38th Annual David W. Smith Workshop on Malformations and Morphogenesis August 26th-29th, 2017

Stoweflake Resort and Conference Center
Stowe, Vermont

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?***
Antonie Kline

2:30-3:15 PM: Break

Session 3

Disorders of Transcriptional Regulation III:
Moderator: Antonie Kline

3:15 PM: ***Identification of a Defining Peripheral Blood DNA Methylation Signature of Kabuki Syndrome Enables Kmt2d Variant Classification***
Charles Schwartz

3:30 PM: ***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

3:45 PM: ***De Novo Variants in Kmt2e, a Candidate Haploinsufficient Gene, are a Novel Cause of Intellectual Disability***
Anne O'Donnell-Luria

4:00 PM: ***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

4:15 PM: ***The Genetic Landscape of Cerebellar Malformations***
William Dobyns

4:30 PM: ***Tuba1a: Outcome of Mosaicism and Phenotypic Analysis***
Marc Williams

4:45 PM: ***Phenotypic Spectrum of Females Carrying Mutations in Zc4h2***
Michael Bamshad

6:30-8:00 PM: **Dinner**

8:00 -10:00 PM: **Poster Session**

Sunday, August 27th

7:00-8:00 AM: **Breakfast**

Session 4

8:00-8:15: **Announcements**
Introduction: Robert Lipinski

8:15-9:15 AM: **Invited Speaker: Edward Morrisey, Perelman School of Medicine at The University of Pennsylvania, Philadelphia, PA.**
"New Insights into Lung Epithelial Ontogeny and its Impact on Postnatal Development and Regeneration"

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

12:00 -1:15 PM **Lunch**

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 Bhoj

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

4:15 PM: ***A Recognizable Phenotype Including Macrocephaly, Ligamentous Laxity and Developmental Delay is Associated with Germline De Novo Taok1 Variants***
Karen Gripp

4:30 PM: ***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

4:45 PM: ***Sonic Hedgehog Signaling Targets Foxf2 During Upper Lip Morphogenesis and Cleft Lip Pathogenesis***
Robert Lipinski

5:00 PM: ***Adamtsl1 Mutations Cause Habsburg Jaw or Mandibular Prognathism***
Nik Kantaputra

6:30-8:00 PM: **Dinner / Hot Air Balloon Rides**

8:00 -10:00 PM: **Entertainment**

Monday, August 28th

7:00-8:00 AM: **Breakfast**

Session 8

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

8:00-8:15 AM: **Announcements**

Introduction: Paul Kruszka
8:15-9:15 AM: **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 Omodysplasia Phenotypes*** **Rolf Stottmann**
4:15-4:30 PM:	**Trainee Awards**
6:30-8:30 PM:	**Dinner**
8:30PM-12:00AM:	**Closing Party**