2018 Ophthalmic Genetics Study Club

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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)

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<table>
<thead>
<tr>
<th>Time</th>
<th>Title</th>
<th>Presenter</th>
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<tbody>
<tr>
<td>Morning Session</td>
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<tr>
<td>7:30 to 7:50</td>
<td>Breakfast</td>
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<tr>
<td>7:50 to 7:55</td>
<td>Welcome</td>
<td>Marilyn Mets</td>
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<td>7:55 to 8:10</td>
<td>Case Presentation</td>
<td>Virginia Miraldi Utz</td>
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<td>8:15 to 8:30</td>
<td>Case Presentation</td>
<td>Melanie Schmitt</td>
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<td>8:35 to 8:50</td>
<td>Posterior Column Ataxia with Retinitis Pigmentosa</td>
<td>Rachel Sullivan</td>
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<td>8:55 to 9:10</td>
<td>A Family with Retinal Degeneration and Kidney Transplants</td>
<td>Aliaa Abdelhakim</td>
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<td>9:15 to 9:30</td>
<td>Expanding the Ocular Phenotype of Saul Wilson Syndrome</td>
<td>Wadih Zein</td>
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<td>9:35 to 9:50</td>
<td>Case Presentation</td>
<td>Meghan Debeneditis</td>
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<td>9:55 to 10:10</td>
<td>Retinal Splits in 3 Sibs</td>
<td>Arif Khan</td>
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<td>10:10 to 10:25</td>
<td>Break</td>
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<td>10:25 to 10:40</td>
<td>The Future of Ophthalmic Genetics</td>
<td>Irene Maumenee</td>
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<td>10:45 to 11:00</td>
<td>Interesting Genetic Testing Finds (2 cases)</td>
<td>Lauren Moissiy</td>
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<td>11:05 to 11:20</td>
<td>Follow-up on Familial Ocular Motor Apraxia</td>
<td>Steven Archer</td>
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<td>11:25 to 11:40</td>
<td>COMMAD: A New Syndrome by Biallelic Mutations in MITF</td>
<td>Brian Brooks</td>
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<tr>
<td>Time</td>
<td>Session</td>
<td>Speaker</td>
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<td>11:45 to 12:00</td>
<td>Vision Loss in Neurofibromatosis I</td>
<td>Alina Dumitrescu</td>
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<td>12:05 to 12:20</td>
<td>A Family with Congenital Retinal Non-Attachment</td>
<td>Selwa A. F. Al-Hazzaa/Basamat Almoallem</td>
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<td>12:20 to 1:05</td>
<td>Lunch Break</td>
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<td>Afternoon Session</td>
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<td>1:05 to 1:20</td>
<td>Case Presentation</td>
<td>Razek Coussa</td>
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<td>1:25 to 1:40</td>
<td>Seeing Stars: The Case for Whole Exome Sequencing in Optic Atrophy</td>
<td>Elizabeth Kellom</td>
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<td>1:45 to 2:00</td>
<td>An Unusual Presentation of Williams’ Syndrome</td>
<td>Richard Lewis</td>
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<td>2:05 to 2:20</td>
<td>Retinal Dystrophy</td>
<td>Monique Leys</td>
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<td>2:25 to 2:40</td>
<td>Interesting Case of RPE65 LCA</td>
<td>Arlene Drack</td>
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<td>2:40 to 2:55</td>
<td>Afternoon break</td>
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<td>2:55 to 3:10</td>
<td>Blurring the Difference of Congenital and Primary Open Angle Glaucoma</td>
<td>David Mackey</td>
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<td>3:15 to 3:30</td>
<td>Case Presentation</td>
<td>Shreya Prabhu</td>
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<td>3:35 to 3:50</td>
<td>Primary Congenital Glaucoma-New TEK Mutations</td>
<td>Terri Young</td>
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<td>3:55 to 4:10</td>
<td>17 Week Gestational Age Retina in Norrie Disease</td>
<td>Shizuo Mukai</td>
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<tr>
<td>4:15 to 4:30</td>
<td>Mystery Family (Almost) Revealed: A Tale of (At Least!) Two Stories</td>
<td>Alex Iannaccone</td>
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<td>4:35 to 4:45</td>
<td>Child with AAMR and Review of the Literature</td>
<td>Jennifer Galvin</td>
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<td>4:45 to 5:00</td>
<td>Electronegative ERGs at The University of Iowa</td>
<td>Wanda Pfeifer</td>
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<td>5:00 pm</td>
<td>Adjournment</td>
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**Case Presentations Pending if Time Allows**

- Scott Brodie: Atypical Cone-Rod Dystrophy
- Elias Traboulsi: Case Presentation
- Arif Khan: My Worst Nightmare
- Wadih Zein: “Atypical” Usher Syndrome
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 Cossa
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 Kellog
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 Stepian
Florina Stoica
Rachel Sullivan
Elias Traboulsi
Terri Young
Wadih Zein

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