Professional Program

10th Biennial International 22q11.2 Conference
July 20 – 22, 2016
Sirmione, Italy

Day 1 - July 20th

7:00 AM  Registration Open

8:00 AM  Welcome: 22q11.2 Society
Peter Scambler and Donna McDonald-McGinn

8:15 AM  Welcome: Local Arrangements Committee
Bruno Marino and Giulietta Angelelli Cafiero

8:30 AM  Angelo DiGeorge Memorial Medal of Honor Presentation
Anne Bassett – 2014 Recipient Presentation

Session I

8:45 AM  Invited Speaker: CNV Detection in Prenatal Diagnosis: From FISH to Cell-free DNA
Francesca Grati
TOMA
Advanced Biomedical Assays, S.p.A.
University of Milan
Milan, Italy

9:05 AM  Q&A

9:10 AM  Submitted Papers - Setting the Stage
McDonald-McGinn
Back to the Future: The Philadelphia Story - Findings in 1305 Patients with 22q11.2 Deletion Syndrome

9:20 AM  Bassett
Multi-system Expression of 22q11.2 Deletion Syndrome in Adults

9:30 AM  Q&A

9:35 AM  Submitted Papers – Screening
Sparsø
Characterizing the 22q11 Microdeletion in a Danish Sample: A Population-Based Screening of 30,000 Newborn Danes
9:41 AM  **Maisenbacher**  
*Size and Location of 22q11.2 Deletions and Duplications Identified in Products of Conception (POC) Samples: Providing Possible Insight into Genes Critical for Early Development*

9:47 AM  **Barry**  
*Identification of the 22q11.2 Deletion Syndrome via Abnormal Newborn Screening for SCID*

9:55 AM  **Q&A**

10:00 AM  **Submitted Papers – Perinatal Ryan**  
*Raising Confidence Threshold Increases the Positive Predictive Value of a SNP-Based NIPT for the 22q11.2 Microdeletion*

10:06 AM  **Schindewolf**  
*Prenatal Sonographic Findings and Perinatal Outcomes in a Cohort of Confirmed 22q11.2 Deletion Fetuses*

10:12 AM  **Tomita-Mitchell**  
*Early Diagnosis of 22q11.2DS Can Decrease Morbidity and Likely Mortality - A Plea for Universal Newborn Screening*

10:16 AM  **Palmer**  
*22q11.2 Deletion Syndrome: Elucidating the Diagnostic Odyssey*

10:20 AM  **Q&A**

10:30 AM  **Coffee Break**

Session II

11:00 AM  **Keynote Speaker: Genetic and Drug-based Strategies to Correct the Mutant Phenotype in Mouse Models**  
*Antonio Baldini*  
*Institute of Genetics and Biophysics*  
*National Research Council*  
*University Federico II*  
*Naples, Italy*

11:30 AM  **Q&A**

11:40 AM  **Submitted Papers – Cardiac Development Scambler**  
*HIRA is Required for Heart Development and Directly Regulates TNNI2 and TNNT3*
<table>
<thead>
<tr>
<th>Time</th>
<th>Title</th>
<th>Speaker</th>
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<tr>
<td>11:50 AM</td>
<td><em>Morrow</em> Histone Modifier Genes Alter Conotruncal Heart Phenotypes in 22q11.2 Deletion Syndrome</td>
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<td>12:00 PM</td>
<td><em>Racedo</em> Reduced Dosage of β‐Catenin Genetically Rescues Intracardiac Anomalies in TBX1 Conditional Null Mouse Model of 22q11.2 Deletion Syndrome</td>
<td>Racedo</td>
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<td>12:10 PM</td>
<td><em>Roberts</em> CYP26B1−/− Embryos Display 22q11 Deletion Syndrome‐Like Cardiovascular Defects</td>
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<td>12:20 PM</td>
<td>Q&amp;A</td>
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<td>12:30 PM</td>
<td>Lunch</td>
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**Session III**

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<th>Time</th>
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<tr>
<td>1:30 PM</td>
<td>Invited Speaker: Outcomes Following Cardiac Interventions</td>
<td>Matteo Trezzi</td>
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<td>Ospedale Bambino Gesu</td>
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<td>Rome, Italy</td>
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<td>1:50 PM</td>
<td>Q&amp;A</td>
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<tr>
<td>1:55 PM</td>
<td>Submitted Papers – Impact of Cardiac Surgery</td>
<td>Tomita-Mitchell</td>
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<td>Perioperative Outcomes Following Cardiovascular Surgery in Patients with 22q11.2 Deletion Syndrome</td>
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<td>2:01 PM</td>
<td>Lambert</td>
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<td>The Frequency of Transfusion during Surgery for Congenital Heart Disease in Patients with 22q11.2 Deletion Syndrome</td>
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<td>2:07 PM</td>
<td>Gaynor and Unolt</td>
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<td>Prevalence of Congenital Heart Disease in Patients with 22q11.2 Deletion Syndrome and Correlation of CHD Severity with Full Scale IQ Scores</td>
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<td>2:13 PM</td>
<td>Duijff</td>
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<td></td>
<td>Neurodevelopmental Outcome in Infants and Toddlers with 22q11.2 Deletion Syndrome (22q11DS): Effect of Birth Weight, Gestational Age, Head Circumference, Gender and Congenital Heart Disease</td>
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<td>2:23 PM</td>
<td>Mascarenhas</td>
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<td>Gastrointestinal Manifestations in 22q11.2 Deletion Syndrome are Not Related to Congenital Heart Disease</td>
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2:30 PM  Q&A

2:40 PM  Interactive Poster Session and Afternoon Tea

Session IV

3:45 PM  Invited Speaker: Immune and Autoimmune Related Issues
          Caterina Cancrini
          Ospedale Bambino Gesù
          Tor Vergata University
          Rome, Italy

4:05 PM  Q&A

4:10 PM  Submitted Papers - Immunodeficiency and Endocrinopathies
          Sullivan
          T Cell Lymphopenia and Cardiac Anomalies in 22q11.2DS

4:20 PM  Bradley
          Genome-Wide B-Cell Gene Expression Profile in 22q11 Deletion Syndrome

4:26 PM  Lambert
          Increased Prevalence of Malignancy in Twins with 22q11.2 Deletion Syndrome

4:32 PM  *Vergaelen
          The 22q11.2 Deletion Syndrome as a Model to Investigate the Role of T-Cells in Psychosis

4:36 PM  *Domachevsky
          The Association Between Inflammatory Markers and Psychosis in 22q11.2 Deletion Syndrome

4:40 PM  Q&A

4:45 PM  Katz
          Hypocalcemia and Congenital Heart Disease in Youth with 22q11.2 Deletion Syndrome

4:51 PM  Houben
          High Prevalence of Hypoparathyroidism in Children with 22q11.2 Deletion Syndrome During Early Puberty

4:57 PM  *Grand
          Hypocalcemia and Full Scale IQ in 22q11.2 Deletion Syndrome
Session V

5:10 PM  
*Submitted Papers – Family Matters*  
**Cole**  
Perinatal Psychological Risk Among Parents of Babies with a Diagnosis of 22q11.2 Deletion Syndrome

5:16 PM  
**Campbell**  
Same But Different: The Importance of Social Support for Fathers of Kids with Developmental Disabilities

5:20 PM  
**Sell**  
Unmet Needs of Parents and Professionals - The 22q11.2DS Dilemma

5:24 PM  
**Sullivan**  
Healthcare Cost Analysis of Patients with 22q11.2DS

5:30 PM  
**Grebe**  
Phoenix Children’s Hospital 22q Clinic: Rewards and Challenges of Our First Two Years

5:35 PM  
Q&A

5:45 PM  
Open Forum – Challenging Cases Submitted from Audience

6:30 PM  
Adjourn

8:00 PM  
*Lakeside Welcome Reception – “Sirmione Spritz and Canapés”*

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Day 2 - July 21st

7:00 AM  
Registration Open

8:00 AM  
*22q11.2 Society - Unsung Hero Award*  
**Sheila Kambin** – 2014 Recipient Presentation

Session VI

8:15 AM  
*Invited Speaker: Brain Anomalies in Tbx1 Mutant Mice*  
**Elizabeth Lindsay Illingworth**  
Institute of Genetics and Biophysics
8:35 AM  Q&A

8:40 AM  Submitted Papers – Pediatric Brain
Earls
A Novel Age-Specific Micropeptide Regulator of Synaptic Plasticity and
Learning Discovered in the 22q11.2 Deletion Region

8:50 AM  LaMantia
TXNRD2-Dependent Redox Metabolism Mediates Neuron Morphogenesis in
the LGDEL 22q11DS Mouse Model

9:00 AM  Meechan
Mapping Cortical Connections to Behavioral Variability in a Model of
22q11.2DS

9:06 AM  Emanuel
IQ and Hemizygosity for the Val168Met Functional Polymorphism of COMT in
22q11.2DS

9:10 AM  Q&A

9:20 AM  Duijff
IBBC-Junior: The International Brain Behavior Consortium on 22q11DS for 0-
8 Year Olds

9:24 AM  Solot
Speech and Language Development in Patients with 22q11.2 Deletion
 Syndrome

9:34 AM  *Van Den Heuvel
Developmental Course of Socio-Communicative Abilities in School-Aged
Children with 22q11.2 Deletion Syndrome

9:38 AM  Moss
Early identification of cognitive and language impairments in children with
22q11.2 deletion syndrome may predict later outcome

9:45 AM  Q&A

9:50 AM  *Niarchou
Attention Deficit Hyperactivity Disorder in Children with 22q11.2 Deletion
 Syndrome and Their Parents

9:56 AM  *Chawner
What are Measures of Autism Spectrum Disorder Capturing in 22q11.2 Deletion Syndrome?

10:06 AM  
**Murphy**  
*Gender Risk for Autism and ADHD in Young People with 22q11DS*

10:10 AM  
**Finucane**  
*A Family Study of Social Responsiveness and Implications for Refining Autism Risk in 22q11.2 Deletion Syndrome*

10:14 AM  
**Fiksinski**  
*Autism Spectrum and Psychosis Risk in the 22q11.2 Deletion Syndrome: Findings from a Prospective Longitudinal Study*

10:25 AM  
**Q&A**

10:30 AM  
**Coffee Break**

**Session VII**

11:00 AM  
**Submitted Abstracts – Pediatric Brain II**  
**Hopkins**  
*Imaging and Neurologic Sequelae in Children with 22q11.2 Deletion Syndrome*

11:10 AM  
**Simon**  
*Three Distinct Brain Structure Patterns as Potential Biomarkers for Subtypes of Chromosome 22q11.2 Deletion Syndrome*

11:16 AM  
**McCabe**  
*Exploring Oculomotor Processing in 22q11.2 Deletion Syndrome*

11:20AM  
**Q&A**

**Session VIII**

11:30 AM  
**Submitted Abstracts - HEENT**  
**Maynard**  
*Diminished Dosage of the 22q11.2 DS Candidate Gene RANBP1 Disrupts Craniofacial Development*

11:40 AM  
**Rosenboom**  
*Haploinsufficiency of TBX1 is Not Responsible for Facial Dysmorphology in Patients with 22q11.2 Deletion Syndrome*

11:44 AM  
**Jackson**
Polysomnographic Screening of Patients with 22q11.2 Deletion Syndrome Before and After Posterior Pharyngeal Flap Surgery for Velopharyngeal Dysfunction

11:50 AM  Kirschner
Assessing the Risk of Obstructive Sleep Apnea After Posterior Pharyngeal Flap Surgery in Patients with 22q11.2 Deletion Syndrome

11:56 AM  Baylis
Speech Outcome After Pharyngeal Flap Surgery in 22q11.2 Deletion Syndrome

12:00 PM  Q&A

12:10 PM  *Sacca
Association of Airway Abnormalities with 22q11.2 Deletion Syndrome

12:16 PM  Willaert
Vestibular Function in 22q11.2 Deletion Syndrome

12:22 PM  Loos
Malformations of the Middle and Inner Ear on CT-Imaging in 22q11 Deletion Syndrome

12:28 PM  Cohen
Audiological Findings in 22q11.2 Deletion Syndrome

12:32 PM  *Verheij
Audiologic Characteristics of 22q11 Deletion Syndrome

12:36 PM  *Kist
Otological Symptoms, Palatal Cleft and Speech Related Abnormalities in a New Cohort of 22q11DS Patients

12:40 PM  Q&A

12:45 PM  Lunch

Session IX

1:45PM  Submitted Abstracts – Adult Brain and Genetic Risk
Høffding
Risk of Mental Disorders in 22q11.2 Deletion and Duplication Syndrome: A Nation-wide Study

1:49 PM  Marshall
Rare Copy Number Variation in 22q11.2DS with and without Schizophrenia: Initial Results from the IBBC
1:59 PM  *Butcher
Whole-Genome Sequencing in 22q11.2 Deletion Syndrome and Complex Neurophenotypes

2:09 PM  **Zhang**
Prediction and Analysis of Risk Genes on the Individual Patient Level for both Syndromic and Non-syndromic Schizophrenia

2:20 PM  Q&A

2:30 PM  **Gur, RA**
Psychosis Risk in 22q11.2 Deletion Syndrome: Findings from the Philadelphia Sample and Implication for IBBC

2:40 PM  **Gothelf**
Negative Prodromal Symptoms Distinguish 22q11.2 Deletion Syndrome from Other Neurodevelopmental Disorders: A Two-Site Study

2:50 PM  *Schneider*
Transition Rates to Psychosis in 22q11 Deletion Syndrome: A longitudinal, Prospective Long-Term Outcome Study

3:00 PM  **Armando**
Age Matters in the Prevalence and Clinical Significance of Ultra-High-Risk for Psychosis Symptoms and Criteria in 22q11DS

3:06 PM  **Gur, Re**
Neurocognitive Performance in 22q11.2 Deletion Syndrome Measured with a Brief Computerized Battery

3:12 PM  **Fini**
Emotion Recognition Deficits and Social Cognition Impairments: Endophenotypes for Psychosis Risk in 22q11.2 Deletion Syndrome

3:20 PM  Q&A

3:30 PM  Interactive Poster Session and Afternoon Tea

Session X

4:00 PM  **Submitted Abstracts – Adult Brain**
*Vingerhoets*
Prevalence of Substance Use and the Relation with Psychosis and COMT in Patients with Chromosome 22q11 Deletion Syndrome
4:06 PM  
**Boot**  
Movement Abnormalities: Common Manifestations in Adults with 22q11.2 Deletion Syndrome?

4:12 PM  
*Butcher*  
Neuroimaging and Clinical Markers of Parkinson's Disease Expression in 22q11.2 Deletion Syndrome

4:22 PM  
**Repetto**  
Sleep Movement Disorders in Adults with 22q11.2 Deletion: New Dopamine-Related Manifestation? A Case Report

4:26 PM  
*van Duin*  
Reward Learning and Dopamine Release in Adults with 22q11DS

4:30 PM  
Q&A

**Session XI**

4:40 PM  
**Submitted Abstracts – Adult Brain Imaging**  
**Bearden**  
22q11.2 Gene Dosage Effects on Subcortical Brain Structure: The Enigma  
22q11.2 Working Group

4:50 PM  
**Kates**  
Twelve Year Longitudinal Trajectories of Neuroanatomy and Neuropsychological Function in 22q11.2 Deletion Syndrome: Predictions to Psychosis

5:00 PM  
**Hooper**  
Longitudinal Mapping of Psychopathology and Associated Brain MRI Changes in Children and Adolescents with 22q11DS

5:06 PM  
**Sandini**  
Anterior Cingulate and Medial Frontal Disconnectivity are Associated with the Presence of Psychosis in 22q11DS

5:10 PM  
*Vingerhoets*  
Gaba and Glutamate Concentrations in Patients with 22q11.2 Deletion Syndrome and Healthy Volunteers: A Randomized Double-Blind 7TESLA Pharmacological MRS Study

5:15 PM  
Q&A
5:30 PM  Adjourn

8:00 PM  Garden Terrace Dinner

Day 3 - July 22\textsuperscript{nd}

7:00 AM  Registration Open

Session XII

8:00 AM  \textit{Invited Speaker – Recurrence of Congenital Heart Defects in Relatives of Patients with 22q11DS: New Clinical and Cytogenetic Evidence}
\textit{Maria Cristina Digilio}
Ospedale Bambino Gesu
Rome, Italy

8:10 AM  \textit{Submitted Papers – Genetics}
\textit{*Demaerel}
An inversion polymorphism on chromosome 22q11.2 predisposes for 22q11 deletions

8:20 AM  \textit{*Hestand}
Sequence based evaluation of the remaining allele in 22q11.2 deletion patients

8:30 AM  Q&A

8:40 AM  \textit{Breetvelt}
\textit{Increased Burden of Rare Coding Variants in the 22q11 Region is Associated with Educational Attainment in a General Population Sample}

8:46 AM  \textit{Heine-Suñer}
\textit{Significant Excess of De Novo CNVs Outside the 22q11.2 Region in 22q11.2 DS Patients}

8:50 AM  \textit{Urban}
\textit{Multilevel Genomics and Epigenomics Analysis of the Molecular Effects of the 22q11 Deletion}

8:56 AM  \textit{Magdinier}
\textit{Does Epigenetics Contribute to the Phenotypic Variability in the DiGeorge Syndrome?}
9:00 AM  Q&A

Session XIII

9:10 AM  **Submitted Papers – 22q11.2 Duplications**
  
  *McDonald-McGinn*
  
  *Caring for Patients with 22q11.2 Duplications and Distal 22q11.2 Deletions in the Setting of a 22q11.2 Clinic*

9:20 AM  **Solot**
  
  *Speech and Language Development in 37 Patients with 22q11.2 Duplication Syndrome*

9:30 AM  **Digilio**
  
  *Congenital Heart Defects in Micro-duplication 22q11.2 Syndrome*

9:35 AM  Q&A

Session XIV

9:40 AM  **Invited Speaker – 22q11.2DS as a Model for Translational Medicine**
  
  *Hakon Hakonarson*
  
  *Center for Applied Genomics*
  
  *The Children’s Hospital of Philadelphia*
  
  *The Perelman School of Medicine of the University of Pennsylvania*

10:00 AM  **Submitted Paper – Complementary Interventions**
  
  *Mariano*
  
  *Cognitive Remediation for Adolescents with 22q11.2 Deletion Syndrome (22q11DS): Examining Effectiveness and Durability of a Remote, Computer-Based Intervention*

10:10 AM  Q&A

10:15 AM  **Coffee Break**

Session XV

10:45 AM  **Submitted Papers – Growing into Adulthood**
  
  *Bassett*
  
  *Mortality and Longevity in Adults with 22q11.2 Deletion Syndrome*

10:55 AM  **Palmer**
  
  *Psychosexual Knowledge and Related Problems in Adults with 22q11.2 Deletion Syndrome*
11:01 AM  *Vergaelen
Fatigue in Adults with 22q11.2 Deletion Syndrome

11:05 AM  Persson
Signs of Dysarthria in Adults with 22q11.2 Deletion Syndrome

11:10 AM  Q&A

11:15 AM  Shugar
Moving Adolescents with 22q11.2 Deletion Syndrome (22q11DS) to Adult Care: Implementation and Evaluation of a Transition Clinic for Teens and Their Caregivers

11:19 AM  Kallish
Healthcare Transitions from Pediatrics to the Adult Medical Setting for Patients with 22q11.2 Deletion Syndrome

11:23 AM  Schoch
Transitioning to Independence in Adolescents and Young Adults with 22q11DS

11:30 AM  Q&A

11:40 AM  Junior Investigator Award Presentation

11:45 AM  Closing Remarks and Announcement of Future Meeting

12:00 PM  Lunch with Families Attending the Caregivers Meeting-Optional

1:00 PM  Adjourn

* - Indicates Junior Investigators
Posters:

- Even numbered posters – authors present on Wednesday (July 20th)
- Odd numbered posters - authors present on Thursday (July 21st)
- *Indicates Top Scoring Posters - authors to take turns presenting 3 minute summaries poster side in order of poster number (Even Posters on Wednesday, Odd Posters on Thursday)

1. **Padula**
   - Multimodal Large-Scale Networks Connectivity in 22q11.2 Deletion Syndrome

2. **Ciampoli**
   - Developmental Trajectories in a Mouse Model of 22q11.2 Deletion Syndrome

3. **Mosheva**
   - Higher Adaptive Functioning and Low Psychiatric Morbidity Characterize Married Individuals with 22q11.2 Deletion Syndrome

4. **Stoddard**
   - Evaluating a Potentially Efficient Preliminary Assessment for Psychosis Proneness Symptoms in Youth with 22q11.2DS

5. **Souders and Maguire**
   - High Prevalence of Sleep Disorders in 22q11.2 Deletion Syndrome

6. **Voll**
   - Prevalence, Predictors, and Long-term Consequences of Obesity in 22q11.2 Deletion Syndrome

7. **Moe**
   - Identification of Previously Undiagnosed Patients with 22q11 Deletion Syndrome in an Adult Congenital Heart Disease Clinic: The Case for Genetic Testing and Comprehensive Medical Evaluation

8. **Sharkus**
   - Mortality associated with 22q11.2 DS

9. **Boot**
   - Hypocalcemia and Hypomagnesemia in Adults with 22q11.2 Deletion Syndrome
10. *Boot
   • Impaired Manual Dexterity in Adults with 22q11.2 Deletion Syndrome

11. *Wootton
   • Ocular Findings Associated with Chromosome 22q11.2 Duplication

12. *Melchiorre
   • Findings in Familial Cases of 22q11.2 DS

13. *Silverman
   • Apgar Scores and Perinatal Course Compared with Long Term Neurocognitive Outcomes in 22q11.2 DS

14. *Crowley
   • The Death of Paper Charts: Underscoring the Necessity for a 22q11.2DS Worldwide Registry

15. *McGinn
   • Germline and Somatic Mosaicism in 22q11.2 DS

16. Urban
   • Genomic and Epigenomic Analysis of Human Neuronal Cells Directly Induced From Adult Human Fibroblasts Carrying the 22q11 Deletion

17. Saitta
   • Use of Exome Sequencing in Chromosome 22q Deletion Syndrome in Patients with Atypical Phenotypic Features

18. *Cunningham
   • Motor Coordination, IQ and Psychopathology in 22q11.2 Deletion Syndrome

19. Chow
   • The Treatment of ADHD in Canadian Children with 22q11.2 Deletions - A Follow-Up Study

20. Angkustsiri
   • What Might Explain Social Impairments in Children with Chromosome 22q11.2 Deletion Syndrome?

21. Simon
   • Children with 22q11.2 Deletion Syndrome Show Lower Spatial and Temporal Acuity than TD Children in Continuously Varying Tasks

22. *Durdle
Comparing Space and Time: An Exploration of the Spatiotemporal Hypergranularity in Children with Chromosome 22q11.2 Deletion Syndrome

23. Baylis
   - MRI Evaluation of Velopharyngeal Structures in Children with 22q11.2DS

24. Baylis
   - Predictors of Hypernasal Speech in Children with 22q11.2 Deletion Syndrome

25. Maguire
   - Application of Practical Guidelines for Managing Patients with 22q11.2 Deletion Syndrome in a Pediatric Surgical Specialty Practice

26. *Weinberger
   - Neurocognitive Profile in Psychotic Versus Nonpsychotic Individuals with 22q11 Deletion Syndrome

27. Fisher
   - Severe Paranoid Psychosis in a Patient with 22q11.2 Deletion Syndrome: Case Report and Beyond

28. Joseph
   - Cognitive Correlates of Cortical Folding in 22q11.2 Deletion Syndrome

29. Amato
   - Anatomical Differences in the Hippocampus May Explain Cognitive Ability on Spatiotemporal Tasks in Children with Chromosome 22q11.2 Deletion Syndrome

30. *Dubourg
   - Reward Processes, White Matter Pathways of the Reward System and Negative Symptoms in 22q11DS

31. *Gudbrandsen
   - Cortical Thickness and Gender Differences in 22q11.2 Deletion Syndrome

32. *Goodwin
   - The Positive and Negative "Lived" Experience of Parenting an Adult Child with 22q11.2 Deletion Syndrome

* - Indicates Junior Investigators
^ Indicates Top Scoring Poster

Program Book Only:
1. Robles-Sanchez
   - Psychopathology in 27 Spanish Children and Adolescents with 22q11.2 Deletion Syndrome