

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** **Gai, H**
Regenerative Thymic Tissues as Curative Cell Therapy for Children with 22q11DS and Severe T Cell Immunodeficiency
- 11:55 AM** Q&A
- 12:05 PM** **Invited Speaker: Intervention and Treatment Affecting Brain and Behavior**
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
- 2:15 PM** **Invited Speaker: Immunology as a Window into Understanding 22q11.2 Issues**
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***

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