Ophthalmic Genetics Study Club 2025
Hybrid Meeting
Orlando, FL

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	101	When One Diagnosis Becomes Two: Lessons from the Diagnostic Workup of Albinism in a Single Family.	Hanna Scanga	University of Pittsburgh	Hanna.scanga@chp.edu
745	102	Custom genetic panel detects second mutation in patient with Leber Congenital Amaurosis – A case report	Valencia Potter	University of Iowa	Valencia-potter@uiowa.edu
0805	103	Exploring the role of *CHRDL1* mutations in X-linked megalocornea: A comprehensive review and case study	Christopher Davey Sponsor: G. Zaidman	New York Medical College School of Medicine	cdavey@student.nymc.edu
0820	104	Unusual etiology of syndromic retinitis pigmentosa and sensorineural hearing loss – case report of thiamine-responsive megaloblastic anemia syndrome	Jesus Vidaurri-Martinez	Duke University School of Medicine	jesus.vidaurri-martinez@duke.edu

0835	105	Mosaic Art	Kimberly Stepien	University of Wisconsin - Madison	kstepien@wisc.edu
0850	106	Association of *MT-CYB* with syndromic retinitis pigmentosa	Aliaa H. Abdelhakim	Columbia University Irving Medical Center	Aha2133@cumc.columbia.edu
0905	107	Genotype and phenotype characteristics of X-linked retinoschisis cases and long-term follow-up results	Nesllihan Sinim Kahraman Alex Levin	Flaum Eye Institute	neslihansinim@gmail.com
Coffee Break 0920-0940AM					
0940	108	Long-term safety and efficacy data of lenadogene nolparvovec gene therapy for LHON	Patrick Yu-Wai-Man	University of Cambridge	py237@cam.ac.uk
0955	109	A case of optic atrophy associated with a novel heterozygous variant in *UCHL1*	Brandon C. Huynh and Matthew D. Benson	University of Alberta, Edmonton, Canada	mbenson@ualberta.ca
1010	110	Gain of function in *BEST1*: Photoreceptor changes and myopia in ADVIRC	Roselind Ni	Wills Eye Hospital	niroselind@gmail.com
1025	112	*SPG11*-related ocular patterns: Kjellin syndrome as a possible developmental marker	Giacomo Bacci	Meyer Childrne's Hospital, Florence, Italy	giacomo.bacci@meyer.it
1040	113	A family with *CACNA2D4*-related retinopathy	Ari August Sponsor: Jose Pulido	Wills Eye Hospital	aaugust@willseye.org
1055	114	Profound Effect of Light on Schisis in X-linked Retinoschisis	Arlene Drack	University of Iowa	arlene-drack@uiowa.edu
1110	115	An additional retinal finding in pediatric retinal dystrophy	Arif Khan, MD	Cleveland Clinic Abu Dhabi	aokhan@yahoo.com

1125	116	Is it this? Or that? Or something else – An unknown case presentation	Gregg Lueder	Washington University	lueder@wustl.edu
1140	117	*CNGA3*-related retinopathy: an unexpected phenotype	Monique Leys, MD	WVU Eye Institute	monique.leys@hsc.wvu.edu; leysm26505@gmail.com
Lunch Break* 1155 pm – 1240 pm					
1240 	118	Genomic Convergence: Deciphering the Coincidence of Oculocutaneous Albinism and Autosomal Dominant Retinitis Pigmentosa	Jordan Lehman	Duke Eye Center	jordan.lehman@duke.edu
1255	119	 **Distinguished Special Lecture** **Lev Prasov MD, PhD** **"Uncovering Multisystem Syndromes Through Ocular Genetics"**			
1340	120	Compound heterozygosity versus homozygosity of autosomal recessive mutations	Irene H Maumenee, MD	CUIMC/Edward S. Harkness Eye Institute	ihm2115@cumc.columbia.edu
1355	121	Ocular manifestations of biallelic *WDR91* variants	Margaret Reynolds	Washington University	margaret.reynolds@wustl.edu
1410	122	Red-fish retinopathy – an EYS phenotype	Scott Brodie, MD, PhD	Columbia University Vagelos College of Physicians and Surgeons	sbrodie52@gmail.com
1425	123	Infant with newly described ocular findings of neuro-oculo-cardio-genitourinary syndrome	Gabriela Rodriguez Sponsor: G Zaidman	New York Medical College	grodrigu@student.touro.edu

1440	124	Cornelia de Lange Syndrome: Solving the Mystery with Episignature	Virginia M. Utz	Cincinnati Children's Hospital	Virginia.utz@cchmc.org
1455	125	Subretinal gene therapy laru-zova for X-linked retinitis pigmentosa (XLRP): Results from Early Phase Trials	Dan Chung	Beacon Therapeutics	dchung@beacondx.com
Coffee Break 1510-1530					
1530	126	Low population penetrance of variants associated with inherited retinal degenerations. **Or** Genetic spectrum of negative electroretinograms in a predominantly pediatric cohort of 177 patients.	Kirill Zaslavsky	Harvard Medical School	
1545	127	Mitochondrial Retinal Dystrophy Associated with m.3243A>G Mutation in the *MT-TL1* Gene	Diti Patel	University of Florida, Jacksonville,	Diti.Patel@jax.ufl.edu
1600	128	New autosomal dominant variant in *RPE65* in a Brazilian family	Mariana Matioli Da Palma	Federal University of São Paulo	marimatioli@yahoo.com.br
1615	129	The Fantasia of Rings: Etiology of Concentric Macular Rings and OCT Corrugations in Oculocutaneous Albinism with Foveal Hypoplasia	Ari August Sponsor: J Pulido	Wills Eye Hospital	aaugust@willseye.org
1630	130	A family with ?FEVR	Meghan DeBenedictis	Cole Eye Institute	Marinom2@ccf.org
1645	131	Biallelic occult macular dystrophy	Noor Ghali	Case Western Reserve University	nghali@umich.edu

			Sponsor: A Khan		
		Concluding Remarks / Adjourn			
		OGSC Business Meeting			