### Agenda

#### Friday August 24, 2018

- Arrivals and Registration

#### Saturday August 25, 2018

<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
<th>Chair/Speaker</th>
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<tbody>
<tr>
<td>7:00 AM – 8:15 AM</td>
<td>Breakfast&lt;br&gt;<strong>Venue:</strong> Vistas Dining Room</td>
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<tr>
<td>8:30 AM-8:45 AM</td>
<td>Introduction and Announcements&lt;br&gt;Micheil Innes and Kym Boycott&lt;br&gt;<strong>KC 101 &amp; 103 (all sessions)</strong></td>
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<tr>
<td>8:45 AM – 10:15 AM</td>
<td>Session I: Founding Fellows Lecture and Teratogens&lt;br&gt;<strong>Chair:</strong> Micheil Innes</td>
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<tr>
<td>8:45 AM</td>
<td>A retrospective look at 45 years of the fetal alcohol spectrum disorder: How did we get here from where we started&lt;br&gt;<strong>Keynote speaker:</strong> Kenneth Lyons Jones - Department of Pediatrics, University of California, San Diego, USA</td>
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<td>9:45 AM</td>
<td>Zika virus as a cause of birth defects: Were the teratogenic effects of Zika virus missed for decades?&lt;br&gt;<strong>Sonja A. Rasmussen - Centers for Disease Control and Prevention, USA</strong></td>
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<tr>
<td>10:00 AM</td>
<td>The pesticide synergist Piperonyl Butoxide inhibits sonic hedgehog signaling and causes holoprosencephaly in mice: Gene-environment interactions and subteratogenic effects&lt;br&gt;<strong>Robert L Lipinski - University of Wisconsin-Madison, USA</strong></td>
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<tr>
<td>10:45 AM – 11:45 AM</td>
<td>Session II: Placenta and Prenatal&lt;br&gt;<strong>Chair:</strong> Francois Bernier</td>
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<tr>
<td>10:45 AM</td>
<td>Stillbirth: Is the placenta the problem?&lt;br&gt;<strong>Elspeth McPherson - Marshfield Clinic Research Institute, USA</strong></td>
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<tr>
<td>11:00 AM</td>
<td>Beyond mesenchymal dysplasia: Clinical characterization of placental pathology in Beckwith-Wiedemann syndrome&lt;br&gt;<strong>Rebecca Linn - Children's Hospital of Philadelphia, USA</strong></td>
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<tr>
<td>11:15 AM</td>
<td>What not to expect when you’re expecting: Unusual cases of placental mosaicism detected on non-invasive prenatal screening&lt;br&gt;<em><em>Amanda Barone</em> - Children's Hospital of Philadelphia, USA</em>*</td>
<td></td>
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<tr>
<td>11:30 AM</td>
<td>Prenatal profile of Pallister Killian syndrome: Retrospective analysis of 114 pregnancies, literature review and approach to prenatal diagnosis&lt;br&gt;<strong>Ian Krantz - Children's Hospital of Philadelphia, USA</strong></td>
<td></td>
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<tr>
<td>11:45 AM – 1:15 PM</td>
<td>Lunch&lt;br&gt;<strong>Venue:</strong> Vistas Dining Room</td>
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### Session III: Central Nervous System Diseases

**Chair:** David Dyment

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<th>Time</th>
<th>Title</th>
<th>Speaker</th>
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<tbody>
<tr>
<td>1:15 PM</td>
<td>The patterns of brain malformations seen in discordant twins</td>
<td>William B Dobyns</td>
<td>University of Washington and Seattle Children’s Hospital, USA</td>
</tr>
<tr>
<td>1:30 PM</td>
<td>Pathogenic $DDX3X$ mutations impair RNA metabolism and disrupt neuronal fate during fetal cortical development</td>
<td>Elliot H. Sherr</td>
<td>University of California, USA</td>
</tr>
<tr>
<td>1:45 PM</td>
<td>POGZ-CHAMP1 are molecular partners associated with neurodevelopmental disorders and phenotypic ontology for olfactory bulb hypoplasia and intestinal malrotation</td>
<td>Joseph T. Shieh</td>
<td>University of California San Francisco, USA</td>
</tr>
<tr>
<td>2:00 PM</td>
<td>Arthrogryposis – Genetic update</td>
<td>Judith G. Hall</td>
<td>University of British Columbia, Canada</td>
</tr>
<tr>
<td>2:15 PM</td>
<td>Variants in $MAP4K4$ cause a novel and potentially treatable form of neurologic dysfunction with cardiac anomalies</td>
<td>Elizabeth J. Bhoj</td>
<td>Children’s Hospital of Philadelphia, USA</td>
</tr>
<tr>
<td>2:30 PM</td>
<td>Variants of $NAA15$: Another newly recognized etiology for neurodevelopmental impairment as illustrated by 38 cases</td>
<td>Alan F. Rope</td>
<td>Kaiser Permanente Northwest, USA</td>
</tr>
<tr>
<td>2:45 PM</td>
<td>$PIK3CB$ causes a novel neurodevelopmental and overgrowth syndrome characterized by macrocephaly and polymicrogyria</td>
<td>Priya T. Bhola*</td>
<td>Children’s Hospital of Eastern Ontario, Canada</td>
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### Session IV: New Insights into Old Syndromes

**Chair:** Oana Caluseriu

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<tr>
<th>Time</th>
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<th>Speaker</th>
<th>Institution</th>
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<tbody>
<tr>
<td>3:30 PM</td>
<td>Kabuki syndrome: International consensus diagnostic criteria</td>
<td>Margaret Adam</td>
<td>University of Washington, USA</td>
</tr>
<tr>
<td>3:45 PM</td>
<td>In depth phenotyping of Snyder-Robinson syndrome: Old and new findings</td>
<td>Angela Peron*</td>
<td>University of Milan, Italy</td>
</tr>
<tr>
<td>4:00 PM</td>
<td>De novo truncating variants in $WHSC1$ recapitulate the Wolf-Hirschhorn (4p16.3 microdeletion) syndrome phenotype</td>
<td>Nada Derar</td>
<td>Medical College of Wisconsin, USA</td>
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### Agenda

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<thead>
<tr>
<th>Time</th>
<th>Session</th>
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</table>
| 3:30 PM    | CHARGE syndrome in the era of molecular diagnosis: Need for higher clinical suspicion - findings in the CCHMC CHARGE center cohort  
*Brittany Simpson* - *Cincinnati Children's Hospital Medical Center, USA* |
| 4:30 PM    | Chimeric versus mosaic genome-wide paternal uniparental isodisomy as etiologies for Beckwith-Wiedemann Syndrome  
*Sarah Sheppard* - *Children’s Hospital of Philadelphia, USA* |
| 4:45 PM    | Influence of molecular classes and growth hormone on dysmorphology features in Prader-Willi syndrome: A multicenter study  
*Virginia Kimonis* - *University of California, Irvine, USA* |
| 5:00 PM    | Further delineation of Temtamy syndrome of corpus callosum and ocular abnormalities  
*Laila Ali Alrakaf* - *Alfaisal University, Saudi Arabia* |
| 5:30 PM    | ASXL-related disorders: The first year of our registry  
*Bianca Russell* - *Cincinnati Children’s Hospital, USA* |
| 5:45 PM    | Method of diagnosis relative to phenotype in Incontinentia Pigmenti  
*Angela Schueerle* - *UT Southwestern Medical Center, USA* |

**6:00 PM – 7:30 PM**  
**Dinner**  
**Venue:** Vistas Dining Room

**7:30 PM – 8:30 PM**  
**Poster Session:** Posters 1-36  
**KC 203/205**

**8:30 PM – 9:30 PM**  
**Poster Session:** Posters 37-71  
**KC 203/205**

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**Sunday August 26, 2018**

**7:00 AM – 8:15 AM**  
**Breakfast**  
**Venue:** Vistas Dining Room

**8:30 AM – 10:30 AM**  
**Session V:** Gene Environment Interaction and Complex Disease  
**Chair:** Joseph T. Shieh

**8:30 AM**  
Genetic and environmental causes of congenital malformation  
**Keynote speaker:** Sally Dunwoodie, University of New South Wales, Australia  
**Introduction:** Kym Boycott

**9:30 AM**  
Variation in aortic disease in mice and humans with elastin insufficiency and Williams Beuren syndrome is modified by secondary changes in extracellular matrix and immune pathway genes  
Beth Kozel – National Heart, Blood, and Lung Institute of NIH, USA

**9:45 AM**  
Pregestational diabetes and neural tube defects in Manitoba  
*Jane Evans* - *University of Manitoba, Canada*
### Agenda

<table>
<thead>
<tr>
<th>Time</th>
<th>Session/Content</th>
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| **10:00 AM**  | Auriculocondylar syndrome: A Craniofacial disorder with a vasospastic complication?  
Matthew A. Deardorff - The Children’s Hospital of Philadelphia, USA |
| **10:15 PM**  | Mutations in the Epithelial Cadherin-P120-Catenin complex are significant contributors to non-syndromic cleft lip with or without cleft palate  
Timothy Cox - University of Washington, USA |
| **10:30 AM – 11:00 AM** | Break KC 203/205                                            |
| **11:00 AM – 12:15 PM** | Session VI: Syndromes - Michael Cohen  
Chair: John Carey |
| **11:00 AM**  | Delineation and definition of the Carey-Fineman-Ziter-Syndrome (CFZS): A tale of (ten) cities  
John C. Carey - University of Utah, USA |
| **11:15 AM**  | The many faces of SOS1: Exon 20 frameshift mutations are associated with multiple mucosal neuroma syndrome – A benign MEN2B phenocopy  
Micheil Innes - Alberta Children’s Hospital Research Institute, Canada |
| **11:30 AM**  | The full spectrum of post-zygotic PIK3CA mutations in non-syndromic lymphatic malformations  
James T. Bennett - University of Washington, USA |
| **11:45 AM**  | SUZ12-related overgrowth: Expanding the phenotype  
William Gibson - University of British Columbia, Canada |
| **12:15 PM – 1:30 PM** | Lunch  
Venue: Vistas Dining Room |
| **1:30 PM – 3:30 PM** | Session VII: Next-Generation Phenotyping 1  
Chair: Anne O’Donnell |
| **1:30 PM**  | Phenotyping with and without typing: Capturing and structuring your patient data  
Keynote speaker: Michael Brudno - Department of Computer Science, University of Toronto, Centre for Computational Medicine, The Hospital for Sick Children, Canada |
| **2:30 PM**  | Deep phenotyping in the NIH undiagnosed diseases network  
John Mulvihill - National Human Genome Research Institute of NIH, USA |
| **2:45 PM**  | Blended phenotypes: An interesting subset of the first 400 patients of the CAUSES research study  
Alison M. Elliott - University of British Columbia, Canada |
| **3:00 PM**  | Confirming the candidacy of 46 disease genes and delineation of their phenotypic expression  
Fowzan S. Alkuraya - Alfaaisal University, Saudi Arabia |
| **3:15 PM**  | HARS syndrome: Expansion of the natural history of the disorder associated with homozygous Y454S mutations in the Histidyl-tRNA Synthetase (HARS) gene  
Victoria Mok Siu - University of Western Ontario, Canada |
# Agenda

<table>
<thead>
<tr>
<th>Time</th>
<th>Session/Activity</th>
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<tbody>
<tr>
<td>3:30 PM – 4:00 PM</td>
<td>Break KC 203/205</td>
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<tr>
<td>4:00 PM – 6:00 PM</td>
<td>Session VIII: Next-Generation Phenotyping 2&lt;br&gt;Chair: Elizabeth Bhoj</td>
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**4:00 PM**

The impact of next generation sequencing on syndrome delineation<br>
*Michael Bamshad - University of Washington, USA*

**4:15 PM**

Alternative diagnoses for individuals referred for exome sequencing with a clinical diagnosis of Dubowitz syndrome<br>
*David Dyment - Children’s Hospital of Eastern Ontario Research Institute, Canada*

**4:30 PM**

Cohort analysis reveals diverse genetic etiology underlying Dubowitz syndrome<br>
*Anne O’Donnell - Broad Institute, USA*

**4:45 PM**

Final demise of the lethal recessive acrofacial dysostosis syndrome Rodriguez type<br>
*Elaine H. Zackai - Children’s Hospital of Philadelphia, USA*

**5:00 PM**

Deep phenotyping using 3D craniofacial morphometry<br>
*Francois Bernier - Alberta Children’s Hospital Research Institute, Canada*

**5:15 PM**

The facial phenotype of Aymé-Gripp syndrome is recognizable by the next generation phenotyping software<br>
*Karen W. Gripp - A. I. duPont Hospital for Children, USA*

**5:30 PM**

Emerging phenotype of a CREBBP-Related disorder distinct from Rubinstein-Taybi syndrome<br>
*Julie Kaplan - Nemours A.I. DuPont Hospital for Children, USA*

**5:45 PM**

Diagnosis and Management of the phenotypic spectrum of twins with Beckwith-Wiedemann Syndrome<br>
*Jennifer L. Cohen* - Children’s Hospital of Philadelphia, USA

**6:00 PM – 7:30 PM**<br>
**Dinner**<br>
Venue: Vistas Dining Room

**8:00 PM – 10:00 PM**<br>
**Activity**<br>
GH 101&103

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**Monday August 27, 2018**

**7:00 AM – 8:15 AM**<br>
**Breakfast**<br>
Venue: Vistas Dining room

**8:30 AM – 10:30 AM**<br>
**Session IX: Treatment 1**<br>
Chair: Sue White

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**8:30 AM**<br>
Emerging therapies in skeletal dysplasias: The journey from diagnosis to precision intervention<br>
**Keynote speaker:** Ravi Savarirayan - Murdoch Children’s Research Institute, and University of Melbourne, Australia
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<tr>
<th>Time</th>
<th>Session</th>
<th>Speaker</th>
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<tbody>
<tr>
<td>9:30 AM</td>
<td>Transformation of clinical care for plexiform</td>
<td>David Viskochil</td>
<td>University of Utah, USA</td>
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<td>neurofibromas in Neurofibromatosis type 1 (NF1)</td>
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<td>with the advent of MEK inhibitors</td>
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<td>9:45 AM</td>
<td>One-year pilot treatment trial oral nitinamide</td>
<td>Brian Brooks</td>
<td>National Eye Institute and National Human Genome Research Institute of NIH, USA</td>
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<td>in Oculocutaneous Albinism, type 1B</td>
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<td>10:00 AM</td>
<td>A child with an advanced lymphatic anomaly</td>
<td>Dong Li</td>
<td>The Children’s Hospital of Philadelphia, USA</td>
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<td>due to a novel gain-of-function mutation in ARAF</td>
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<td>has remission following treatment with a MEK inhibitor</td>
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<td>10:15 AM</td>
<td>Landscape of treatable rare diseases</td>
<td>Alison Eaton</td>
<td>University of Calgary, Canada</td>
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<tr>
<td>Break KC 203/205</td>
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<tr>
<td>11:00 AM</td>
<td>Session X: Treatment 2</td>
<td>Dorothy K. Grange</td>
<td>Washington University School of Medicine, St. Louis Children's Hospital, USA</td>
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<td>Cantu syndrome: Treatment of a mouse model</td>
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<td>with the sulfonylurea glibenclamide and</td>
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<td></td>
<td>potential for human therapy</td>
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<tr>
<td>11:15 AM</td>
<td>Role of Leptin and Adiponectin in PIK3CA-</td>
<td>Kim M Keppler</td>
<td>National Human Genome Research Institute of NIH, USA</td>
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<td>Related Overgrowth Spectrum (PROS) –</td>
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<td>Implications for Phenotype and Response to</td>
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<td>Sirolimus Therapy</td>
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<td>11:30 AM</td>
<td>FASTKD2 mutations associated with development</td>
<td>Michael J. Lyons</td>
<td>Greenwood Genetic Center, USA</td>
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<td>delay, tremor, and chronic kidney disease</td>
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<td>responsive to Coenzyme Q10</td>
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<td>12:00 PM</td>
<td>Dietary treatment for Neurofibromatosis type 1</td>
<td>Rachel Lombardo</td>
<td>Cincinnati Children’s Hospital and Medical Center, USA</td>
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<td>Dietietary treatment for Neurofibromatosis</td>
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<td>type 1</td>
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<td>Impact of molecular diagnosis on management,</td>
<td>Anne Slavotinek</td>
<td>University of California, San Francisco, USA</td>
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<td>treatment, and long term outcomes in congenital</td>
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<td>diarrheal disorders</td>
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<td>12:15 PM</td>
<td>Nonsense suppression therapy can rescue BMP4</td>
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<td>nonsense mutation in vitro and the bmp4st72</td>
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<td>allele in vivo in zebrafish</td>
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<tr>
<td>12:30 PM</td>
<td>Lunch: Vistas Dining Room (until 1:30)</td>
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<td>Free Time</td>
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## Agenda

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<th>Time</th>
<th>Event</th>
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<tbody>
<tr>
<td>12:30 PM - 2:00 PM</td>
<td>Fellows session</td>
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<tr>
<td>5:30 PM – 7:30 PM</td>
<td>Dinner&lt;br&gt;<strong>Venue:</strong> Vistas Dining Room</td>
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<tr>
<td>8:00 PM – 10:00 PM</td>
<td><strong>Unknowns Session</strong>&lt;br&gt;GH 101&amp;103&lt;br&gt;<strong>Chairs:</strong> Marilyn Jones and Micheil Innes</td>
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### Tuesday August 28, 2018

<table>
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<tr>
<th>Time</th>
<th>Event</th>
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<tbody>
<tr>
<td>7:00 AM – 8:15 AM</td>
<td><strong>Breakfast:</strong>&lt;br&gt;<strong>Venue:</strong> Vistas Dining Room</td>
</tr>
<tr>
<td>8:30 AM – 10:30 AM</td>
<td><strong>Session XI: Epigenetics</strong>&lt;br&gt;<strong>Chair:</strong> Matthew A. Deardorff</td>
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</table>

### 8:30 AM

**Functional molecular diagnostic opportunities created by cross-talk between histone modifications and DNA methylation**

**Keynote speaker:** Rosanna Weksberg - Department of Pediatrics, University of Toronto, Genetics and Genome Biology Program, Hospital for Sick Children, Canada

### 9:30 AM

ADNP syndrome has a unique genomic DNA methylation signature that suggests *PACSIN1* may contribute to the disease mechanism

**Eric Bend*** - Greenwood Genetic Center, USA

### 9:45 AM

Identification of a unique DNA methylation signature associated with Nicolaides-Baraitser syndrome

**Resham Ejaz*** - University of Toronto, Canada

### 10:00 AM

Why do *FMR1* full mutations become methylated? An outstanding question

**Giovanni Neri** - Institute of Genomic Medicine, Catholic University School of Medicine, Italy

### 10:15 AM

The phenotype spectrum of Beckwith-Wiedemann syndrome due to loss of methylation at imprinting center 2

**Jennifer M. Kalish** - The Children’s Hospital of Philadelphia, USA

### 11:00 AM – 11:00 AM

**Break KC 203/205**

### 11:00 AM – 12:30 PM

**Session XII: New and Emerging Syndromes I**

**Chair:** Fowzan Alkuraya

### 11:00 AM

Examination of the landscape of Histone Lysine Methylases and Demethylases in human developmental disorders leads to the identification of novel syndromes

**Victor Faundes*** - University of Manchester

### 11:15 AM

Mutations in the BAF-complex subunit *DPF2* associated with Coffin-Siris syndrome

**André Reis** - Institute of Human Genetics, FAU Erlangen-Nürnberg, Germany
## Agenda

### 11:30 AM
Missense mutations disrupting the helicase domain of chromatin remodeller CHD3 cause a novel neurodevelopmental syndrome with intellectual disability, macrocephaly and impaired speech and language  
*Philippe M. Campeau - CHU Sainte-Justine Research Center Canada*

### 11:45 AM
Clinical and molecular characterization of TFE3 mutation-related neurocutaneous syndrome  
*Daphné Lehalle - Université de Bourgogne Franche-Comté, France*

### 12:00 PM
Characterization of novel X-linked, IGSF1-associated human and canine overgrowth  
*Thoa K. Ha* - University of California San Francisco, USA

### 12:15 PM
Craniofacial anomalies, ptosis, agenesis of the corpus callosum, and developmental delay associated with loss of function variants in ZNF462: Humans and zebrafish  
*Paul Kruszka - National Human Genome Research Institute of NIH, USA*

### 12:30 PM – 1:45 PM  
**Lunch**  
*Venue: Vistas Dining Room*

### 1:45 PM – 4:30 PM
**Session XIII: New and Emerging Syndromes 2**  
**Chair: Alison M. Elliott**

#### 1:45 PM
The human engrailed-1 (EN1) locus: Two different types of mutations resulting in limb and brain malformation phenotypes  
*Andrea Superti-Furga - Lausanne University Hospital, Switzerland*

#### 2:00 PM
Do heterozygous loss-of-function variants in MTOR cause a treatable neurodevelopmental phenotype?  
*Susan White - Victorian Clinical Genetics Services, Australia*

#### 2:15 PM
Missense mutation in actin-building protein plastin 3 cause X-linked multiple congenital anomalies with diaphragmatic hernia  
*Robin D. Clark - Loma Linda University School of Medicine, USA*

#### 2:30 PM
Phenotypic spectrum of Au-Kline syndrome: 12 new cases and review of the literature  
*P.Y. Billie Au - University of Calgary, Canada*

#### 2:45 PM
Expansion of the GTPBP2-Associated Neuro-ectodermal Syndrome Phenotype  
*Melissa Carter - Children’s Hospital of Eastern Ontario, Canada*

### 3:00 PM  
**Break KC 203/205**

### 3:15 PM
*UBE2A* related XLID – a recognizable X-linked intellectual disability syndrome  
*Roger Stevenson - Greenwood Genetic Center, USA*
<table>
<thead>
<tr>
<th>Time</th>
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<tbody>
<tr>
<td>3:30 PM</td>
<td>A novel neuromuscular disorder linked to <em>STX4</em></td>
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<td><em>Carlos E. Prada - Cincinnati Children’s Hospital Medical Center, USA</em></td>
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<tr>
<td>3:45 PM</td>
<td>Truncating variants in <em>PPMID</em> case a recognizable dysmorphic and behavioral phenotype with similarities to Williams syndrome</td>
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<td><em>Cynthia J. Curry - University of California, San Francisco/Fresno, USA</em></td>
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<tr>
<td>4:00 PM</td>
<td>Armfield XLID syndrome has a distinct phenotype (short stature, small hands/feet, ocular anomalies, seizures), results from mutations in <em>FAM50A</em>, and is a spliceosomopathy</td>
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<td><em>Charles E. Schwartz - Greenwood Genetic Center, USA</em></td>
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<td>4:15 PM</td>
<td>10 Faces You Should Know</td>
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<td><em>Charles E. Schwartz - Greenwood Genetic Center, USA</em></td>
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<tr>
<td>4:30 PM</td>
<td>Presentation of Fellows/Trainee Awards</td>
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| 6:30 PM – 1:00 AM | Dinner Reception and Dance  
*Venue: KC 201/203*                |

**Wednesday August 29, 2018**

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<tr>
<th>Time</th>
<th>Session</th>
</tr>
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| 7:00 AM – 9:00 AM | Breakfast  
*Venue: Vistas Dining Room*                        |