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 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  **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