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

Invited Speaker: Intervention and Treatment Affecting Brain and Behavior

12:05 PM 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

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
Immunology as a Window into Understanding 22q11.2 Issues

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
<table>
<thead>
<tr>
<th>Time</th>
<th>Presentation</th>
<th>Title</th>
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<tbody>
<tr>
<td>5:10 PM</td>
<td>van Mil, S</td>
<td><em>Late Mortality in a Genetic Subtype of Tetralogy of Fallot</em></td>
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<tr>
<td>5:20 PM</td>
<td>Lambert, M</td>
<td><em>Evaluation of Bleeding Risk with Cardiac Surgery in 22q11.2DS: A Case Control Study</em></td>
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<td>5:30 PM</td>
<td>Q&amp;A</td>
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<tr>
<td>5:40 PM</td>
<td>Katz, L</td>
<td><em>Association between Cardiac Surgery and Hypocalcemia in 22q11.2DS</em></td>
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<td>5:50 PM</td>
<td>Boot, E</td>
<td><em>Endocrine Alterations in Adults with 22q11.2DS</em></td>
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<td>6:00 PM</td>
<td>Houben, M</td>
<td><em>Growth in Dutch Children with 22q11.2 Deletion Syndrome - Construction of Reference Growth Charts and Analysis of Determinants of Growth</em></td>
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<td>6:10 PM</td>
<td>Q&amp;A</td>
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<td>Adjourn</td>
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<td>8:00 PM</td>
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<td><em>Poolside and Ping Pong Reception</em></td>
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* - 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

10:30 AM  Willaert, A  
Submitted Papers: Hearing and Language  
Homozygous Tbx1 Missense Mutation in Mice Causes Complete Hearing Loss

10:40 AM  Verheij, E  
Submitted Papers: Hearing and Language  
Anatomical Malformations of the Middle and Inner Ear in 22q11.2DS

10:50 AM  Elden, L  
Submitted Papers: Hearing and Language  
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  
Submitted Papers: Hearing and Language  
Language Impairment in the 22q11.2DS

11:30 AM  Solot, C  
Submitted Papers: Hearing and Language  
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

12:30 PM  McGinn, DE*  
Submitted Papers – Cognition, Intellect and More  
Maternal Origin of Familial 22q11.2 Deletions Negatively Impacts FSIQ Scores
12:40 PM  Breetvelt, E
A Normative Chart for Cognitive Development in 22q11DS: Implications for 22q11DS and Beyond

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

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

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

3:15 PM  Fiksinski, A*
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
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
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

4:40 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