Ophthalmic Genetics Study Club

Hybrid Meeting

Thursday, September 29, 2022

Hancock Building close walk from
Children's Hospital
875 N. Michigan Avenue, 14th Floor, Room 108

Moderators: Elias I. Traboulsi & Virginia M. Utz

This year's meeting is dedicated to Dr. A. Linn Murphree

	Title	Presenter	Institution
8:00	**BREAKFAST & CATCHING UP**		
8:30 – 12:00 - **SESSION 1**		**MODERATOR: ELIAS TRABOULSI**	
8:30 **Virtual Presentation**	Expanding the spectrum of Oculocutaneous Albinism: does isolated foveal hypoplasia really exists?	**Giacomo Bacci, MD, PhD** *Staff*	Meyer Children's Hospital, Fi, Italy
8:45	The Management of Inherited Retinal Disease by an Ophthalmic Genetic Service: An Analysis of Provider Referral Patterns, Delays in Diagnosis and the Diagnostic Yield of Genetic Testing	**Natario Couser, MD, MS** *Staff*	Virginia Commonwealth University
9:00	A woman referred for evaluation of Stargardt disease	**Arif Khan, MD**	Cleveland Clinic AbuDhabi
9:15	The problem of monocular cases of inherited retinal disease	**Scott E. Brodie, MD**	NYU Langone Health
9:30	Right disease, wrong gene or wrong gene, right disease	**Alex V. Levin, MD, MHSc, FAAP, FAAO, FRCSC**	Flaum Eye Institute
9:45	Use of VEP in Inherited Retinal Degenerations	**Arlene V. Drack, MD**	University of Iowa
10:00 **Virtual Presentation**	Multimodal imaging characteristics and correlation with genetic	**Savleen Kaur, MD**	Department Of Ophthalmology, Post Graduate

	profile of achromatopsia in North Indian population.		Institute of Medical Education and Research (PGIMER) Chandigarh, India
10:15 **Virtual Presentation**	Registry of 85 informative south Asian pedigrees with primary concomitant strabismus: A meta-analysis	**Zia Chaudhuri, MD, PhD**	LHMC & Associated Hospitals, PGIMER & Dr. RML Hospital, New Delhi, India
10:30	Repeatability of a Truncated Visual Field Protocol in Pediatric Inherited Retina Disease Patients	**Ramiro Maldonado, MD**	Duke University
10:45	Case Presentation	**Meghan DeBenedictis, CGC, MEd**	Cole Eye Institute
11:00	**In Honor of A. Linn Murphree**	**Janey Wiggs, MD, PhD**	MEEI, Harvard Medical School
1:00 – 4:45 - **SESSION 2**		**MODERATOR: GINNY M. UTZ**	
1:00	Case Presentation	**Rachel Huckfeldt, MD**	MEEI, Harvard Medical School
1:15 **Virtual Presentation**	Case Presentation	**Rebecca Procopio, MD**	Wills Eye Hospital
1:30 **Virtual Presentation**	Case Presentation	**Sarah Chorfi, MD**	MEEI, Harvard Medical School

1:45	Gene independent approaches to cone photoreceptor survival and rescue in rod cone dystrophies	**Daniel Chung, DO**	Sparing Vision
2:00	Case Presentation	**Jose Pulido, MD**	Wills Eye Hospital
2:15 **Virtual Presentation**	Off-Label Voretigene Neparvovec-rzyl for Dominant *RPE65* Dystrophy	**Robert Sisk, MD**	University of Cincinnatti
2:30	*RPE65* Gene Therapy in Saudi Arabia	**Selwa Al-Hazzaa, MD**	Al-Faisal University
2:45 **Virtual Presentation**	Variant Classification and Reporting Differences Can Lead to Variable Test Yield on Retinal Dystrophy Panels	**Diana Brightman, Ph.D, MS, CGC**	Cincinnati Children's Hospital
3:00	Case Presentation	**Jennifer Galvin, MD**	Yale New Haven Hospital
3:15 **Virtual Presentation**	Genetic Causes for IRDS in Ethiopean Jews	**Miriam Ehrenberg, MD**	Schneider Children's Medical Center, Israel
3:30	Brothers with optic neuropathy: LHON or not?	**Lev Prasov, MD**	University of Michigan
3:45 **Virtual Presentation**	Isolated *ectopia lentis* in Polynesian patients	**Sarah Hull, MD**	Auckland Eye New Zealand
4:00	A "Spleenided Case!"	**Kimberly Stepien. MD**	University of Wisconsin

4:15	*PIEZO1* and *PIEZO2* Pathogenic Variants Identified in Primary Congenital Glaucoma	**Terri Young, MD, MBA**	University of Wisconsin – Madison, School of Medicine and Public Health
4:30	Case Presentation	**Virginia Utz, MD**	Cincinnati Children's Hospital
4:45 – 5:00 - **BUSINESS MEETING**			