

2018 Ophthalmic Genetics Study Club



Friday, October 26, 2018
7:30 am to 5:00 pm

Kindly hosted by Dr. Marilyn Mets & the Division of Ophthalmology

Ann & Robert H. Lurie Children's Hospital
225 East Chicago Avenue
11-142 (11th floor)



Time	Title	Presenter
Morning Session		
7:30 to 7:50	Breakfast	
7:50 to 7:55	Welcome	Marilyn Mets
7:55 to 8:10	Case Presentation	Virginia Miraldi Utz
8:15 to 8:30	Case Presentation	Melanie Schmitt
8:35 to 8:50	Posterior Column Ataxia with Retinitis Pigmentosa	Rachel Sullivan
8:55 to 9:10	A Family with Retinal Degeneration and Kidney Transplants	Aliaa Abdelhakim
9:15 to 9:30	Expanding the Ocular Phenotype of Saul Wilson Syndrome	Wadih Zein
9:35 to 9:50	Case Presentation	Meghan Debenedictis
9:55 to 10:10	Retinal Splits in 3 Sibs	Arif Khan
10:10 to 10:25	Break	
10:25 to 10:40	The Future of Ophthalmic Genetics	Irene Maumenee
10:45 to 11:00	Interesting Genetic Testing Finds (2 cases)	Lauren Moissiy
11:05 to 11:20	Follow-up on Familial Ocular Motor Apraxia	Steven Archer
11:25 to 11:40	COMMAD: A New Syndrome by Biallelic Mutations in MITF	Brian Brooks

11:45 to 12:00	Vision Loss in Neurofibromatosis I	Alina Dumitrescu
12:05 to 12:20	A Family with Congenital Retinal Non-Attachment	Selwa A. F. Al-Hazzaa/Basamat Almoallem

12:20 to 1:05	Lunch Break	
Afternoon Session		
1:05 to 1:20	Case Presentation	Razek Coussa
1:25 to 1:40	Seeing Stars: The Case for Whole Exome Sequencing in Optic Atrophy	Elizabeth Kellom
1:45 to 2:00	An Unusual Presentation of Williams' Syndrome	Richard Lewis
2:05 to 2:20	Retinal Dystrophy	Monique Leys
2:25 to 2:40	Interesting Case of RPE65 LCA	Arlene Drack
2:40 to 2:55	Afternoon break	
2:55 to 3:10	Blurring the Difference of Congenital and Primary Open Angle Glaucoma	David Mackey
3:15 to 3:30	Case Presentation	Shreya Prabhu
3:35 to 3:50	Primary Congenital Glaucoma- New TEK Mutations	Terri Young
3:55 to 4:10	17 Week Gestational Age Retina in Norrie Disease	Shizuo Mukai
4:15 to 4:30	Mystery Family (Almost) Revealed: A Tale of (At Least!) Two Stories	Alex Iannaccone
4:35 to 4:45	Child with AAMR and Review of the Literature	Jennifer Galvin
4:45 to 5:00	Electronegative ERGs at The University of Iowa	Wanda Pfeifer
5:00 pm	Adjournment	

Case Presentations Pending if Time Allows

Scott Brodie
Elias Traboulsi
Arif Khan
Wadih Zein

Atypical Cone-Rod Dystrophy
Case Presentation
My Worst Nightmare
“Atypical” Usher Syndrome

Scott Brodie
Scott Brodie
Scott Brodie

Severe Cone Dystrophy in a 7 year old girl: follow up
Autosomal Dominant High Hyperopia with mild retinoschisis
Usher Syndrome in a 5 year old boy?

Attendees

Aliaa Abdelhakim
Selwa Adulla Al-Hazzaa
Basamat Almoallem
Steven Archer
Bronwyn Bateman
Keyla Del Mar Bordonada Collazo
Scott Brodie
Brian Brooks
Daniel Chung
Razek Coussa
Thales Antonio Cabral de Guimaraes
Meghan Debenedictis
Monte Del Monte
Arlene Drack
Alina Dumitrescu
Jennifer Galvin
Shawn Hayes
Laryssa Huryn
Alex Iannaccone
Manuel Benjamin
Shuheik Kameya
Elizabeth Kellom
Arif Khan
Richard Lewis
Monique Leys
David Mackey
Ramiro Maldonado
Irene Maumenee
Marilyn Mets
Virginia Miraldi Utz
Basamat Moallem
Lauren Moissiy
Shizuo Mukai
Mark Orvidas
Wanda Pfeifer
Shreya Prabhu
Lev Prasov
Hanta Ralay Ranaivo
David Rousso
Eeva-Marja Sankila
Melanie Schmitt
Eduardo Silva
Kimberly Stepien
Florina Stoica
Rachel Sullivan
Elias Traboulsi
Terri Young
Wadiah Zein

Email Address

aha2133@cumc.columbia.edu
hazzaa@kfshrc.edu.sa
Basamat.almoallem@gmail.com
sarcher@umich.edu
bronwyn.bateman@comcast.net
keyla.bordonada@nih.gov
scott.brodie@nyumc.org
brooks_b@mail.nih.gov
daniel.chung@sparktx.com
coussar@ccf.org
thalesacguimaraes@hotmail.com; tguimaraes@willseye.org
marinom2@ccf.org
madm@umich.edu
arlene-drack@uiowa.edu
alina-dumitrescu@uiowa.edu
jennygalvin@gmail.com
shawn.hayes@retrophin.com
laryssa.huryn@nih.gov
alessandro.iannaccone@duke.edu
Ibanez_jayibanezmd@gmail.com
shuheik@nms.ac.jp
ekellom@wisc.edu
aokhan@yahoo.com
rlewis@bcm.edu
leysm@wvmedicine.org
D.Mackey@utas.edu.au
ram.maldonado@gmail.com
ihm2115@cumc.columbia.edu
mmets@luriechildrens.org
virginiautz@yahoo.com; virginia.utz@cchmc.org
basamat.almoallem@gmail.com
lauren.moissiy@blueprintgenetics.com
Shizuo_Mukai@meci.harvard.edu
mark.orvidas@retrophin.com
wanda-pfeifer@uiowa.edu
shreyap@med.umich.edu
lev.prasov@nih.gov
hralay@luriechildrens.org
david.rousso@sparktx.com
eeva-marja.sankila@helsinki.fi
maschmitt@wisc.edu
esilva2579@hotmail.com
kstepien@wisc.edu
florinastoica@gmail.com
rmsullivan2@wisc.edu
traboue@ccf.org
tyoung6@wisc.edu
wadiah.zein@nih.gov