vitreoretinal surgery. He currently serves as a Professor of Ophthalmology at Seoul National University Bundang Hospital, where he also holds the positions of Chief of the Department of Ophthalmology and Director of the Medical Device Research and Development Center.

Dr. Woo specializes in the diagnosis and treatment of a broad spectrum of retinal and ocular conditions, including age-related macular degeneration, diabetic retinopathy, inherited retinal diseases, uveitis, retinal vascular diseases, and retinal detachment. He is highly skilled in advanced surgical procedures such as vitreoretinal surgery and cataract surgery. He completed his MD from Seoul National University College of Medicine in 1999, followed by a BsD and PhD in Ophthalmology from Seoul National University in 2008 and 2012 respectively. Dr. Woo has been part of the faculty at Seoul National University Bundang Hospital since 2008 and assumed the role of department chief in 2024.

Dr. Woo is the leader of the KEGC (Korean Eye Gene Consortium) and the vice-president of the EAIRDs (East Asian Inherited Retinal Disease Society). He is an active member of several national and international societies, including the Korean Retina Society, Korean Ophthalmology Society, Korean Uveitis Society, Korean Society of Clinical Electrophysiology and Vision, the American Academy of Ophthalmology (AAO), the Retina Society, EURETINA, the American Society of Retina Specialists (ASRS), ISCEV, and the Asia-Pacific Vitreo-retina Society (APVRS) and Academy of Ophthalmology (APAO). Through his academic leadership, clinical practice, and research contributions, Dr. Woo continues to advance the field of ophthalmology both in Korea and globally.

Answers to Trivia

1) Cherry Red Spot
2) Crabbe's Disease
3) Corneal Dystrophy
4) Achromatopsia

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