



Stanford | Stanford Center for
OPHTHALMOLOGY | Optic Disc Drusen
Byers Eye Institute

4th Annual Stanford Center for Optic Disc Drusen Conference Agenda

Monday, April 17, 2023 • 8AM-4PM PDT

**In-person: Conference Room D110, Stanford Research Park,
3145 Porter Dr., Palo Alto, CA 94304**

Virtual: via Zoom webinar

Visit the ODD website:
med.stanford.edu/optic-disc-drusen

Visit our department website:
med.stanford.edu/ophthalmology

EVENT ORGANIZERS



Joyce Liao, MD, PhD
Director, Center for Optic Disc Drusen at Stanford
Professor of Ophthalmology and of Neurology



Jeffrey Goldberg, MD, PhD
Blumenkranz Smead Professor and Chair of Ophthalmology



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Time	Topic	Speakers/Moderators
7:30am – 7:55am PDT	In-person: Stanford Research Park Conference Center. 3145 Porter Drive, Palo Alto, CA, 94304 Conference room D110 Parking is free or walk from ophthalmology labs Check in at registration desk and enjoy breakfast Virtual: Zoom webinar opens at 7:45am PDT	Any issues, please email us at OpticDiscDrusen@stanford.edu
8:00am – 8:02am	Welcoming remarks	Jeffrey Goldberg, MD, PhD
8:02am – 8:15am	Introduction to Stanford Center for Optic Disc Drusen, updates, and our research goals	Joyce Liao, MD, PhD
8:15am	Introduction of Session 1 Moderators	Jeffrey Goldberg, MD, PhD
8:15am – 9:40am (85 min)	Session 1 Optic disc drusen clinical presentation, imaging, and disease pathogenesis	Moderators: Heather Moss, MD, PhD Shannon Beres, MD
8:15am – 8:30am (15 min)	Clinical presentation and multimodal imaging of optic disc drusen in adults	Joyce Liao, MD, PhD
8:30am – 8:45am (15 min)	Clinical presentation and imaging of optic disc drusen in children	Shannon Beres, MD
8:45am – 9:00am (15 min)	Big data and electronic health records for ophthalmology	Sophia Wang, MD, MS
9:00am – 9:15am (15 min)	Quantitative analysis of optic disc drusen using metabolic imaging	Sangeethabalasri Pugazhendhi, MBBS
9:15am – 9:30am (15 min)	Are optic disc drusen kidney stones of the optic nerve?	Alan Pao, MD
9:30am – 9:40am (10 min)	Discussion	All
9:40am – 10:00am (20 min)	Break (food and drinks in café area)	



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Time	Topic	Speakers/Moderators
10:00am	Introduction of Session 2 Moderators	Joyce Liao, MD, PhD
10:00am – 10:45am (45 min)	Session 2: Genomic analysis of optic disc drusen	Moderators: Vinit Mahajan, MD, PhD Joyce Liao, MD, PhD
10:00am – 10:05am (5 min)	Genomic analysis of autosomal dominant optic disc drusen: patient recruitment	Joyce Liao, MD, PhD
10:05am – 10:15am (10 min)	Whole exome sequencing vs whole genome sequencing	Karanvir Kaushal, PhD
10:15am – 10:25am (10 min)	Whole genome sequencing: data analysis	Meng Wang, MS
10:25am – 10:35am (10 min)	What's next after whole genome sequencing?	Karanvir Kaushal, PhD
10:35am – 10:45am (10 min)	Discussion	All
10:45am	Introduction of Session 3 Moderators	David Myung, MD, PhD
10:45am – 12:00pm (60 min)	Session 3: The anterior optic nerve: biology, mitochondria, and ultrastructural study	Moderators: Patrick Yu-Wai-Man, MD, PhD David Myung, MD, PhD
10:45am – 11:00am (15 min)	Anterior optic nerve and mitochondria disease	Patrick Yu-Wai-Man, MD, PhD
11:00am – 11:15am (15 min)	Anterior optic nerve and glia: syndromic optic disc drusen and ciliopathies	Yang Sun, MD, PhD
11:15am – 11:30am (15 min)	Human retinal ganglion cell model of ectopic calcification	Hirenkumar Patel, PhD
11:30am – 11:45pm (15 min)	Ultrastructural analysis of human mitochondria in eye-brain diseases	Wah Chiu, PhD
11:45am – 12:00pm	Discussion	All



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12:15pm – 12:55pm (40 min)	Patient Q&A Session, Open Discussion/Lunch In-person: Stanford Research Park Conference Center D110 Virtual: via Zoom webinar	Moderators: Shannon Beres, MD Joyce Liao, MD, PhD Heather Moss, MD, PhD
1:00pm	Introduction of Session 4 Moderators	Mary Elizabeth Hartnett, MD
1:00pm – 2:30pm (75 min)	Session 4: Novel therapies and clinical trial for optic disc drusen	Moderators: Jeffrey Goldberg, MD, PhD Mary Elizabeth Hartnett, MD
1:00pm – 1:15pm (15 min)	Mitochondrial optic neuropathies: possible role in optic nerve head drusen (virtual)	Alfredo Sadun, MD, PhD
1:15pm – 1:30pm (15 min)	Preclinical translational studies of optic neuropathies and drug screening	Yang Hu, MD, PhD
1:30pm – 1:55pm (25 min)	Treatment of mitochondrial optic neuropathies	Patrick Yu-Wai-Man, MD, PhD
1:55pm – 2:15pm (20 min)	Optic nerve regeneration and other treatment considerations	Jeffrey Goldberg, MD, PhD
2:15pm – 2:30pm (15 min)	Discussion	All
2:30pm	Wrap up	Joyce Liao, MD, PhD Jeffrey Goldberg, MD, PhD
2:45pm – 4:00pm (75 min)	Reception and Poster Session <i>(hors d'oeuvre appetizers and open bar)</i> Poster presenters: Please stand by your poster at the first 30 min (2:45-3:15pm) of the poster session. The attendees are excited to talk to the people who did the work. Note, the organizers will come by with gift cards for the top 3 poster prizes during the poster session In-person: please pick up your souvenirs at the registration desk during the poster session or when you leave. Virtual: We hope you can join us in person in the future!	All



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Shannon Beres, MD

Clinical Associate Professor of Neurology and of Ophthalmology
Stanford University

Dr. Shannon Beres is a pediatric and adult neuro-ophthalmologist. She is currently working on projects using hand-held optical coherence tomography in infants and young children. She contributes to multi-site studies in pediatric optic neuritis, cranial neuropathies, pseudotumor cerebri syndrome, and optic pathway gliomas.



Wah Chiu, PhD

Wallenberg-Bienenstock Professor of Bioengineering
Director of Cryogenic electron microscopy and Bioimaging
Stanford Linear Accelerator Center (SLAC) National Accelerator
Laboratory, Stanford University

Dr. Chiu's work has made multiple transformational contributions in developing single particle CryoEM for the structural determination of molecular machines at atomic resolution. His recent research is devoted to characterization of subcellular structure signatures of cells normal and pathological states with and without therapeutic intervention. Dr. Chiu is an elected member of the National Academy of Sciences.



Jeffrey Goldberg, MD, PhD

Blumenkranz Smead Professor and Chair of Ophthalmology
Stanford University

Dr. Goldberg's clinical effort is focused on medical and surgical intervention for treatment of glaucoma and other optic nerve diseases. His research is directed at neuroprotection and regeneration of retinal ganglion cells and the optic nerve. His laboratory is developing novel stem cell and nanotherapeutics approaches for treatment of vision loss.



Mary Elizabeth Hartnett, MD

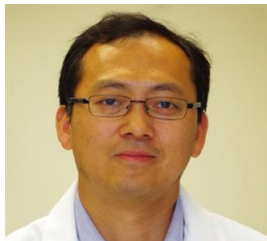
Michael F. Marmor Professor in Retinal Science and Diseases,
Professor of Ophthalmology
Stanford University

Dr. Hartnett is the Director of Pediatric Retina at Stanford and principal investigator of a retinal angiogenesis laboratory, in which she studies causes and treatments for diseases including retinopathy of prematurity and age-related macular degeneration.



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Yang Hu, MD, PhD

Associate Professor of Ophthalmology
Stanford University

Dr. Yang Hu is working on neuroprotection and axon regeneration for optic neuropathies. His lab focuses on the molecular mechanisms responsible for axon injury-induced neuronal degeneration with the goal of building on this understanding to develop effective combined strategies to promote neuroprotection, regeneration and functional recovery.



Karanvir Kaushal, PhD

Postdoctoral Scholar, Liao Lab
Stanford University

Karanvir Kaushal earned his PhD from Postgraduate Institute of Medical Education and Research (PGIMER), India, and joined Liao Lab as a postdoctoral scholar in Jan 2022. His research focuses on multi-omic profiling of patients with optic disc drusen and ischemic optic neuropathy. His goal is to identify novel biomolecules and pathways in order to improve the diagnosis and treatment of patients with optic neuropathies.



Joyce Liao, MD, PhD

Director, Center for Optic Disc Drusen at Stanford
Professor of Ophthalmology and of Neurology
Stanford University

Dr. Liao is dedicated to making basic discoveries and improving clinical care and treatment of patients with eye-brain issues like optic disc drusen and ischemic optic neuropathy. She runs a basic science laboratory focused on optic nerve diseases and retinal ganglion cells and a human clinical research group investigating risk factors for disease and testing highly promising novel therapies. Her goal is to improve the diagnosis and treatment of patients suffering from vision loss due to these optic neuropathies.



Vinit Mahajan, MD, PhD

Professor of Ophthalmology and Vice Chair for Research
Stanford University

Dr. Mahajan directs a NIH-funded Omics Laboratory that uses high-throughput methods in genomics, proteomics, and phenomics to identify molecules involved in vitreoretinal diseases. Mahajan and his team performed the first CRISPR gene editing therapy for eye disease in human stem cells. Using translational proteomics, Mahajan's multidisciplinary team is developing new precision health approaches using molecular biomarkers to diagnose retinal disease, select personalized therapies, and decode the anatomic structures of the human eye.



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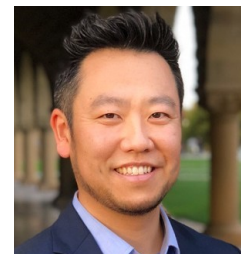
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Heather Moss, MD, PhD

Associate Professor of Ophthalmology and of Neurology
Stanford University

Dr. Moss is a physician-scientist who provides expert neuro-ophthalmic clinical care. She directs an innovative clinical-research program in biomarker discovery. Dr. Moss's clinical and research interests focus on idiopathic intracranial hypertension, optic neuritis, other optic neuropathies, and multiple sclerosis. She is also an expert in telemedicine.



David Myung, MD, PhD

Associate Professor of Ophthalmology and, by courtesy, of
Chemical Engineering, Stanford University

Dr. Myung is an ophthalmologist and attending physician specializing in cataract and corneal surgery and external diseases of the eye. He is the Director of the Ophthalmic Innovation Program. Myung leads a research group that takes an interdisciplinary approach toward fostering regeneration of ocular tissues. He is also Director of the Stanford Teleophthalmology Automated Testing and Universal Screening Program, which is pushing the boundaries of telemedicine and artificial intelligence (AI) to improve eye care worldwide.



Alan Chun-Yao Pao, MD

Associate Professor of Medicine/Nephrology
Stanford University

Dr. Pao aims to advance the understanding and treatment of kidney stone disease. He is Director of the Kidney Stone Center at Stanford and VA Palo Alto Health Care System. He is broadly interested in how the kidneys control salt, water, and electrolyte homeostasis in the body. His research group uses cultured kidney cells, transgenic mice, human plasma/urine samples, and electronic health record data to study the pathogenesis of kidney stone disease. His therapeutic focus is on the development of small molecule compounds that can be used to reduce stone risk or disrupt stone formation.



Hirenkumar Patel, PhD

Postdoctoral Scholar, Liao Lab
Stanford University

Dr. Patel received his PhD in 2018 from Mumbai University, India. He is a postdoctoral Scholar in the Liao Lab. He works in the area of stem cells and their differentiation to retinal ganglion cells (RGC). Presently, he is working on the derivation of fibroblast cell lines from patients' skin biopsies and evaluation of their basic cellular function and metabolic activity under different calcification conditions.



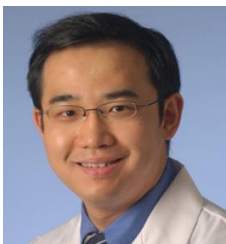
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Sangeethabalasri Pugazhendhi, MBBS
Postdoctoral Scholar, Liao Lab
Stanford University

Dr. Pugazhendhi received her medical degree from PSG Institute of Medical Sciences & Research, India, in 2019. She is currently a postdoctoral scholar in the Liao Lab and leads the effort in clinical research studies of optic disc drusen and other optic neuropathies. Her research focus is on the use of innovative ophthalmic imaging as biomarkers for predicting vision loss in neuro-ophthalmic patients.



Yang Sun, MD, PhD

Professor of Ophthalmology and Vice Chair of Academic Affairs
Stanford University

Dr. Yang Sun is a clinician-scientist with a clinical specialty in glaucoma. He practices at both Byers Eye Institute at Stanford as well as at VA Palo Alto Health Care System. He is an NIH and VA-funded investigator with a research focus on the role of inositol metabolism in eye development and disease. The current research interests in his lab include the elucidation of the mechanisms in cilia-mediated signaling in developmental and degenerative eye conditions.



Alfredo A. Sadun, MD, PhD

Flora L. Thornton Endowed Chair and Chief of Ophthalmology,
Doheny, Vice-Chair of Ophthalmology
University of California Los Angeles

Dr. Sadun is an international authority in neuro-ophthalmology, especially in diseases of the optic nerve. He maintains an active laboratory with research centered on the clinical, psychophysical and laboratory studies of diseases of the optic nerve, diseases of mitochondrial impairment, optic nerve regeneration, and neuro-protection. Dr. Sadun has made pivotal discoveries on Leber's hereditary optic neuropathy, and he was the first to identify an optic neuropathy associated with Alzheimer's disease.



Patrick Yu-Wai-Man, MD, PhD

Chair and Professor of Ophthalmology
University of Cambridge, England

Professor Yu-Wai-Man is an academic neuro-ophthalmologist with a major research interest in mitochondrial genetics and inherited eye diseases. He is Chair of Ophthalmology at the University of Cambridge, affiliated with the Cambridge Centre for Brain Repair. His research group is exploring the disease mechanisms leading to progressive retinal ganglion cell loss in inherited optic neuropathies by using a combination of patient tissues, induced pluripotent stem cells and animal models.



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Meng Wang, MS

Bioinformatics Programmer
Baylor College of Medicine

Ms. Wang is a Bioinformatics Programmer at Professor Rui Chen Lab in the Department of Molecular & Human Genetics at Baylor College of Medicine. Her research in the Chen Lab focuses on human genetics research of Inherited Retinal Diseases (IRDs) using whole genome sequencing (WGS), cell-level gene isoform detection using ONT long read single cell RNA sequencing, and detection and validation of structural variations. Also working to develop multiple databases and user interfaces which can be applied to lab daily use.



Sophia Wang, MD, MS

Assistant Professor of Ophthalmology
Stanford University

Dr. Wang's research focuses on using and integrating a wide variety of "big data" sources spanning both structured and unstructured forms, including national datasets, insurance claims data, surgical video, and electronic health records, all to investigate and improve ophthalmic outcomes. Her work to develop artificial intelligence algorithms to predict ophthalmology patients' outcomes using natural language processing for electronic health records data is supported by several sources, including a Research to Prevent Blindness Career Development Award, a K23 Career Development Award from the National Eye Institute, and the Young Clinician Scientist Award from the American Glaucoma Society.