Ophthalmic Genetics Study Club 2024
Hybrid Meeting
Lurie Children's Hospital, Chicago, USA

Moderators: Elias I. Traboulsi & Virginia M. Utz

Schedule	Title	Presenter	Institution	Email Address
7:00	**Breakfast/Catching up**			
725	Welcome	Elias I. Traboulsi, MD, MEd Virginia Utz, MD	Cleveland Clinic Cole Eye Institute	traboue@ccf.org
730 	MORC2-related Neurodevelopmental and Neuropathy Spectrum Disorder and Klinefelter syndrome: Genetic and Ocular Findings	Rebecca Procopio, MS, CGC	Wills Eye Hospital	rebecca.procopio34@gmail.com
745	**RPE65 Update:** -Safety and Durability of Voretigene Neparvovec for Biallelic *RPE65*-Mediated Inherited Retinal Disease: Phase 3 Results at 8 and 9 Years -Genetic epidemiology of *RPE65*-mediated inherited retinal disease in the United States	David L. Rousso, Ph.D Aditya Saxena, Ph.D	Spark Therapeutics, Inc.	david.rousso@sparktx.com Aditya.Saxena@sparktx.com
0805	The Long and Short of it: Expanding the Phenotype and	Janice Lasky Zeid, M.D.	Ann & Robert H. Lurie Children's Hospital of	jzeid@luriechildrens.org

	Ocular Manifestations of Neurofibromatosis 1 Microdeletion Syndrome		Chicago/Northwestern University	
0820	Pars Plana Vitrectomy in Congenital X-Linked Retinoschisis	Nieraj Jain, M.D.	Department of Ophthalmology, Emory University School of Medicine	nieraj.jain@emory.edu
0835	Could Estrogen reduce the Split?	Kimerly Stepien, MD	University of WI-Madision Department of Ophthalmology and Visual Sciences	kstepien@wisc.edu
0850	Ophthalmological Manifestations in a Pediatric Population with Stickler Syndrome Types I and II	Andy Drackley, MS, CGC	Lurie Children's Hospital of Chicago	adrackley@luriechildrens.org.
0905	Macular dystrophy, what is the missing puzzle piece?	Fyqah Almahmoudi, MD	King Fahd Armed Forces Hospital, Saudi Arabia	fyqah112@gmail.com
Coffee Break 0920-0940AM				
0940	Genetic Basis for Bilateral Optic Atrophy, Bilateral Medial Rectus Hypoplasia, Oligophrenia and Stroke in 31 y/o Male	Patrick J. Droste MS, MD	Pediatric Ophthalmology PC	drdroste@comcast.net
0955	Phenotypic variability of MAF-Associated Congenital Cataracts: Isolated or Syndromic?	Jennifer Rossen, MD	Lurie Children's Hospital of Chicago	jerossen@luriechildrens.org
1010 ★	Compound Heterozygous Nonsense and Frameshift Mutations in Exon 43 of EYS Causing Reduced Penetrance	Megan Soucy, MS, CGC	Columbia University - Department of Ophthalmology	mes2350@cumc.columbia.edu

1025	Genotypic and Phenotypic Expansion of ADAMTS18-Related Ocular Pathology	Ari August, MD	Wills Eye Hospital	aaugust@willseye.org
1040	Genetic variants in patients with bilateral congenital cataracts in a large cohort from North India	Savleen Kaur, MD, DNB, FICO, FAICO	Post Graduate Institute of Medical Education and Research, India	mailsavleen@gmail.com
1055	Lessons learned from the ClinGen Gene and Variant Curation Expert Panels	Lev Prasov, MD, PhD	University of Michigan	lprasov@umich.edu
1110	Macular scars from prior ocular toxoplasmosis infection?	Arif Khan, MD	Cleveland Clinic Abu Dhabi	aokhan@yahoo.com
1125	Compound heterozygous *CYP1B1* variants in juvenile open angle glaucoma (JOAG)	Brenda L. Bohnsack, MD, PhD	Ann & Robert H. Lurie Children's Hospital of Chicago	bbohnsack@luriechildrens.org
1140	*INTS11* neurodevelopmental disorder with electronegative ERG	Monique Leys, MD	WVU Eye Institute	monique.leys@hsc.wvu.edu; leysm26505@gmail.com
Lunch Break* 1155 pm – 1240 pm				
1240	Tribute to Frank Judisch, MD	Arlene Drack, MD	University of Iowa	arlene-drack@uiowa.edu
1250	**Distinguished Special Lecture** **Johane Robitaille, MD** **"Saturday Night FEVR and Three Decades of Research in Maritime Canada"**			

1340	Thirty-year follow-up of surgical repair of congenital ectopia lentis including DNA analysis	Irene H Maumenee, MD	CUIMC/Edward S. Harkness Eye Institute	ihm2115@cumc.columbia.edu
1355	Homozygous deletion in P-CARE results in a maculopathy and cone-rod dystrophy	Ian MacDonald, MSc, MD, CM	University of Montreal	macdonal@ualberta.ca
1410	Pigmentary retinopathy and optic disc swelling in siblings with *ABCA4* mutations	Scott Brodie, MD, PhD	Columbia University Vagelos College of Physicians and Surgeons	seb2207@cuimc.columbia.edu
1425	Association of colobomas with variants in the Rho family of small GTPases and effector genes	Diana Brightman, PhD, MS	Cincinnati Children's Hospital Medical Center	Diana.brightman@cchmc.org
1440	Amblyopia Not Responding to Treatment	Alina Dumitrescu, MD	University of Iowa	alina-dumitrescu@uiowa.edu
1455	Microcephaly and chorioretinopathy in a child born to an Ophthalmology resident: perspectives as a doctor and mother in training	Emily Levine, MD	Dartmouth Hitchcock Medical Center	emily.s.levine@hitchcock.org
Coffee Break 1510-1530				
1530	Results of the first patient-customized intravitreally-administered antisense oligonucleotide (ASO) for a patient with retinal dystrophy caused by an intronic pseudogene.	Emily McCourt, MD	University of Colorado	emily.mccourt@gmail.com

Field Code Changed

1545	A case of fish-eye disease caused by novel variants in LCAT	Matt Benson, MD	University of Alberta	mbenson@ualberta.ca
1600	Deep phenotyping reveals novel ophthalmic findings in Smith-Lemli-Optiz Syndrome	Kati Veres, MD	National Eye Institute/NIH	kati.veres@nih.gov
1615	Case Presentation	Noor Ghali, M.S.	Case Western Reserve University SOM	nxg342@case.edu
1630	Ellipsoid zone optical gap and cone-rod dystrophy secondary to ABCA4 p.Pro143Leu variant	Kirill Zaslavsky	Massachusetts Eye and Ear Infirmary	kzaslavsky@meei.harvard.edu
1645	Ultracompact hand-held swept source optical coherence tomography (SS-HH-OCT) as a novel diagnostic modality for early-onset retinal dystrophies (EORDS)	Ramiro Maldonado, MD	Duke Eye Center	Ramiro.maldonado@duke.edu
17:00	Presentation	Dan Chung, DO	SparingVision	Daniel.chung@sparinvision.com
If time permits	Case Presentation	Scott Brodie, MD, PhD	Columbia University Vagelos College of Physicians and Surgeons	seb2207@cuimc.columbia.edu
	Concluding Remarks / Adjourn			
	OGSC Business Meeting			