

# 2018 Ophthalmic Genetics Study Club

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Friday, October 26, 2018  
 Ann & Robert H. Lurie Children's Hospital  
 225 East Chicago Avenue  
 11-142 (11<sup>th</sup> floor)

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7:30 am to 5:00 pm

*Breakfast & Lunch is Provided with registration*

Time	Title	Presenter
<b>Morning Session</b>		
7:30 to 7:55	<b>Breakfast</b>	
7:55 to 8:10	Case Presentation	Virginia Miraldi Uta
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	<b>Break</b>	
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

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12:20 to 1:05	Lunch Break	
<b>Afternoon Session</b>		
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	Case Presentation	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***

Elias Traboulsi  
 Arif Khan  
 Wadih Zein

Case Presentation  
 My Worst Nightmare  
 "Atypical" Usher Syndrome

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## Attendees

Aliaa Abdelhakim  
Steven Archer  
Bronwyn Bateman  
Keyla Del Mar Bordonada Collazo  
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  
Shuhei Kameya  
Elizabeth Kellom  
Arif Khan  
Richard Lewis  
Monique Leys  
David Mackey  
Ramiro Maldonado  
Irene Maumenee  
Marilyn Mets  
Virginia Miraldi Utz  
Lauren Moissiy  
Shizuo Mukai  
Mark Orvidas  
Wanda Pfeifer  
Shreya Prabhu  
Lev Prasov  
Hanta Ralay Ranaivo  
Melanie Schmitt  
Florina Stoica  
Rachel Sullivan  
Elias Traboulsi  
Terri Young  
Wadih Zein

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