# Workshop Schedule

## Friday, August 27th

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| Registration: | 4:00-6:30 pm |
| Dinner | 6:30-8:00 pm |
| Reception | 8:00-10:00 |

## Saturday, August 28th

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| 7:00-8:15 am | **Breakfast:** |
| 8:15am | **Welcome and Introductions** |
| 8:30-10:15 am | **Session 1: Novel strategies to understand the causes/mechanisms of birth defects** (Moderator: Angela Lin) |
| 8:30 am | Les Biesecker - Using massively parallel sequencing technologies to provide improved molecular delineation of human malformation syndromes |
| 9:15 am | Bamshad - Exome sequencing identifies a gene for Kabuki syndrome |
| 9:30 am | Boycott - Next-Generation sequencing strategies give insight into the mechanism of birth defects in a Canadian isolated population |
| 9:45 am | Bleyl - Comparison of pooled allelic ratios (CoPAR) analysis: an efficient method for mapping genetic traits in extended pedigrees |
| 10:00 am | Allanson – Nablus mask-like facial syndrome and blepharo-naso-facial syndrome are the same entity. Refinement of the critical region of chromosome 8q22.1 points to a potential candidate gene |
| 10:15-10:45 am | **BREAK** |
| 10:45-12:00 pm | **Session 2: Novel strategies to understand the causes/mechanisms of birth defects (cont.)** (Moderator: Mike Bamshad) |

10:45 am Krantz - Applying novel genomic tools towards understanding an old chromosomal diagnosis: Using genome-wide expression and SNP genotyping to identify the true cause of Pallister-Killian syndrome

11:00 am Paciorkowski - Bioinformatic analysis of published and novel copy number variants suggests candidate genes and networks for infantile spasms

11:15 am Bernier - Identification of a novel *Fibulin 4* mutation in a long-term survivor with autosomal recessive cutis laxa (ARCL): the importance, power and methodology of incorporating obligate carrier relatives in homozygosity mapping projects

11:30 pm Muenke – Analysis of novel holoprosencephaly risk alleles identified through a bioinformatics approach

11:45 pm Bernstein – Modeling the cardiac and central nervous system pathophysiology of Timothy syndrome using human induced pluripotent stem cells

12:00-1:30 pm **Lunch**

1:30-3:00 pm **Session 3: Clinical I** (Moderator: Melissa Parisi)

1:30 pm Manning - Proposed revision of the "Dysmorphology Score" in the assessment of fetal alcohol spectrum disorders

1:45 pm McPherson - Stillbirth: A multifactorial problem

2:00 pm Bowdin - Chromosome microarray abnormalities in fetuses with sonographic diagnosis of isolated micrognathia

2:15 pm R. Stevenson - Sirenomelia sans sirenomelia

2:30 pm Mark - Unraveling the mystery of umbilical cord coiling

2:45 pm Cassidy - Infertility in Prader-Willi syndrome?: A normal son born to a 27 year old with PWS

3:00-3:30 pm **BREAK**

3:30-5:15 pm **Session 4: Disorders of the limbs/skeleton** (Moderator: Bryan Hall)

3:30 pm Beck - Defects of embryonic myosin in Freeman-Sheldon syndrome cause reduced force and prolonged relaxation of skeletal myofibers

3:45 pm Gurnett - Evidence that *PITX1* haploinsufficiency causes clubfoot in humans and mice

4:00 pm J. Hall - Amyoplasia - revisited

4:15 pm D. Stevenson - Comparison of bone resorption and the skeletal phenotype in Costello syndrome, CFC syndrome, and Noonan syndrome

4:30 pm M. Jones - Clinical outcomes in children with achondroplasia
4:45 pm Elliot - Insights gained from angiography in the etiology and patterning of limb vascular disorders
5:00 pm A. Hunter – Human equivalent of mouse Disorganization: fact or fantasy?

6:00-7:30 pm **Dinner**

7:30-9:00 pm **DAVID SMITH FELLOWS SESSION** (Moderator: Jim Hanson)

**Speakers:** Ken Jones
Bryan Hall
Roger Stevenson
Jim Hanson

## Sunday, August 29th

7:00-8:30 am **Breakfast**

8:30-10:30 am **Session 5: Genetic and environmental risk factors for birth defects** (Moderator: Cindy Moore)

8:30 am Randy Jirtle - Epigenetics, imprinting, and the developmental origin of disease
9:15 am Carey - Mycophenolate mofetil embryopathy: natural history and follow-up of original cases
9:30 am Safina - Disorganization spectrum with amniotic constriction rings and consideration of past teratogen or environmental exposures
9:45 am Rasmussen – Maternal use of venlafaxine and risk for birth defects
10:00 am Sadler - Heart development, serotonin, and SSRI-induced heart defects
10:15 am Moore - Assisted reproductive technologies and multiple congenital anomalies

10:30-11:00 am **BREAK**

11:00–12:30 pm **Session 6: Genetic and environmental risk factors for birth defects (cont.)** (Moderator: Sonja Rasmussen)

11:00 am Finnell - Gene-nutrient-environment Interactions and risk for neural tube defects
11:15 am Palacios - Folate receptor α autoantibodies and risk of complex birth defects

11:30 am Luquetti - Prevalence analysis of 1,379 cases of isolated microtia from South America

11:45 am Feldkamp - Is gastroschisis really an isolated sporadic defect? Familial cases of gastroschisis in Utah, 1997-2008

12:00 pm Keppler-Noreuil - Clinical and risk factor analysis of cloacal exstrophy in the National Birth Defects Prevention Study (NBDPS)

12:15 pm Niyazov - Cloacal dysgenesis sequence: elucidating the mechanism of multiple malformations using fetal imaging, genetic testing and autopsy

12:30-2:00 pm **Lunch**

2:00-3:30 pm **Session 7: Adults with dysmorphic syndromes** (Moderator: Toni Kline)

2:00 pm C. Stevens - Adults with Rubinstein-Taybi syndrome

2:15pm Hopkin - Dysmorphic syndromes in adults presenting for genetic services.

2:30pm Schrander-Stumpel - Ageing in Prader-Willi syndrome

2:45pm Clericuzio – Mechanisms of adult disease in WAGR Syndrome

3:00pm Pyeritz - Aortopathy Is a common manifestation of Fabry disease

3:15pm Robinson - Outer canthal distance in young adults with fetal alcohol syndrome: preliminary report

3:30-4:00 pm **BREAK**

4:00-6:00 pm **Session 8: Molecular basis of disease** (Moderator: Max Muenke)

4:00 pm Rope - The Kelley phenotype: a novel X-linked multiple congenital anomaly syndrome associated with an *EBP* mutation

4:15 pm Deardorff - Overlapping disorders of cohesin

4:30 pm Dyment - Mutations of *PPARG* cause a congenital, generalized lipodystrophy: insight from follow-up of an adult patient

4:45 pm Sherr - The schizophrenia gene, *DISC1*, and callosal agenesis: disruption of cerebral connectivity as a cause of neurobehavioral disorders

5:00 pm Jezewski - Variable penetrance, expressivity and pleiotropy of *MSX1* alleles

5:15 pm Mefford - Array CGH in patients with single suture craniosynostosis reveals duplication of *RUNX2* in two affected cousins

5:30 pm Hogue - Homozygosity for a *FBN1* missense mutation in an adult patient with atypical features of Marfan syndrome

5:45 pm Moog - Homozygous Loss of *CHRNA7* on Chromosome 15q13.3 causes severe encephalopathy with seizures and hypotonia

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

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

## Monday, August 30th

7:00-8:30 am **Breakfast**

8:30-10:15 am **Session 9: Disorders of Sensory Perception**
(Moderator: Jeff Golden)

8:30 am Steve Wooding - Genetics of taste receptors
9:15 am Dugan – Anosmia / hyposmia segregates with the LADD syndrome in family of a proband with a novel *FGF10* mutation
9:30 am Kline - Workings of the inner ear and sinuses in Cornelia de Lange syndrome
9:45 am Pineda-Alvarez - A wide range of ophthalmologic anomalies are part of the broad holoprosencephaly spectrum
10:00 am Slavotinek – A new family with Manitoba-Oculo-Tricho-Anal (MOTA) syndrome and mutations in *FREM1*

10:15-10:45 am **BREAK**

10:45–12:30 pm **Session 10: The face in dysmorphic syndromes**
(Moderator: John Carey)

10:45 am Shriver - Does exaggerating normal range gene effects recapitulate syndromic facies?
11:00 am Lipinski – Examination of the face-brain dysmorphology patterns in a mouse model of hedgehog signaling antagonist-induced cleft lip and palate
11:15 am Martin - Cheiloscopy and clefting – now we're really getting a groove on
11:30 am Boyd – Cranio-lenticular-sutural dysplsia and other craniofacial syndromes caused by endoplasmic reticulum export defects
11:45 am Bird – Nablus mask-like facial syndrome: attempting genotype-phenotype correlations

12:00 pm K. Evans - Oculoauriculofrontonasal syndrome: new pathognomonic nasal anomalies in an old syndrome

12:15 pm Gripp – Dolichocilia and loose anagen hair in Costello syndrome: cell type specific effects of rasopathy causing mutations?

12:30-2:00 pm **Lunch**

2:00-3:30 pm **Fellows Session**

1. Preparing and submitting a NIH grant: Jim Hanson, Melissa Parisi
2. Navigating the editorial process: John Carey

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

8:00-10:00 pm **Unknowns and Interesting Knowns Session**

## Tuesday, August 31st

7:00-8:30 am **Breakfast**

8:30-10:15 am **Session 11: Race, ethnicity, and birth defects**
(Moderator: Holly Ardinger)

8:30 am Laura Arbour - First Nations communities in Canada and risk for birth defects/genetic diseases

9:15 am Innes - The influence of a unique genetic history on birth defects in the Hutterite population

9:30 am Leppig – Clinical evaluation of Vietnamese patients with developmental disabilities: lessons and challenges using dysmorphology evaluations and genomic testing

9:45 am Seaver – Brittle bones in babies: osteogenesis imperfecta (OI) in Asian and Pacific Islanders in Hawai'i

10:00 am Shalev - A carrier screening program for severe and frequent genetic diseases in the Arab population of Israel

10:15-10:45 pm **Break**

10:45 – 12:30 pm **Session 12: Central Nervous System**
(Moderator: Steve Bleyl)

10:45 am Dobyns - Gomez-Lopez-Hernandez and VACTERL-H syndromes account for a majority of patients with rhombencephalosynapsis

11:00 am Golden - Differential effects of a polyalanine tract expansion in *Arx* on neural development and gene expression

11:15 am Chitayat - Posterior fossa abnormalities in patients with Beckwith-Wiedemann syndrome

11:30 am Dinulos - Vanishing white matter disease: it may scare you to death

11:45 am Doherty – Absent cerebellum with turricephaly, aqueductal stenosis, bitemporal alopecia and bilateral corneal scarring: Gomez-Lopez-Hernandez syndrome or something new?

12:00 pm Millen - Characterization of a novel Dandy-Walker malformation locus on chr. 3p14

12:15 pm Mirzaa - MPPH_CM (megalencephaly-polymicrogyria-polydactyly-hydrocephalus-capillary malformation): revised spectrum and overlap with two familial syndromes

12:30-2:00 pm **Lunch**

2:00-3:30 pm **Session 13: Cytogenetics**
(Moderator: Kathy Leppig)

2:00 pm K. Jones - Submicroscopic deletion of 9q34.3 disrupting *EHMT1* gene in a patient with acrocallosal syndrome

2:15 pm Keegan - Comprehensive genetic analysis of OEIS complex reveals no evidence for a recurrent microdeletion or duplication

2:30 pm Lauzon - Array CGH elucidation of a complex chromosome 21 rearrangement in a child with syndromic thrombocytopenia and predisposition to acute myelogenous leukemia

2:45 pm Muller - Deletion 9q22.3: Gorlin syndrome plus metopic craniosynostosis, macrosomia, and developmental delay

3:00 pm Richer - A family with microdeletion of 2q14 associated with IUGR, OFC at the 2nd%, cognitive impairment and hypercoagulability

3:15 pm Graham - Distinctive phenotype in 8 patients with deletion of chromosome 1q24-q25

3:30-4:00 pm **BREAK**

4:00-6:00 pm **Session 14: Clinical II**
(Moderator: Judith Allanson)

4:00 pm Chung - In-vitro treatment with pravastatin and dexamethasone normalizes abnormal deposition of elastic fibers in dermal fibroblasts derived from patients with restrictive dermopathy - possible therapeutic implications

4:15 pm B. Hall - Novel *SCY1BP1L* gene mutation and Arnold-Chiari malformation I in a case of geroderma osteodysplastica

4:30 pm J. Evans - Cervical vertebral fusions (Klippel-Feil Anomaly) in Manitoba: differences in aboriginal and non-aboriginal populations

4:45pm Fernandez - A consanguineous Newfoundland family has a provisionally unique hypomelanosis disorder that most closely resembles Hermansky-Pudlak syndrome.

5:00 pm Cunningham - Rare Insulin-Like growth factor 1 receptor variants associated with isolated single suture craniosynostosis

5:15 pm Haldeman-Englert - Genes and biological processes disrupted in rare developmental delay and multiple congenital anomaly syndromes

5:30 pm Siebert - The application of optical projection tomography to the study of bladder outlet obstruction

5:45 pm Sanchez-Lara - Sleep problems in children with Sotos syndrome

6:00 pm Closing Comments

6:30 pm **Dinner (Party)**

## Wednesday, September 1st

7:00–9:00 am **Breakfast**

**Departure**