

# 11<sup>th</sup> Biennial International 22q11.2 Conference

*“Celebrating 20 Years of Education and Collaboration”*

July 11 – 13, 2018

Whistler, British Columbia, Canada

## Day 1

## July 11<sup>th</sup>

7:00 AM

*Registration Opens – Harmony Foyer*

8:00 AM

*Welcome from the 22q11.2 Society – Harmony Ballroom*  
*Peter Scambler and Donna McDonald-McGinn*

8:10 AM

*Welcome from the Local Arrangements Committee – Harmony Ballroom*  
*Anne Bassett, Jacob Vorstman and Pooja Panwar*

8:15 AM

*Official Meeting Opens – Harmony Ballroom*

8:20 AM

***2018 Angelo DiGeorge Memorial Medal of Honor Presentation***  
*2016 Recipient Ann Swillen – Presenting*

**Session I:**

***Detection - Care - Collaboration***

*Chairs: Peter Scambler and Donna McDonald-McGinn*

***Invited Speaker:***

***Bedside to Bench and Back Again – A Caregiver’s Tale***

8:30 AM

**1**

***Anne Bassett***  
*University of Toronto*  
*Toronto, ON, Canada*

***Submitted Papers: Detection***

8:45 AM

**2**

***Donna McDonald-McGinn***  
*22q and Two – 22q11.2 Deletion and Coexisting Conditions*

8:55 AM

**3**

***Margaret Harr***  
*Screening for 22q11.2 Deletions and Duplications in a Large Biobank Data-Set at a Tertiary Medical Center Uncovers New and Undiagnosed Cases*

- 9:05 AM **4** **Zachary Demko**  
*Analytical Validation of a SNP-Based Non-Invasive Prenatal Test to Detect the Fetal 22q11.2 Deletion in a Cohort of Maternal Plasma Samples*
- 9:15 AM **5** **Melissa Maisenbacher**  
*Detection of Mothers at Risk for the 22q11.2 Deletion by NIPT Highlights Lack of Maternal and Fetal Confirmatory Testing*
- 9:25 AM Q&A
- Submitted Papers: Care**
- 9:40 AM **6** **Alice Bailey**  
*Multidisciplinary Care for Patients with Chromosome 22q11.2 CNVs*
- 9:45 AM **7** **Emily Gallagher**  
*Multidisciplinary Care Utilization in Patients with 22q11.2 Deletion Syndrome at Seattle Children's Hospital*
- 9:50 AM **8** **Scott Hickey**  
*The Impact of Interdisciplinary Team Care on Adherence to Clinical Care Guidelines in Children with 22q11.2DS*
- 9:55 AM **9** **Caren Kelman**  
*Psychosocial Needs Necessitating Social Work Intervention for Patients and Families Affected by the 22q11.2 Deletion and Duplication Syndromes*
- 10:00 AM Q&A
- Submitted Papers: Collaboration**
- 10:10 AM **10** **McGinn, MJ\***  
*Current Care Practices For 22q11.2 Deletion and Duplication Syndromes across the Americas: Towards Establishing A Formal Network*
- 10:16 AM **11** **Chiaki Kitamura**  
*Exploring Support Needs of Individuals and Families Affected by 22q11.2DS in Japan*
- 10:20 AM **12** **Christine Loock**  
*Quality Improvement and Research Collaboration Opportunities in British Columbia and Canada Using Data Linkages for Patients and Families Living with 22q11.2DS*

10:25 AM

Q&A

10:30 AM

*Coffee Break – Harmony Foyer*

**Session II:**

***On the Road to Improving Care via Novel Interventions***

*Chairs: Anne Bassett and Ann Swillen*

***Invited Speaker:***

***TBX1 and 22q11.2DS: Transcriptional Mechanisms and Phenotypic Rescue***

11:00 AM

13

**Antonio Baldini**

*Institute of Genetics and Biophysics, National Research Council, University Federico II  
Naples, Italy*

***Submitted Papers: Working Towards a Cure***

11:30 AM

14

**Vittorio Sebastiano**

*In Vitro Modeling of 22q11 Endodermal Anomalies Using Human Pluripotent Stem Cells: Understanding the Disease to Develop a Cure*

11:40 AM

15

**Katja Weinacht**

*Regenerative Thymic Tissues as Curative Cell Therapy for Children with 22q11DS and Severe T Cell Immunodeficiency*

11:50 AM

Q&A

***Invited Speaker:***

***Intervention and Treatment Affecting Brain and Behavior***

12:05 PM

16

**Elizabeth Illingworth**

*Institute of Genetics and Biophysics, National Research Council  
Naples, Italy*

***Submitted Papers: Targeted Therapy***

12:35 PM

17

**Doron Gothelf**

*A Meta-Analysis of Psychiatric Treatments in 22q11DS*

12:40 PM

18

**Therese Van Amelsvoort**

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

- 12:50 PM**      **19**      **Marco Armando**  
*Omega-3 Polyunsaturated Fatty Acids Improve Neurocognitive Functions and Reduce the Conversion Rate to Psychosis in Patients with 22q11.2DS*
- 1:00 PM**      **20**      **Daniel Meechan**  
*A Developmental/Molecular Mechanism and Targeted Therapy for Cognitive Disruption in 22q11.2DS*
- 1:10 PM**      **Q&A**
- 1:30 PM**      **Group Photo followed by Luncheon and Poster Viewing – Harmony Foyer**
- Session III:**      **The Classics (per DiGeorge) – Immune, Cardiac, and Endocrine**  
*Chairs: Antonio Baldini and Kathleen Sullivan*
- Invited Speaker:**  
**Immunology as a Window into Understanding 22q11.2 Related Issues**
- 2:15 PM**      **21**      **Kathleen Sullivan**  
*Children’s Hospital of Philadelphia and the Perelman School of Medicine at the University of Pennsylvania  
Philadelphia, PA, USA*
- Submitted Papers: Thymus**
- 2:45 PM**      **22**      **Beruh Dejene**  
*The Thymus in 22q11.2DS*
- 2:55 PM**      **23**      **Terrence Crowley\***  
*Variable Immune Deficiency Related to Deletion Size in Chromosome 22q11.2DS*
- 3:00 PM**      **24**      **Solveig Oskarsdottir**  
*Long Term Follow-Up of Patients with 22q11DS and Low Trecs in the Newborn Period*
- 3:10 PM**      **25**      **Carmela Giancotta**  
*Autoimmunity and its Association with T Cells and B Cell Subsets in Patients with Del22q11.2 Syndrome*
- 3:15 PM**      **26**      **Agostina Marolda**  
*Immunological Hallmarks of Hematological Autoimmunity in 22q11.2 Deletion Syndrome (22q11.2DS) Patients: a Multicentric Case-Control Study*

3:20 PM

Q&A

3:35 PM **27** **Submitted Papers: Heart**

**Marta Unolt**

*Left Pulmonary Artery in 22q11.2DS: Echocardiographic Findings in Humans and Expression of Tbx1 and Knockout Mice*

3:45 PM

**28** **Peter Scambler**

*Tbx1 Is Required for Vagal Innervation of the Heart*

3:55 PM

**29** **Glen Iannucci\***

*TBX1 Mutation as a Cause of Non-Syndromic Familial Vascular Rings*

4:00 PM

**30** **Damien Heine-Suñer**

*Vitamin A Supplementation in the Diet, but Not Deficiency, Modulates the Incidence of Congenital Heart Defects in a 22q11DS Mouse Model*

4:10 PM

Q&A

4:20 PM

*Poster Session with Authors (Odd #) and Afternoon Tea – Harmony Foyer*

**Session IV:**

**The Classics and More**

*Chairs: Nicole Sarles-Philip and Solveig Oskarsdottir*

**Submitted Papers: Cardiac and Endocrine**

5:15 PM

**31** **Stephanie Jeong\***

*22q11.2 Duplication: An Important Cause of Hypoplastic Left Heart Syndrome*

5:25 PM

**32** **Spencer van Mil\***

*Late Mortality in a Genetic Subtype of Tetralogy of Fallot*

5:35 PM

**33** **Michele Lambert**

*Evaluation of Bleeding Risk with Cardiac Surgery in 22q11.2DS: A Case Control Study*

5:45 PM

**34** **Lorraine Katz**

*Association between Cardiac Surgery and Hypocalcemia in 22q11.2DS*

5:55 PM

**35** **Erik Boot**

*Endocrine Alterations in Adults with 22q11.2DS*

- 6:05 PM      **36**      **Michiel Houben**  
*Growth in Dutch Children with 22q11.2 Deletion Syndrome - Construction of Reference Growth Charts and Analysis of Determinants of Growth*
- 6:15 PM      Q&A
- 6:30 PM      *Adjourn Day 1*
- 8:00 PM**      ***Poolside and Ping Pong Reception – Harmony Terrace***

**Day 2**      **July 12<sup>th</sup>**

- 7:30 AM      *Registration Opens – Harmony Foyer*
- 8:00 AM      ***20<sup>th</sup> Anniversary Special Service Award Presentation – Harmony Ballroom***  
*Peter Scambler and Anne Bassett – Presenting*
- Session V:**      ***Major Associated Systems***  
*Chairs: Bernice Morrow and Oksana Jackson*
- 8:10 AM      **37**      ***Bernice Morrow***  
*Albert Einstein College of Medicine  
New York, NY, USA*
- 8:25 AM      **38**      ***Modupe Adetunji \****  
*Immature Platelet Fraction is Elevated in Individuals with 22q11.2DS Even with Near Normal Platelet Counts*
- 8:35 AM      **39**      ***Jelle Homans\****  
*Scoliosis in Association with the 22q11.2DS*
- 8:40 AM      **40**      ***Jelle Homans\****  
*The Surgical Outcome of Scoliosis Surgery within the 22q11.2DS*

8:45 AM	41	<b>Maria Mascarenhas</b> <i>Exploring the Gastrointestinal Phenotype in 22q11.2DS</i>
8:55 AM	42	<b>Anthony LaMantia</b> <i>A Neurodevelopmental Basis for Perinatal Feeding and Swallowing Disorders in 22q11.2</i>
9:05 AM	43	<b>Thomas Maynard</b> <i>Ranbp1 Haploinsufficiency Contributes to Cranial Neural Crest Anomalies in 22q11.2DS</i>
9:15 AM		Q&A
9:30 AM	44	<b>Oksana Jackson</b> <i>Palatal Abnormalities in 22q11.2DS</i>
9:40 AM	45	<b>Brian Kellogg</b> <i>Revision Speech Surgery in Patients with 22q11.2DS: A Retrospective Review of Speech Outcomes</i>
9:50 AM	46	<b>Alexander de la Mar</b> <i>Comparison of Speech Outcome after Pharyngoplasty in 22q11.2DS: Cranial Based Pharyngeal Flap versus the Modified Honig Procedure</i>
10:00 AM	47	<b>Lisa Elden</b> <i>Anatomical Malformations of the Middle and Inner Ear in 22q11.2DS</i>
10:05 AM	48	<b>Lisa Elden</b> <i>A Cohort Study: Indications and Outcomes of Otolaryngologic Surgeries in Pediatric Patients with 22q11.2DS</i>
10:15 AM	49	<b>Christina Gonzalez-Gandolfi*</b> <i>22q11.2 LCR22D-LCR22E Distal Deletion: Findings in One Illustrative Family</i>
10:20 AM		Q&A
10:30 AM		<i>Coffee Break – Harmony Foyer</i>

**Session VI:**

**Speech, Language and Intellect**

*Chairs: Elizabeth Illingworth and Cindy Solot*

**Invited Speaker:**

**Hippocampal-Prefrontal Miscommunication and Cognitive Deficits**

11:00 AM

50

**David Kupferschmidt**

*National Institute of Health  
Bethesda, MD, USA*

**Submitted Papers: Language, Cognition and Intellect**

11:30 AM

51

**Tessel Boerma\***

*Language Impairment in the 22q11.2DS*

11:40 AM

52

**Cindy Solot**

*Are Language Scores an Early Predictor of Cognitive Decline?*

11:50 AM

Q&A

12:00 PM

53

**Daniel McGinn\***

*Maternal Origin of Familial 22q11.2 Deletions Negatively Impacts FSIQ Scores*

12:10 PM

54

**Elemi Breetvelt**

*A Normative Chart for Cognitive Development in 22q11DS: Implications for 22q11DS and Beyond*

12:20 PM

55

**Ania Fiksinski\***

*The Impact of Parental IQ on the Variable Penetrance of Intellectual Impairment in 22q11DS*

12:30 PM

56

**Ann Swillen**

*Comparison of Cognitive Abilities and Social Responsiveness Skills in Children with 22q11.2DS and Children with Idiopathic Intellectual Disability*

12:40 PM

Q&A

**Invited Speaker:**

**Towards Understanding the Neuronal Circuits of Psychosis in 22q11.2 Deletion Syndrome**

12:50 PM

57

**Stanislav Zakharenko**

*St. Jude Children's Research Hospital  
Memphis, TN, USA*



1:20 PM

Q&A

1:30 PM

*Luncheon and Poster Viewing – Harmony Foyer*

**Session VII:**

***Developmental Trajectories and Psychotic Illness***

*Chairs: Raquel Gur and Maude Schneider*

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

2:15 PM

58

**Laurie Earls**

*The Impact of the Aging Epitranscriptome on Neurophysiologic Phenotypes of 22q11.2DS*

2:25 PM

59

**Raquel Gur**

*The International 22q11.2DS Brain Behavior Consortium: Challenges and Opportunities*

2:35 PM

60

**Ania Fiksinski\***

*Trajectories of Processing Speed and Risk for Psychotic Disorders in 22q11DS: A Longitudinal Study*

2:45 PM

61

**Maria Pontillo**

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

2:55 PM

62

**Wendy Kates**

*Longitudinal Trajectories of Psychiatric Diagnoses and Predictors of Persistence in Youth with 22q11.2DS*

3:05 PM

Q&A

3:15 PM

63

**Maude Schneider**

*Longitudinal Evolution of Negative Symptoms in 22q11.2DS and Predictive Value for Transition to Psychosis*

3:25 PM

64

**Maude Schneider**

*Visual Processing of Complex Social Scenes in 22q11.2DS: Relevance for Social Impairments?*

3:35 PM

65

**Raquel Gur**

*The Evolution of Psychosis in 22q11.2 Deletion Syndrome: Risk and Resilience*

- 3:45 PM**      **66**      **Kathryn McCabe**  
*A Pilot Study Characterizing the Social Impairment Phenotype of Children with 22q11.2DS and Children with Idiopathic Autism Spectrum Disorder*
- 3:49 PM**      **67**      **Elfi Vergaelen\***  
*Differences in Self-and Parent Reporting and the Relation with an At Risk State in Adolescents and Adults with 22q11.2DS*
- 3:55 PM**      **Q&A**
- 4:05 PM**      **68**      **Therese van Amelsvoort**  
*Stress Reactivity, Cortisol Levels and Experience Sampling in Adults with 22q11.2DS*
- 4:15 PM**      **69**      **Marco Armando**  
*Coping Strategies Mediate the Effect of Stressful Life Events on Schizotypal Traits and Psychotic Symptoms in 22q11.2 Deletion Syndrome*
- 4:25 PM**      **70**      **Kathleen Angkustsiri**  
*Parenting, Anxiety and Adaptive Function in Children with Chromosome 22q11.2DS*
- 4:35 PM**      **Q&A**
- 4:45 PM**      *Poster Session with Authors (Even #) and Afternoon Tea – Harmony Foyer*
- 5:45 PM**      **Concurrent Sessions**
- I.**      **Unknown/Difficult Cases – Audience Participation (2 slides/case)**  
*Limited Number – Please email ([jeongsk@email.chop.edu](mailto:jeongsk@email.chop.edu)) intent to present in advance and load during the break – Harmony Ballroom*
- II.**      **Integrative Health – Caring for Oneself via Mindfulness, Massage, Aromatherapy, Yoga and Narrative Medicine**  
*Maria Mascarenhas, MJ McGinn, and Edward Moss – Harmony Foyer*
- 6:30 PM**      **Adjourn Day 2**
- 7:45 PM**      ***First Nations Procession Assembles – Four Seasons Entrance***
- 8:00 PM**      ***Procession Begins Promptly***

**8:15 PM** *Cocktail Hour and Gala Dinner - Squamish and Lil'wat Cultural Centre*

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**Day 3** **July 13<sup>th</sup>**

**7:30 AM** *Registration Opens – Harmony Foyer*

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

**8:10 AM** **2018 Unsung Hero Award Presentation**  
*2016 Recipient Maria Kamper – Presenting*

**Session VIII:** **Genetics and Brain Expression**  
*Chairs: Laurie Earls and Loydie Jerome-Majewska*

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

**8:20 AM** **71** **Sebastien Jacquemont**  
*University of Montreal  
Montreal, Canada*

**Submitted Papers: Genetic Variants and Risk**

**8:50 AM** **72** **Elemi Breetvelt**  
*Burden of Rare Coding Variants in the 22q11.2 Deletion Region is Associated with Educational Attainment and Schizophrenia Risk in Two General Population Cohorts*

**8:55 AM** **73** **Marie De Borre\***  
*Contribution of Rare Hemizygous Variants to Phenotypic Variability in 22q11.2DS*

**9:05 AM** **74** **Jhih-Rong Lin**  
*Integrated Genome-Wide Analyses of Rare Variants for Schizophrenia Risk in 22q11.2DS*

**9:15 AM** **75** **Maria Niarchou\***  
*Genetic Risk for Schizophrenia and Development of Anxiety Disorders and Negative Symptoms in 22q11.2DS*

9:25 AM

Q&A

**Submitted Papers: Neurologic Features**

9:40 AM

76

**Madeline Chadehumbe**

*Neurologic Challenges in 22q11.2DS*

9:50 AM

77

**Christopher Eaton\***

*Epilepsy and Seizures in Young People with 22q11.2DS: Prevalence and Links with Neurodevelopmental Disorders*

9:55 AM

78

**Linda Campbell**

*Emotion Dysregulation in 22q11.2 Deletion Syndrome*

10:00 AM

79

**Davide Vecchio\***

*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*

10:05 AM

80

**Jill Arganbright**

*Sleep Patterns and Problems among Children with 22q11 Deletion Syndrome*

10:10 AM

81

**Hayley Moulding\***

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

10:20 AM

Q&A

10:30 AM

*Coffee Break – Harmony Foyer*

**Session IX:**

**Movement Disorders**

*Chairs: Erik Boot and Doron Gothelf*

**Submitted Papers: Understanding Parkinson's and Motor Findings**

11:00 AM

82

**Erik Boot**

*Parkinsonian Motor Features in Adults with 22q11.2DS*

11:10 AM

83

**Sinéad Morrison\***

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

- 11:16 AM**      **84**      **Matthias Fischer**  
*Anxiety Phenotypes and Biomarkers for Parkinson's disease in 22q11.2DS*
- 11:26 AM**      **85**      **Gabriela Repetto**  
*Analysis of Prodromal Manifestations of Parkinson's disease in Adults with 22q11.2DS*
- 11:30 AM**      **Q&A**
- Session X:**      **Imaging**  
*Chairs: Carrie Bearden and Jacob Vorstman*
- 11:40 AM**      **86**      **Carrie Bearden**  
*The Enigma 22q11.2DS Working Group: Insights into Neurodevelopment and Psychosis*
- 11:50 AM**      **87**      **Therese Van Amelsvoort**  
*Frontal Dopamine D2/3 Receptor Binding in Adults with 22q11.2DS: A [18F] Fallypride Positron Emission Tomography Study*
- 12:00 PM**      **88**      **Clodagh Murphy**  
*The Neuroanatomy of Autism Spectrum Disorder in 22q11.2DS*
- 12:10 PM**      **89**      **Joanne Doherty**  
*Excitatory-Inhibitory Balance in 22q11.2DS: A Pilot Magnetic Resonance Spectroscopy and Magnetoencephalography Study*
- 12:16 PM**      **90**      **Clodagh Murphy**  
*Developing Protocols to Enable MRI Brain Scanning in Infants and Young Children with 22q11.2DS*
- 12:20 PM**      **Q&A**
- Session XI:**      **Looking Ahead**  
*Chairs: Gabriella Repetto and Rosanna Weksberg*
- 12:30 PM**      **91**      **Tracy Heung\***  
*Predictors of All-Cause Mortality in Adults with 22q11.2DS*

- 12:40 PM 92 **Anne Bassett**  
*Medical Multimorbidity in Adults with 22q11.2DS*
- 12:50 PM 93 **Joanne Loo\***  
*Personalized Medical Information Cards for Adults with 22q11.2DS*
- 1:00 PM 94 **Doron Gothelf**  
*Education and Employment Trajectories from Childhood to Adulthood in Individuals with 22q11.2DS*
- 1:10 PM 95 **Paula Goldenberg**  
*Functional Outcomes in 27 Adults with 22q11.2DS*
- 1:15 PM 96 **Lisa Palmer**  
*Identifying Issues Related to Sexual Health in Adults with 22q11.2DS*
- 1:25 PM Q&A
- 1:30 PM *Luncheon and Poster Viewing – Harmony Foyer*
- Session XII: The Future of Clinical and Genetic Research**  
*Chairs: Joris Vermeesch and Christian Marshall*
- Invited Speaker:**  
***Mechanisms of 22q11.2 Deletions and Duplications***
- 2:15 PM 97 **Joris Vermeesch**  
*KU Leuven  
Leuven, Belgium*
- Submitted Papers: Structure, Function and Family**
- 2:45 PM 98 **Lisanne Vervoort\***  
*Optical Mapping of 22q11.2 Low Copy Repeats Reveals Structural Hypervariability*
- 2:55 PM 99 **Beverly Emanuel**  
*Differences in the Frequency of Structural Variation May Affect the Prevalence of the 22q11.2 Deletion Syndrome Amongst Populations*
- 3:05 PM Q&A
- 3:15 PM 100 **Alexander Urban**  
*Haplotype Specific Analysis of Chromosome 22q Folding Patterns in 22q11.2DS*

- 3:25 PM**      **101**    **Michelle Siu**  
*Elucidating Pathophysiology using Genome-Wide DNA Methylation Analysis*
- 3:35 PM**      **102**    **Damien Heine-Suñer**  
*Recurrent de novo 22q11.2 LCR22D-LCR22D Deletion Originating from a Maternal 22q11.2 LCR22B-LCR22D Duplication within a Family*
- 3:40 PM**      **103**    **Nicholas Delihis**  
*A Family of Long Non-Coding RNA Genes in 22q11.2 Shows a High Specificity in Chromosomal Location in Low Copy Repeats and These Genes Contain the Translocation Breakpoint Type A Sequence*
- 3:45 PM**      **Q&A**
- Invited Speaker:**  
***The Future of Genomics – Impact on 22q11.2DS and Vice Versa***
- 3:55 PM**      **104**    **Christian Marshall**  
*University of Toronto  
Toronto, ON, Canada*
- 4:25 PM**      **Q&A**
- 4:30 PM**      **Afternoon Tea – Harmony Foyer**
- Session XIII:**      **Discoveries Every Minute**  
*Chairs: Damian Heine-Suñer and Sebastien Jacquemont*
- Invited Speaker:**  
***CHD7 and Epigenetics***
- 5:00 PM**      **105**    **Peter Scambler**  
*University College London and Great Ormond Street Institute of Child Health  
London, UK*
- Late Breaking Submitted Papers**
- 5:15 PM**      **106**    **Beata Nowakowska**  
*Alterations in TANGO2 on the Intact Chromosome 22q11.2 Allele as a Possible Cause of Sudden Death*
- 5:25 PM**      **107**    **Loydie Jerome-Majewska**  
*A Novel Snap29 Mutant Mouse Line Models a Subset of Abnormalities found in CEDNIK and 22q11.2 Deletion Syndrome Patients*

- 5:35 PM      **108** **Anne Bassett**  
*Schizophrenia-Relevant Collections of Genes from the Rest of the Genome Contribute to Schizophrenia Expression in 22q11.2DS*
- 5:45 PM      **109** **Nigel Williams**  
*Schizophrenia Polygenic Risk Score Analysis in 22q11.2 Deletion Syndrome*
- 5:55 PM      **110** **Jacob Vorstman**  
*The International 22q11DS Brain and Behavior Consortium: Polygenic Risk Score Analysis IQ, IQ-Decline and Subthreshold Psychosis*
- 6:05 PM      Q&A
- 6:20 PM**      ***Junior Investigator Award***  
***2016 Award Recipient Ania Fiksinski - Presenting***
- 6:25 PM**      ***Closing Remarks and Announcement of Future Meeting***
- 6:30 PM      *Adjourn Meeting*
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### ***Poster Presentations:***

**\* - Indicates Junior Investigator**

**+ - Indicates Top Scoring Poster**

❖ *Odd numbered posters – authors present on Wednesday (July 11<sup>th</sup>)*

❖ *Even numbered posters – authors present on Thursday (July 12<sup>th</sup>)*

111.      **Erica Schindewolf\***  
*Prenatal Thymus Size Analysis Predicting T Cell Count in the First Year of Life*
112.      **Russel Jelsema**  
*Non-Invasive Prenatal Testing for Fetal 22q11.2DS*
113.      **Jessica Russo\* +**  
*Impact of Assisted Reproductive Technology on Prevalence and Associated Features in 22q11.2DS*

**\* - Indicates Junior Investigators**



114. **Jill Arganbright**  
*Caring for Children with 22q11.2DS: Current State of 22q Multidisciplinary Team Clinics*
115. **Michael Van Lue**  
*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*
116. **Maria Kamper**  
*22q Coordination Portal and App*
117. **Daniela Schweitzer**  
*Care for Patients with 22q11.2DS within and Outside a Craniofacial Team*
118. **Courtney Hall**  
*Strategies for Delivering Coordinated Interdisciplinary Team Care to Patients with 22q11.2DS*
119. **Anne Lawlor**  
*Developing Integrated Care in the Context of Rare Chromosomal Conditions: 22q11.2DS – A Parent/Clinician Collaboration: The Irish Story*
120. **Sara Ruzzi\***  
*Challenges in Providing Comprehensive Care for Patients with 22q11.2DS Living in Geographic/Cultural Isolates – A Case Report*
121. **Paula Goldenberg**  
*Use of Social Media Targeting Patients and Families Changes National and Global Health Care Outcomes for People with Chromosome 22 Conditions*
122. **Robert Smith\*<sup>+</sup>**  
*Can Maternal Diabetes Exacerbate Phenotypic Features in Patients with 22q11.2 Copy Number Variants?*
123. **Staci Kallish**  
*Hypoparathyroidism Presenting as Acute Cardiomyopathy in Undiagnosed 22q11.2 Deletion Syndrome*
124. **Priyal Patel**  
*Bleeding Phenotype in Children with 22q11DS – Preliminary Results from a Cross-Sectional Study*
125. **Lauren Madhoun**  
*Dysphagia in Young Children with 22q11.2DS*

126. **Daniela Schweitzer**  
*The Role of the Speech-Language Pathologist in Craniofacial Team Care for Patients with 22q11.2DS*
127. **Toko Hayakawa**  
*Language Home Environment of Young Children with 22q11.2DS*
128. **Adriane Baylis**  
*Vocal Output and Parent Input: A Lena Study of Speech in Young Children with 22q11.2DS*
129. **Adriane Baylis** <sup>+</sup>  
*Velopharyngeal Structural and Muscle Variations in Children with 22q11.2DS*
130. **Christopher Hartnick**  
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