

11th Biennial International 22q11.2 Conference

“Celebrating 20 Years of Education and Collaboration”

July 11 – 13, 2018

Whistler, British Columbia, Canada

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

Chadhumbe, 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

Cunningham, A*

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** **Mosheva, M***
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** **Zhang, X**
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	<u>Unknown/Difficult Cases – Audience Participation (2 slides/case)</u> <i>Limited Number – Please email intent to present in advance and load during the break</i>
6:10 PM	Junior Investigator Award
6:15 PM	Closing Remarks and Announcement of Future Meeting
6:30 PM	Adjourn

* - Indicates Junior Investigators