Preliminary Workshop at a Glance

Friday, August 25th

1:00-9:00 pm: Registration

6:00-9:00 pm: Reception

Saturday, August 26th

7:15-8:30 AM: Breakfast

Session 1

8:30 AM: Welcome

Introduction: Karen Gripp

8:45-9:30 AM: Founding Fellow Lecture, Judith G. Hall
“Pallister Hall Syndrome – A Tale of Buried Treasure”

Introduction: Ian Krantz

9:30-10:30 AM: Invited Speaker: Dale Dorsett, Saint Louis University, Saint Louis, Missouri
“Drosophila melanogaster as a Model for Transcriptional Dysregulation Syndromes”

10:30-11:00 AM: Break

Disorders of Transcriptional Regulation I:
Moderator: Matthew Deardorff

11:00 AM: Disorders of Transcriptional Regulation (DTRs) – A Growing Group of Disorders with Phenotypic Overlap with Cornelia De Lange Syndrome. Ian Krantz

11:15 AM: Dias-Logan Syndrome: Delineating a Newly Recognized Disorder of Transcriptional Regulation
Angela Peron (Trainee)
11:30 AM:  *Mutations in H3f3a And H3f3b Encoding Histone 3.3 Cause The First Reported Germline Histone Syndrome: Report Of 23 Patients With Neurodevelopmental And Congenital Manifestations*  
Elizabeth Bhoj

11:45 AM:  *Mutations in Ebf3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia and Facial Dysmorphism*  
Katta Girisha

12:00 PM:  *A Potential Treatment for the Undergrowth Phenotype Associated with the NSD1/ 5q35.2-5q35.3 Duplication Syndrome*  
Fabiola Quintero

12:15 pm-1:30 PM:  Lunch

**Session 2**

**Disorders of Transcriptional Regulation II:**
*Moderator: Timothy Cox*

1:30 PM:  *A Homozygous Splice Site Variant in Taf8 Causes Intellectual Disability, Brain Abnormalities and In Vitro Evidence of Absent Taf8 with Alterations in the Tfiid Complex*  
Cynthia Curry

1:45 PM:  *De Novo, Deleterious Sequence Variants in the Transcription Factor Pbx1 are Associated with Intellectual Disability and Ear, Branchial Arch, Renal, Cardiac and Diaphragmatic Abnormalities*  
Anne Slavotinek

2:00 PM:  *De Novo Variants in The Histone Methylase Kmt5b Cause a Novel Disorder of Transcription*  
Sarah Sheppard (Trainee)

2:15 PM:  *Bicornuate Uterus in Cornelia de Lange Syndrome: Overlap with Hand-Foot-Genital Syndrome?*  
Antonic Kline

2:30-3:15 PM:  Break

**Session 3**

**Disorders of Transcriptional Regulation III:**
*Moderator: Antonie Kline*
Identification of a Defining Peripheral Blood DNA Methylation Signature of Kabuki Syndrome Enables Kmt2d Variant Classification
Charles Schwartz

Deletion of an Evolutionarily Conserved Chromatin Insulator Element Associated with Elevated Retinoid Signaling as the Genetic Basis for an OAVS-Like Presentation in Mice
Timothy Cox

De Novo Variants in Kmt2e, a Candidate Haploinsufficient Gene, are a Novel Cause of Intellectual Disability
Anne O'Donnell-Luria

Smc1a Mutations Cause Mechanistically Separable Allelic Disorders: Atypical Cornelia De Lange Syndrome and a Rett-Like Epileptic Encephalopathy.
Matthew Deardorff

Neurologic, Neuromuscular and Craniofacial syndromes:
Moderator: Ian Glass

The Genetic Landscape of Cerebellar Malformations
William Dobyns

Tuba1a: Outcome of Mosaicism and Phenotypic Analysis
Marc Williams

Phenotypic Spectrum of Females Carrying Mutations in Zc4h2
Michael Bamshad

Session 4

Announcements
Introduction: Robert Lipinski

Syndromes & Isolated Birth Defects Involving Malformations of the Developing Foregut I: Moderator: Robert Lipinski

9:15 AM: Acinar Dysplasia, Tbx4 and Lung Branching Morphogenesis Cara Skraban

9:30 AM: Segmental Tracheal Atresia: It's Not All Chaos Jane Evans

9:45 AM: Loss-of-Function Variants in Med12 are a Cause of Hardikar Syndrome Chaya Murali (Trainee)

10:00-10:30 AM: Break

Session 5

Syndromes & Isolated Birth Defects Involving Malformations of the Developing Foregut II: Moderator: Omar Abdul-Rahman

10:30 AM: Intestinal Malrotation: A Review and Report of a Family with Craniosynostosis and Malrotation Bryan Hall

10:45 AM: The Role of the Kruppel-Like Transcription Factor Klf5 in Foregut Development Mary Beth Dinulos

11:00 AM: The Role of Foxf1 in Foregut Development Paul Mark

11:15 AM: Esophageal Atresia/Tracheoesophageal Fistula: Rady Children’s Hospital San Diego Experience Carolina Galarreta Aima

11:30 AM: Genetic Diagnoses and Associated Malformations in Fetuses Prenatally Diagnosed with Esophageal Atresia Mersedeh Rohanizadegan (Trainee)

11:45 AM: Biallelic Mutations in Waardenburg Syndrome Genes Cause Recognizable Arthrogryposis Syndromes Roger Stevenson
Session 6

Introduction: Rolf Stottmann

1:15-2:15 PM: Invited Speaker: Dr. Robert Krauss, Icahn School of Medicine Mount Sinai, New York, NY.
   “Mechanisms of Gene-Environment Interaction in Holoprosencephaly”

Teratogens and Malformations:
Moderator: Elizabeth Bhaj

2:15 PM: Update on a Timeline of Critical Developmental Stages for the Teratogenic Causation of Birth Defects
   Art Aylsworth

2:30 PM: The Risk of Maternal Autoimmune Disease and Associated Treatments on the Development of Infantile Hemangioms in the Offspring
   Ken Jones

2:45 PM: Unilateral Abdominal Wall Hypoplasia- A Feature of Diabetic Embryopathy?
   Cathy Stevens

3:00 PM: Congenital Zika Virus Infection with Arthrogryposis and Paralysis of the Diaphragm
   Cynthia Moore

3:15 PM: Genetic Sensitivity to Depakote-Induced Birth Defects: Efforts to Identify Susceptibility Genes
   Richard Finnell

3:30-4:00 PM: Break

Session 7

New syndromes and new insights into old syndromes:
Moderator: Melanie Manning

4:00 PM: Of Mice and Men: First Example of Homozygous Variant in Ednra Causing Lethal Craniofacial Anomalies Similar to those in Knockout Mice
   Elaine Zackai
A Recognizable Phenotype Including Macrocephaly, Ligamentous Laxity and Developmental Delay is Associated with Germline De Novo Taok1 Variants
Karen Gripp

Biallelic Mutations in Pisd Identified in Siblings with Congenital Cataracts and Extreme Short Stature Adds a Novel Disorder to the Emerging Family of Mitochondrial Chaperonopathies
Micheil Innes

Sonic Hedgehog Signaling Targets Foxf2 During Upper Lip Morphogenesis and Cleft Lip Pathogenesis
Robert Lipinski

Adamts11 Mutations Cause Habsburg Jaw or Mandibular Prognathism
Nik Kantaputra

Session 8

Dysmorphology (syndromes and malformations) in minority and unique populations I:

Introduction: Paul Kruszka

8:00-8:15 AM: Announcements

Invited Speaker:
Maximillian Muenke, NIH/NHGRI:
“Delineating Dysmorphology in Underrepresented Minority Populations”

Moderator Paul Kruszka

9:15 AM: Unusual Physical Findings in Known Genetic Syndromes Affecting the African American and Hispanic Populations: Report from the Largest Genetic Center in the DMV Area.
Eyby Leon

9:30 AM: Embryonic Lethal Mendelian Phenotypes: A Large Cohort from a Consanguineous Population
Fowzan Alkuraya
9:45 AM:  
*Cornelia De Lange Syndrome in Diverse Populations*
Leah Dowsett (Trainee)

10:00-10:30 AM:  
Break

Session 9

*Dysmorphology (syndromes and malformations) in minority and unique populations II:*
*Moderator: Fowzan Alkuraya*

10:30 AM:  
*Congenital Heart Malformations in Sub-Saharan Africa and Asia*
Paul Kruszka

10:45 AM:  
*Clinical Spectrum and Molecular Genetics of Kabuki Syndrome in Hong Kong – 22 Years’ Experience*
Ho-Ming Luk

11:00 AM:  
*Ritscher-Schinzel/3-C Syndrome – Further Delineation of a First Nations Cohort and Implications in Cholesterol Homeostasis*
Alison Elliot

11:15 AM:  
*Clinical Features of Beckwith-Wiedemann Syndrome in Diverse Populations*
Jennifer Kalish

11:30 AM:  
*A New Ashkenazi Jewish Syndrome? Nup188 and its Role in a Newly Described Oculo-Facial-Neuro Syndrome.*
Jennifer Cohen (Trainee)

11:45 AM:  
*Homozygous Boricua Tbck Mutation Causes Neurodegeneration and Aberrant Autophagy*
Xilma Ortiz-Gonzalez

12:00 PM:  
*Cataloguing Rare Genetic Disorders Found Amongst Irish Travellers*
Sally A Lynch

12:15 PM:  
Lunch

12:15-1:30 PM:  
Fellows Session (Lunch)

Free Afternoon

6:30-8:00 PM:  
Dinner

8:00-10:00 PM:  
Unknown Session
Moderators: Marilyn Jones and Mike Innes

Tuesday, August 29th

7:30-8:30 AM: Breakfast

Session 11

Introduction: David Viskochil

8:30-9:30 AM: Invited Speaker:
“The Natural History of Genetic Disorders: the Centerpiece of the ‘Central Dogma’ of Clinical Genetics”
John C. Carey, University of Utah Health Care and Primary Children’s Hospital, Salt Lake City, Utah

Natural History of Syndromes I:
Moderator: Paul Mark

9:30 AM: Phenotype and Natural History in 49 Individuals with Satb2-Associated Syndrome
Yuri Zarate

9:45 AM: Natural History of Spine Disease in the Mucopolysaccharidoses: Integrating Human and Animal Model Data
Dena Matalon (Trainee)

10:00 AM: The Natural History Of Craniosynostosis And Tumor Risk
Joseph Shieh

10:15 AM: Natural History of Nevoid Basal Cell Carcinoma (Gorlin) Syndrome
Nina Gold (Trainee)

10:30-11:00 AM: Break

Session 12

Natural History of Syndromes II:
Moderator: Tara Wenger

11:00 AM: They’re not all Giving Us the (Fifth) Finger: Natural History of 46 Unreported Patients from the Coffin-Siris Syndrome/Baf Pathway Registry
Samantha Schrier Vergano

11:15 AM: Review & Natural History of the Naa10-Associated Disorders
Alan Rope

11:30 AM: *Cantu Syndrome Natural History Studies: Clinical Investigations on 18 Patients and Report on International Redcap Registry Data on 58 Patients*  
Dorothy Grange

11:45 AM: *An Investigation into the Natural History of Bardet-Biedl Syndrome*  
Farrah Rajab (Trainee)

12:00 PM: *Medically Actionable Comorbidities in Adults with Costello Syndrome*  
K. Nicole Weaver

12:15-1:45 PM: Lunch

Session 13

*Natural History of Syndromes III:*
*Moderator: Nicole Weaver*

1:45 PM: *Rasa1-Related Disorders*  
David Stevenson

2:00 PM: *Maladaptive Behaviors in Children with Angelman Syndrome*  
Lynne Bird

2:15 PM: *Neonatal Marfan Syndrome – A Comprehensive Review and Assessment of Prognosis*  
David Weaver

2:30 PM: *Cardiovascular Manifestations and Evaluation of High Blood Pressure in Williams-Beuren Syndrome*  
Miguel Del Campo

2:45 PM: *Characterization and Natural History of Genital Tract Anomalies and Tumors in Proteus Syndrome*  
Kim Keppler-Noreuil

3:00-3:30 PM: Break

Session 14

*Miscellaneous Topics*
*Moderator: Cara Skraban*

3:30 PM: *Cardiac Anomalies in Monozygotic Twins*  
Elizabeth McPherson
3:45 PM: Landscape of Pleiotropic Proteins Causing Human Malformation Syndromes
Kym Boycott

4:00 PM: Mouse Knock-In of a Premature Stop Allele of Frizzled-2 Recapitulates
Human Autosomal Dominant Omomydysplasia Phenotypes
Rolf Stottmann

4:15-4:30 PM: Trainee Awards

6:30-8:30 PM: Dinner

8:30PM-12:00AM: Closing Party