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

10:55 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
Elizabeth Illingworth
Institute of Genetics and Biophysics, National Research Council
Naples, Italy

12:05 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
Kathleen Sullivan
Children’s Hospital of Philadelphia and the Perelman School of Medicine at the University of Pennsylvania
Philadelphia, PA, USA

2:15 PM
Kathleen Sullivan

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

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

1:00 PM  
A. 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  
S. Hooper  
*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  
L. Earls  
*The Impact of the Aging Epitranscriptome on Neurophysiologic Phenotypes of 22q11.2DS*

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

3:15 PM  
A. Fiksinski  
*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
<table>
<thead>
<tr>
<th>Time</th>
<th>Presenter</th>
<th>Title and Details</th>
</tr>
</thead>
<tbody>
<tr>
<td>9:50 AM</td>
<td>Vecchio, D*</td>
<td>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</td>
</tr>
<tr>
<td>9:55 AM</td>
<td>Moulding, H*</td>
<td>Sleep Problems and the Relationship With Psychiatric and Neurodevelopmental Difficulties in Young People with 22q11.2DS</td>
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<td>10:05 AM</td>
<td>Q&amp;A</td>
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<tr>
<td>10:15 AM</td>
<td>Coffee Break</td>
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<tr>
<td>10:45 AM</td>
<td>Boot, E</td>
<td>Parkinsonian Motor Features in Adults with 22q11.2DS</td>
</tr>
<tr>
<td>10:55 AM</td>
<td>Morrison, S*</td>
<td>Using Objective Measures of Sensorimotor Control to Improve Our Understanding of Motor Difficulties and the Links with Other Neurodevelopmental Problems in 22q11.2DS</td>
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<tr>
<td>11:01 AM</td>
<td>Fisher, M</td>
<td>Anxiety Phenotypes and Biomarkers for Parkinson’s disease in 22q11.2DS</td>
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<tr>
<td>11:11 AM</td>
<td>Repetto, G</td>
<td>Analysis of Prodromal Manifestations of Parkinson’s disease in Adults With 22q11.2DS</td>
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<tr>
<td>11:15 AM</td>
<td>Q&amp;A</td>
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<td>11:25 AM</td>
<td>Bearden, C</td>
<td>The Enigma 22q11.2DS Working Group: Insights into Neurodevelopment and Psychosis</td>
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<tr>
<td>11:35 AM</td>
<td>van Duin, E*</td>
<td>Frontal Dopamine D2/3 Receptor Binding in Adults with 22q11.2DS: A [18F] Fallypride Positron Emission Tomography Study</td>
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<tr>
<td>11:45 AM</td>
<td>Rogdaki, M</td>
<td>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</td>
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<tr>
<td>11:55 AM</td>
<td>Q&amp;A</td>
<td></td>
</tr>
</tbody>
</table>
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
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*