The 10th Biennial International 22q11.2 Conference

Hotel Acquaviva del Garda
Sirmione, Italy
July 20 – 22, 2016
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
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
11:50 AM  Morrow
Histone Modifier Genes Alter Conotruncal Heart Phenotypes in 22q11.2 Deletion Syndrome

12:00 PM  *Racedo
Reduced Dosage of β-Catenin Genetically Rescues Intracardiac Anomalies in TBX1 Conditional Null Mouse Model of 22q11.2 Deletion Syndrome

12:10 PM  *Roberts
CYP26B1/- Embryos Display 22q11 Deletion Syndrome-Like Cardiovascular Defects

12:20 PM  Q&A

12:30 PM  Lunch

Session III

1:30 PM  Invited Speaker: Outcomes Following Cardiac Interventions
Matteo Trezzi
Ospedale Bambino Gesu
Rome, Italy

1:50 PM  Q&A

1:55 PM  Submitted Papers – Impact of Cardiac Surgery
Tomita-Mitchell
Perioperative Outcomes Following Cardiovascular Surgery in Patients with 22q11.2 Deletion Syndrome

2:01 PM  Lambert
The Frequency of Transfusion during Surgery for Congenital Heart Disease in Patients with 22q11.2 Deletion Syndrome

2:07 PM  Gaynor and Unolt
Prevalence of Congenital Heart Disease in Patients with 22q11.2 Deletion Syndrome and Correlation of CHD Severity with Full Scale IQ Scores

2:13 PM  Duijff
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

2:23 PM  Mascarenhas
Gastrointestinal Manifestations in 22q11.2 Deletion Syndrome are Not Related to Congenital Heart Disease
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

5:05 PM  Q&A
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”

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
National Research Council
Naples, Italy
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<thead>
<tr>
<th>Time</th>
<th>Session/Preparation</th>
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<tbody>
<tr>
<td>8:35 AM</td>
<td>Q&amp;A</td>
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<tr>
<td>8:40 AM</td>
<td>Submitted Papers – Pediatric Brain</td>
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<td><strong>Earls</strong></td>
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<td>A Novel Age-Specific Micropetide</td>
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<td>Regulator of Synaptic Plasticity</td>
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<td>and Learning Discovered in the 22q11.2 Deletion Region</td>
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<td>8:50 AM</td>
<td><strong>LaMantia</strong></td>
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<td>TXN RD2-Dependent Redox Metabolism</td>
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<td>Mediates Neuron Morphogenesis in</td>
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<td>the LGDEL 22q11DS Mouse Model</td>
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<td>9:00 AM</td>
<td><strong>Meechan</strong></td>
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<td>Mapping Cortical Connections to</td>
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<td>Behavioral Variability in a Model</td>
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<td>of 22q11.2DS</td>
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<td>9:06 AM</td>
<td><strong>Emanuel</strong></td>
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<td>IQ and Hemizygosity for the Val168Met Functional Polymorphism of COMT in 22q11.2DS</td>
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<td>9:10 AM</td>
<td>Q&amp;A</td>
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<td>9:20 AM</td>
<td><strong>Duijff</strong></td>
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<td>IBBC-Junior: The International</td>
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<td>Brain Behavior Consortium on 22q11DS for 0-8 Year Olds</td>
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<td>9:24 AM</td>
<td><strong>Solot</strong></td>
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<td>Speech and Language Development in</td>
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<td>Patients with 22q11.2 Deletion</td>
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<td>Syndrome</td>
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<td>9:34 AM</td>
<td>*Van Den Heuvel</td>
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<td>Developmental Course of Socio-</td>
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<td>Communicative Abilities in School-</td>
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<td>Aged Children with 22q11.2 Deletion</td>
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<td>9:38 AM</td>
<td><strong>Moss</strong></td>
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<td>Early identification of cognitive</td>
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<td>and language impairments in children with 22q11.2 deletion syndrome may predict later outcome</td>
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<td>9:45 AM</td>
<td>Q&amp;A</td>
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<td>9:50 AM</td>
<td>*Niarchou</td>
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<td>Attention Deficit Hyperactivity</td>
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<td>Disorder in Children with 22q11.2</td>
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<td>Deletion Syndrome and Their Parents</td>
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<td>9:56 AM</td>
<td>*Chawner</td>
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<td></td>
<td>What are Measures of Autism Spectrum Disorder Capturing in 22q11.2 Deletion Syndrome?</td>
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<td>10:06 AM</td>
<td>Murphy</td>
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<td>Finucane</td>
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<td>10:14 AM</td>
<td>Fiksinski</td>
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**Session VII**

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<tr>
<td>11:00 AM</td>
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<td><strong>Submitted Abstracts – Pediatric Brain II</strong></td>
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<td></td>
<td>Hopkins</td>
<td>Imaging and Neurologic Sequelae in Children with 22q11.2 Deletion Syndrome</td>
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<td>11:10 AM</td>
<td>Simon</td>
<td>Three Distinct Brain Structure Patterns as Potential Biomarkers for Subtypes of Chromosome 22q11.2 Deletion Syndrome</td>
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<tr>
<td>11:16 AM</td>
<td>McCabe</td>
<td>Exploring Oculomotor Processing in 22q11.2 Deletion Syndrome</td>
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<td>11:20 AM</td>
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<td>Q&amp;A</td>
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**Session VIII**

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<tr>
<td>11:30 AM</td>
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<td><strong>Submitted Abstracts - HEENT</strong></td>
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<td>Maynard</td>
<td>Diminished Dosage of the 22q11.2 DS Candidate Gene RANBP1 Disrupts Craniofacial Development</td>
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<tr>
<td>11:40 AM</td>
<td>Rosenboom</td>
<td>Haploinsufficiency of TBX1 is Not Responsible for Facial Dysmorphology in Patients with 22q11.2 Deletion Syndrome</td>
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<tr>
<td>11:44 AM</td>
<td>Jackson</td>
<td>Polysomnographic Screening of Patients with 22q11.2 Deletion Syndrome Before and After Posterior Pharyngeal Flap Surgery for Velopharyngeal Dysfunction</td>
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<td>11:50 AM</td>
<td><strong>Kirschner</strong></td>
<td>Assessing the Risk of Obstructive Sleep Apnea After Posterior Pharyngeal Flap Surgery in Patients with 22q11.2 Deletion Syndrome</td>
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<td>11:56 AM</td>
<td><strong>Baylis</strong></td>
<td>Speech Outcome After Pharyngeal Flap Surgery in 22q11.2 Deletion Syndrome</td>
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<td>12:00 PM</td>
<td>Q&amp;A</td>
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<td>12:10 PM</td>
<td><em>Sacca</em></td>
<td>Association of Airway Abnormalities with 22q11.2 Deletion Syndrome</td>
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<td>12:16 PM</td>
<td><strong>Willaert</strong></td>
<td>Vestibular Function in 22q11.2 Deletion Syndrome</td>
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<td>12:22 PM</td>
<td><strong>Loos</strong></td>
<td>Malformations of the Middle and Inner Ear on CT-Imaging in 22q11 Deletion Syndrome</td>
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<td>12:28 PM</td>
<td><strong>Cohen</strong></td>
<td>Audiological Findings in 22q11.2 Deletion Syndrome</td>
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<td>12:32 PM</td>
<td><em>Verheij</em></td>
<td>Audiologic Characteristics of 22q11 Deletion Syndrome</td>
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<td>12:36 PM</td>
<td><em>Kist</em></td>
<td>Otological Symptoms, Palatal Cleft and Speech Related Abnormalities in a New Cohort of 22q11DS Patients</td>
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<td>12:40 PM</td>
<td>Q&amp;A</td>
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<td>12:45 PM</td>
<td>Lunch</td>
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**Session IX**

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<tr>
<td>1:45 PM</td>
<td><strong>Høffding</strong></td>
<td>Submitted Abstracts – Adult Brain and Genetic Risk</td>
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<td><strong>Marshall</strong></td>
<td>Risk of Mental Disorders in 22q11.2 Deletion and Duplication Syndrome: A Nation-wide Study</td>
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<td>1:49 PM</td>
<td><strong>Marshall</strong></td>
<td>Rare Copy Number Variation in 22q11.2DS with and without Schizophrenia: Initial Results from the IBBC</td>
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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
Day 3 - July 22nd

7:00 AM  Registration Open

Session XII

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

8:10 AM  Submitted Papers – Genetics
*Demaevel
An inversion polymorphism on chromosome 22q11.2 predisposes for 22q11 deletions

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

8:30 AM  Q&A

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

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

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

8:56 AM  Magdinier
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 Microduplication 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

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 Mosaicim 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