11th Biennial International 22q11.2 Conference

"Celebrating 20 Years of Education and Collaboration"

July 11 – 13, 2018

Whistler, British Columbia, Canada

Day 1 July 11th

7:00 AM **Registration Opens**

8:00 AM ***Welcome from the 22q11.2 Society***
Peter Scambler and Donna McDonald-McGinn

8:10 AM ***Welcome from the Local Arrangements Committee***
Anne Bassett, Jacob Vorstman and Pooja Panwar

8:15 AM ***Official Meeting Open***

8:20 AM ***2018 Angelo DiGeorge Memorial Medal of Honor Presentation***
2016 Recipient Ann Swillen *- Presenting*

Session I: ***Detection - Care - Collaboration***

Invited Speaker: Bedside to Bench and Back Again – A Caregiver's Tale

8:30 AM ***Anne Bassett,*** *University of Toronto*
Toronto, Canada

8:45 AM **Q&A**

Submitted Papers: Detection

8:50 AM ***McDonald-McGinn, DM***
22q and Two – 22q11.2 Deletion and Coexisting Conditions

9:00 AM ***Harr, M***
Screening for 22q11.2 Deletions and Duplications in a Large Biobank Data-Set at a Tertiary Medical Center Uncovers New and Undiagnosed Cases

9:10 AM ***Demko, Z***
Analytical Validation of a SNP-Based Non-Invasive Prenatal Test to Detect the Fetal 22q11.2 Deletion in a Cohort of Maternal Plasma Samples

9:20 AM ***Maisenbacher, M***
Detection of Mothers at Risk for the 22q11.2 Deletion by NIPT Highlights Lack of Maternal and Fetal Confirmatory Testing

9:30 AM **Q&A**

Submitted Papers: Care

9:40 AM ***Bailey, A***
Multidisciplinary Care for Patients with Chromosome 22q11.2 CNVs

9:45 AM ***Hickey, S***
The Impact of Interdisciplinary Team Care on Adherence to Clinical Care Guidelines in Children with 22q11.2DS

9:50 AM ***Kelman, C***
Psychosocial Needs Necessitating Social Work Intervention for Patients and Families Affected by the 22q11.2 Deletion and Duplication Syndromes

9:55 AM **Q&A**

10:00 AM **Coffee Break**

Submitted Papers: Collaboration

10:30 AM ***McGinn, MJ****
Current Care Practices For 22q11.2 Deletion and Duplication Syndromes across the Americas: Towards Establishing A Formal Network

10:36 AM ***Kitamura, C***
Exploring Support Needs of Individuals and Families Affected by 22q11.2DS in Japan

10:40 AM ***Shiozaki, A***
The Need for User-Oriented "The Japanese Version Original Care Guide for 22q11.2DS" From Patient and Family Perspectives

10:44 AM ***Loock, C***
Quality Improvement and Research Collaboration Opportunities in British Columbia and Canada Using Data Linkages for Patients and Families Living with 22q11.2DS

10:48 AM **Q&A**

Session II: ***On the Road to Improving Care via Novel Interventions***

Invited Speaker: Transcriptional Mechanisms and Phenotypic Rescue

10:55 AM ***Antonio Baldini***
Institute of Genetics and Biophysics, National Research Council, University Federico II Naples, Italy

11:25 AM **Q&A**

11:35 AM ***Sebastiano, V***
In Vitro Modeling of 22q11 Endodermal Anomalies Using Human Pluripotent Stem Cells: Understanding the Disease to Develop a Cure

11:45 AM ***Weinacht, K***
Regenerative Thymic Tissues as Curative Cell Therapy for Children with 22q11DS and Severe T Cell Immunodeficiency

11:55 AM **Q&A**

Invited Speaker: Intervention and Treatment Affecting Brain and Behavior

12:05 PM ***Elizabeth Illingworth***
Institute of Genetics and Biophysics, National Research Council
Naples, Italy

12:35 PM **Q&A**

12:45 PM ***Gothelf, D***
A Meta-Analysis of Psychiatric Treatments in 22q11DS

12:50 PM ***Vingerhoets, C***
Gaba and Glutamate in Patients with 22q11.2DS and Healthy Volunteers and the Relation with Cognition: A Randomized Double-Blind 7Tesla Pharmacological MRS Study

1:00 PM ***Armando, M***
Omega-3 Polyunsaturated Fatty Acids Improve Neurocognitive Functions and Reduce the Conversion Rate to Psychosis in Patients with 22q11.2DS

1:10 PM ***Meechan, D***
A Developmental/Molecular Mechanism and Targeted Therapy for Cognitive Disruption in 22q11.2DS

1:20 PM **Q&A**

1:30 PM **Lunch and Poster Viewing**

Session III: ***The Classics (per DiGeorge) – Immune, Cardiac, and Endocrine***

Invited Speaker: Immunology as a Window into Understanding 22q11.2 Issues

2:15 PM ***Kathleen Sullivan***
Children's Hospital of Philadelphia and the Perelman School of Medicine at the University of Pennsylvania
Philadelphia, PA, USA

2:45 PM **Q&A**

Submitted Papers: Thymus

2:55 PM ***Dejene, B***
The Thymus in 22q11.2DS

3:05 PM ***Crowley, TB****
Variable Immune Deficiency Related to Deletion Size in Chromosome 22q11.2DS

3:15 PM ***Framme, J***
Long Term Follow-Up of Patients with 22q11DS and Low Trecs in the Newborn Period

3:21 PM ***Schindewolf, E****
Prenatal Thymus Size Analysis Predicting T Cell Count in the First Year of Life

3:25 PM **Q&A**

Submitted Papers: Heart

3:35 PM ***Mastromoro, G***
Left Pulmonary Artery in 22q11.2DS: Echocardiographic Findings in Humans and Expression of Tbx1 and Knockout Mice

3:45 PM ***Scambler, P***
Tbx1 Is Required for Vagal Innervation of the Heart

3:55 PM ***Amengual-Cladera, E***
Vitamin A Supplementation in the Diet, but Not Deficiency, Modulates the Incidence of Congenital Heart Defects in a 22q11ds Mouse Model

4:05 PM **Q&A**

4:15 PM **Poster Session with Authors (Odd Numbers) and Afternoon Tea**

Submitted Papers: Cardiac and Endocrine

5:00 PM ***Jeong, S****
22q11.2 Duplication: An Important Cause of Hypoplastic Left Heart Syndrome

5:10 PM ***van Mil, S***
Late Mortality in a Genetic Subtype of Tetralogy of Fallot

5:20 PM ***Lambert, M***
Evaluation of Bleeding Risk with Cardiac Surgery in 22q11.2DS: A Case Control Study

5:30 PM **Q&A**

5:40 PM ***Katz, L***
Association between Cardiac Surgery and Hypocalcemia in 22q11.2DS

5:50 PM ***Boot, E***
Endocrine Alterations in Adults with 22q11.2DS

6:00 PM ***Houben, M***
Growth in Dutch Children with 22q11.2 Deletion Syndrome - Construction of Reference Growth Charts and Analysis of Determinants of Growth

6:10 PM **Q&A**

6:20 PM **Adjourn**

8:00 PM ***Poolside and Ping Pong Reception***

***** - Indicates Junior Investigators***

Day 2 July 12th

7:30 AM **Registration Open**

8:00 AM ***20th Anniversary Special Service Award Presentation***
Peter Scambler and Anne Bassett – *Presenting*

Session IV: ***Major Associated Systems***

Invited Speaker: CRKL1 and Mammalian Structural Birth Defects

8:10 AM ***Bernice Morrow***
Albert Einstein College of Medicine
New York, NY, USA

8:25 AM **Q&A**

Submitted Papers: A Multisystem Condition

8:30 AM ***Adetunji, M***
Immature Platelet Fraction is Elevated in Individuals with 22q11.2DS Even with Near Normal Platelet Counts

8:40 AM ***Homans, J****
Scoliosis in Association with the 22q11.2DS

8:45 AM ***Homans, J****
The Surgical Outcome of Scoliosis Surgery within the 22q11.2DS

8:50 AM ***Mascarenhas, M***
Exploring the Gastrointestinal Phenotype in 22q11.2DS

9:00 AM ***LaMantia, A***
A Neurodevelopmental Basis for Perinatal Feeding and Swallowing Disorders in 22q11.2

9:10 AM **Q&A**

9:20 AM ***Paronett, E***
Ranbp1 Haploinsufficiency Contributes to Cranial Neural Crest Anomalies in 22q11.2DS

9:30 AM ***Jackson, O***
Palatal Abnormalities in 22q11.2DS

9:36 AM ***Kellogg, B***
Revision Speech Surgery in Patients with 22q11.2DS: A Retrospective Review of Speech Outcomes

9:42 AM ***de la Mar, A***
Comparison of Speech Outcome after Pharyngoplasty in 22q11.2DS: Cranial Based Pharyngeal Flap versus the Modified Honig Procedure

9:50 AM **Q&A**

10:00 AM **Coffee Break**

Submitted Papers: Hearing and Language

10:30 AM ***Willaert, A***
Homozygous Tbx1 Missense Mutation in Mice Causes Complete Hearing Loss

10:40 AM ***Verheij, E***
Anatomical Malformations of the Middle and Inner Ear in 22q11.2DS

10:50 AM ***Elden, L***
A Cohort Study: Indications and Outcomes of Otolaryngologic Surgeries in Pediatric Patients with 22q11.2DS

11:00 AM **Q&A**

11:10 AM ***Smith, R****
22q11.2 LCR22D-LCR22E Distal Deletion: Findings in One Illustrative Family

11:20 AM ***Boerma, T***
Language Impairment in the 22q11.2DS

11:30 PM ***Solot, C***
Are Language Scores an Early Predictor of Cognitive Decline?

11:40 AM **Q&A**

11:50 AM ***Invited Speaker: Hippocampal-Prefrontal Miscommunication and Cognitive Deficits***
David Kupferschmidt
National Institute of Health
Bethesda, MD, USA

12:20 PM **Q&A**

Submitted Papers – Cognition, Intellect and More

12:30 PM ***McGinn, DE****
Maternal Origin of Familial 22q11.2 Deletions Negatively Impacts FSIQ Scores

12:40 PM ***Breetvelt, E***
A Normative Chart for Cognitive Development in 22q11DS: Implications for 22q11DS and Beyond

12:50 PM ***Fiksinki, A****
The Impact of Parental IQ on the Variable Penetrance of Intellectual Impairment in 22q11DS

1:00 PM ***Van den Heuvel****
Comparison of Cognitive Abilities and Social Responsiveness Skills in Children with 22q11.2DS and Children with Idiopathic Intellectual Disability

1:10 PM ***Hooper, S***
The Relationship of Intellectual Functions to Psychosis and Subthreshold Psychotic Symptoms in Individuals with 22q11.2DS Using Latent Profile Models

1:20 PM **Q&A**

1:30 PM **Lunch and Poster Viewing**

Session V: ***Developmental Trajectories and Psychotic Illness***

2:15 PM ***Invited Speaker: Micro-RNA and 22q11.2DS***
Stanislav Zakharenko
St. Jude Children's Research Hospital
Memphis, TN, USA

2:45 PM **Q&A**

Submitted Papers – The Developmental Continuum of 22q11.2DS

2:55 PM ***Earls, L***
The Impact of the Aging Epitranscriptome on Neurophysiologic Phenotypes of 22q11.2DS

3:05 PM ***Gur, R***
The International 22q11.2DS Brain Behavior Consortium: Challenges and Opportunities

3:15 PM ***Fiksinski, A****
Trajectories of Processing Speed and Risk for Psychotic Disorders in 22q11DS: A Longitudinal Study

3:25 PM **Q&A**

3:35 PM ***Pontillo, M***
Neurocognitive Profile and Onset of Psychosis in Children, Adolescents and Young Adults with 22q11DS: A Longitudinal Study

3:45 PM ***Kates, W***
Longitudinal Trajectories of Psychiatric Diagnoses and Predictors of Persistence in Youth with 22q11.2DS

3:55 PM ***Schneider, M***
Longitudinal Evolution of Negative Symptoms in 22q11.2DS and Predictive Value for Transition to Psychosis

4:05 PM **Q&A**

4:15 PM **Poster Session with Authors (Even Numbers) and Afternoon Tea**

5:00 PM ***Gur, R***
The Evolution of Psychosis in 22q11.2 Deletion Syndrome: Risk and Resilience

5:10 PM ***DuBourg, L****
Visual Processing of Complex Social Scenes in 22q11.2DS: Relevance for Social Impairments?

5:20 PM ***McCabe, K***
A Pilot Study Characterizing the Social Impairment Phenotype of Children with 22q11.2DS and Children with Idiopathic Autism Spectrum Disorder

5:24 PM ***Vergaelen, E****
Differences in Self-and Parent Reporting and the Relation with an At Risk State in Adolescents and Adults with 22q11.2DS

5:30 PM **Q&A**

5:40 PM ***van Duin, E****
Stress Reactivity, Cortisol Levels and Experience Sampling in Adults with 22q11.2DS

5:50 PM ***Armando, M***
Coping Strategies Mediate the Effect of Stressful Life Events on Schizotypal Traits and Psychotic Symptoms in 22q11.2 Deletion Syndrome

6:00 PM ***Angkustsiri, K***
Parenting, Anxiety and Adaptive Function in Children with Chromosome 22q11.2DS

6:10 PM **Q&A**

6:20 PM **Adjourn**

8:00 PM ***Gala Dinner: Squamish and Lil'wat Nations Cultural Centre***




***** - Indicates Junior Investigators***

Flashback to Sirmione '16

Day 3 July 13th

7:30 AM **Registration Open**

8:00 AM ***22q11.2 Society Website Update – Joanne Loo***

8:05 AM ***2018 Unsung Hero Award Presentation***
2016 Recipient Maria Kamper - *Presenting*

Session VI: ***Genetics and Brain Expression***

Invited Speaker: Measuring and Predicting the Effect Size of Non-Recurrent CNVs on Cognitive and Behavioral Traits

8:15 AM ***Sebastien Jacquemont***
University of Montreal
Montreal, Canada

8:45 AM **Q&A**

Submitted Papers: Genetic Variants and Risk

8:55 AM ***Breetvelt, E***
Burden of Rare Coding Variants in the 22q11.2 Deletion Region is Associated with Educational Attainment and Schizophrenia Risk in Two General Population Cohorts

9:00 AM ***De Borre, M****
Contribution of Rare Hemizygous Variants to Phenotypic Variability in 22q11.2DS

9:10 AM ***Lin, J***
Integrated Genome-Wide Analyses of Rare Variants for Schizophrenia Risk in 22q11.2DS

9:20 AM ***Niarchou, M****
Genetic Risk for Schizophrenia and Development of Anxiety Disorders and Negative Symptoms in 22q11.2DS

9:30 AM **Q&A**

Submitted Papers: Associated Neurologic Features

9:40 AM ***Chadehumbe, M***
Neurologic Challenges in 22q11.2DS

9:45 AM ***Eaton, C****
Epilepsy and Seizures in Young People with 22q11.2DS: Prevalence and Links with Neurodevelopmental Disorders

9:50 AM ***Vecchio, D****

Intellectual Disability, Autism Spectrum Disorder and Seizures Due to 22q11.2-q11.23 Microduplications: Clinical and Molecular Characterization of a New Neurodevelopmental Disorders Genetic Driver

9:55 AM ***Moulding, H****

Sleep Problems and the Relationship With Psychiatric and Neurodevelopmental Difficulties in Young People with 22q11.2DS

10:05 AM **Q&A**

10:15 AM **Coffee Break**

Submitted Papers: Understanding Parkinson's and Motor Findings

10:45 AM ***Boot, E***

Parkinsonian Motor Features in Adults with 22q11.2DS

10:55 AM ***Morrison, S****

Using Objective Measures of Sensorimotor Control to Improve Our Understanding of Motor Difficulties and the Links with Other Neurodevelopmental Problems in 22q11.2DS

11:01 AM ***Fisher, M***

Anxiety Phenotypes and Biomarkers for Parkinson's disease in 22q11.2DS

11:11 AM ***Repetto, G***

Analysis of Prodromal Manifestations of Parkinson's disease in Adults With 22q11.2DS

11:15 AM **Q&A**

Submitted Papers: Viewing the Brain from Every Angle

11:25 AM ***Bearden, C***

The Enigma 22q11.2DS Working Group: Insights into Neurodevelopment and Psychosis

11:35 AM ***van Duin, E****

Frontal Dopamine D2/3 Receptor Binding in Adults with 22q11.2DS: A [18F] Fallypride Positron Emission Tomography Study

11:45 AM ***Rogdaki, M***

The State or Trait Component of Dopamine and Glutamate Dysfunction in the Risk for Psychosis: An In Vivo Multimodal Imaging Study of Individuals with 22q11.2DS

11:55 AM **Q&A**

12:05 PM ***Gudbrandsen, M***
The Neuroanatomy of Autism Spectrum Disorder in 22q11.2DS

12:15 PM ***Doherty, J***
Excitatory-Inhibitory Balance in 22q11.2DS: A Pilot Magnetic Resonance Spectroscopy and Magnetoencephalography Study

12:21 PM ***Murphy, C***
Developing Protocols to Enable MRI Brain Scanning in Infants and Young Children with 22q11.2DS

12:25 PM **Q&A**

Session VII: ***Looking Ahead***

12:35 PM ***Invited Speaker: The Future of Genomics - Impact on 22q11.2DS and Vice Versa***
Christian Marshall
University of Toronto
Toronto, ON, Canada

1:05 PM **Q&A**

1:15 PM **Lunch and Poster Viewing**

2:00 PM ***Submitted Papers: Adults and Outcomes***
Heung, T*
Predictors of All-Cause Mortality in Adults with 22q11.2DS

2:10 PM ***Malecki, S***
Medical Multimorbidity in Adults with 22q11.2DS

2:20 PM ***Loo, J****
Personalized Medical Information Cards for Adults with 22q11.2DS

2:30 PM ***Gothelf, D****
Education and Employment Trajectories from Childhood to Adulthood in Individuals with 22q11.2DS

2:40 PM ***Goldenberg, P***
Functional Outcomes in 27 Adults with 22q11.2DS

2:45 PM ***Palmer, L***
Identifying Issues Related to Sexual Health in Adults with 22q11.2DS

2:55 PM **Q&A**

Session VIII: ***The Future of Clinical and Genetic Research***

3:05 PM ***Invited Speaker: Mechanisms of 22q11.2 Deletions and Duplications***
Joris Vermeesch
KU Leuven
Leuven, Belgium

3:35 PM **Q&A**

3:45 PM ***Siu, M***
Elucidating Pathophysiology using Genome-Wide DNA Methylation Analysis

3:55 PM ***Urban, A***
Haplotype Specific Analysis of Chromosome 22q Folding Patterns in 22q11.2DS

4:05 PM ***Xie, M***
Variation in the Frequency of an Inversion Polymorphism May Affect the Prevalence of the 22q11.2DS amongst Populations

4:15 PM **Q&A**

4:25 PM ***Invited Speaker: CHD7 and Epigenetics***
Peter Scambler
University College London and Great Ormond Street Institute of Child Health
London, UK

4:40 PM **Q&A**

Late Breaking Submitted Papers: Discoveries Every Minute

4:50 PM ***TBA***

5:00 PM ***TBA***

5:10 PM ***TBA***

5:20 PM ***TBA***

5:30 PM **Q&A**

5:40 PM ***Unknown/Difficult Cases – Audience Participation (2 slides/case)***
Limited Number – Please email intent to present in advance and load during the break

6:10 PM **Junior Investigator Award**

6:15 PM **Closing Remarks and Announcement of Future Meeting**

6:30 PM **Adjourn**

***** - *Indicates Junior Investigators***

Poster Presentations:

***** - Indicates Junior Investigator*** **+ - Indicates Top Scoring Poster***

- *Odd numbered posters – authors present on Wednesday (July 11th)*
- *Even numbered posters - authors present on Thursday (July 12th)*

1. ***Jelsema, R***
   Non-Invasive Prenatal Testing for Fetal 22q11.2DS

2. ***Russo, J**** ***+***
   Impact of Assisted Reproductive Technology on Prevalence and Associated Features in 22q11.2DS

3. ***Arganbright, J***
   Caring for Children with 22q11.2DS: Current State of 22q Multidisciplinary Team Clinics

4. ***Van Lue, M***
   Use of A Single Access Data Portal to Analyze Demographic and Utilization Data of Patients with 22qDS Treated at a Tertiary-Care Pediatric Hospital from 2013-2017

5. ***Kamper, M***
   22q Coordination Portal and App

6. ***Johns, A***
   Care for Patients with 22q11.2DS within and Outside a Craniofacial Team

7. ***Hall, C***
   Strategies for Delivering Coordinated Interdisciplinary Team Care to Patients with 22q11.2DS

8. ***Lawlor, A***
   Developing Integrated Care in the Context of Rare Chromosomal Conditions: 22q11.2DS – A Parent/Clinician Collaboration. The Irish Story

9. ***Ruzzi, S***
   Challenges in Providing Comprehensive Care for Patients with 22q11.2DS Living in Geographic/Cultural Isolates – A Case Report

10. ***Goldenberg, P***
    Use of Social Media Targeting Patients and Families Changes National and Global Health Care Outcomes for People with Chromosome 22 Conditions

11. ***Gonzalez-Gandolfi, C**** [+]
    Can Maternal Diabetes Exacerbate Phenotypic Features in Patients with 22q11.2 Copy Number Variants?

12. ***Patel, P***
    Bleeding Phenotype in Children with 22q11DS – Preliminary Results from a Cross-Sectional Study

13. ***Madhoun, L***
    Dysphagia in Young Children with 22q11.2DS

14. ***Tanner, A***
    The Role of the Speech-Language Pathologist in Craniofacial Team Care for Patients with 22q11.2DS

15. ***Hayakawa, T***
    Language Home Environment of Young Children with 22q11.2DS

16. ***Baylis, A***
Vocal Output and Parent Input: A Lena Study of Speech in Young Children with 22q11.2DS

17. ***Kollara Sunil, L*** [+]
Velopharyngeal Structural and Muscle Variations in Children with 22q11.2DS

18. ***Hartnick, C***
Mobilization of the Carotid Arteries to Allow for Pharyngeal Flap in Children with 22q Anomalies and VPI

19. ***Haenssler, A***
Analysis of the Impact of Cranial Base Abnormalities on Cerebellar Volume and Velopharyngeal Variables Related to Speech in 22q11.2DS

20. ***Cummings, C***
Phonetic and Phonological Analysis of Speech in Infants and Toddlers with 22q11.2DS

21. ***Magee, L***
Pre- and Post-Operative Speech and Psychosocial Functioning in Patients with 22q11.2DS Patients Presenting with Velopharyngeal Dysfunction

22. ***Kragness, S***
Age-Dependent Brain Expression Pattern of a Novel Micropeptide Encoded in the 22q11.2 Deletion Region

23. ***Morrison, S***
Longitudinal Cognitive Development and Association with Prodromal Psychotic Symptoms in Adolescents with 22q11.2DS

24. ***Cutler-Landsman, D***
From Research to Practice--Optimizing the Learning Path for Students with 22q11.2DS

25. ***Parkinson, D*** [+]
The Impact of Prepubescent and Adolescent Socialization on Adult Hippocampal Physiology

26. ***Fiksinski, A*** [*] [+]
Neurocognition and Adaptive Functioning in a Genetic High-Risk Model of Schizophrenia

27. ***Wallin, L***
Mental Health in 22q11.2DS from Childhood to Adult Age: A Prospective Longitudinal Study of 90 Individuals

28. Pontillo, M [+]

Negative Psychotic Symptoms in 22q11.2 Deletion and their Association with the Neuropsychological Profile

29. Francisco, A

Brain Measures of Basic Auditory Processing in Adolescents and Adults with 22q11.2DS

30. ***Van, L*** [*] [+]

Treatment of Schizophrenia in 22q11.2DS

31. Kumakura, Y

Meeting the Diverse Needs of Adolescents and Young Adults with 22q11.2DS

32. D'Arcy, S [*]

Characterizing Cooking Habits and Confidence in Food Skills in Adults with 22q11.2DS

33. Tindale, E

How Do You Feel? A Parent's Perspective

34. Loock, C

Active Patient Involvement in Medical Education at the University of British Columbia: Partnering with 22q11.2DS Families

35. Baskin, J

Mid-childhood adaptive function in individuals with 22q11.2 deletion syndrome is associated with immune-deficiency, but not oral/palatal or cardiac phenotypes