QUANTITATIVE AND SYSTEMS BIOLOGY COLLOQUIUM:
Inositol signaling and eye diseases

Yang Sun
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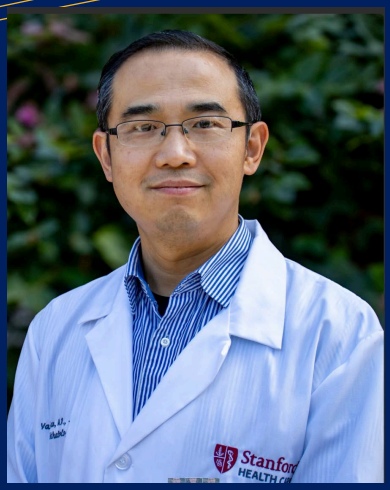

Date:

3/13/2025

Time:

10:30 AM - 11:45 AM

Location:

COB 1 114

About the Speaker:

Dr. Yang Sun is a clinician-scientist in ophthalmology and a Professor and Vice Chair, Academic Affairs, of Ophthalmology at Stanford University and Byers Eye Institute. He is the Laurie Kraus Lacob Faculty Scholar at Stanford Child Health Research Institute. Dr. Sun received his BA in Biophysics from Johns Hopkins University, followed by a MD.PhD. degree from Washington University School of Medicine. He completed Ophthalmology residency at Stanford University and a prestigious Heed fellowship at University of Michigan, Ann Arbor. Dr. Sun was an Assistant Professor at Indiana University, where he was promoted to an Associate Professor with tenure, before returning to Stanford University. He has been continuously funded by National Eye Institute and Veterans Administration. Dr. Sun's research in glaucoma has also been funded by American Glaucoma Society, Lowe Syndrome Association, Knights Templar Eye Foundation, and Matilda Ziegler Foundation. Dr. Sun holds several U.S. patents on novel regulators of eye pressure and is the primary investigator on a number of glaucoma clinical trials. He is a member of Stanford BioX faculty and he was elected as a member of American Society of Clinical Investigators.

Abstract:

Hosphoinositides are a group of signaling lipids that play crucial roles in various cellular processes, including membrane trafficking, cytoskeletal organization, and signal transduction. Dysregulation of the enzymes that regulate these molecules can cause human disease such as Lowe syndrome and Joubert syndrome. Lowe syndrome, a rare X-linked multisystem disorder presenting with major abnormalities in the eyes, kidneys, and central nervous system, is caused by mutations in OCRL gene. Encoding an inositol polyphosphate 5-phosphatase, OCRL catalyzes the hydrolysis of PI(4,5)P2 into PI4P. There are no effective targeted treatments for Lowe syndrome. I will discuss the recent advances in Lowe syndrome and our understanding of OCRL in cell biology. Another cilia related condition is Joubert syndrome, a rare genetic disorder characterized by the malformation of the cerebellar vermis, presenting a distinctive "molar tooth sign" on brain imaging. This condition leads to a range of symptoms, including hypotonia, ataxia, developmental delays, and abnormalities in eye movements and breathing patterns . We recently discovered a novel link between cilia and mitochondria and I will discuss the basic and translational findings related to Joubert syndrome.

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