22q11.2 Society
est. 2013
Quaestio · Curatio
Universum Veritas

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

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

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

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 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***
****Demaerel***
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 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***

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*

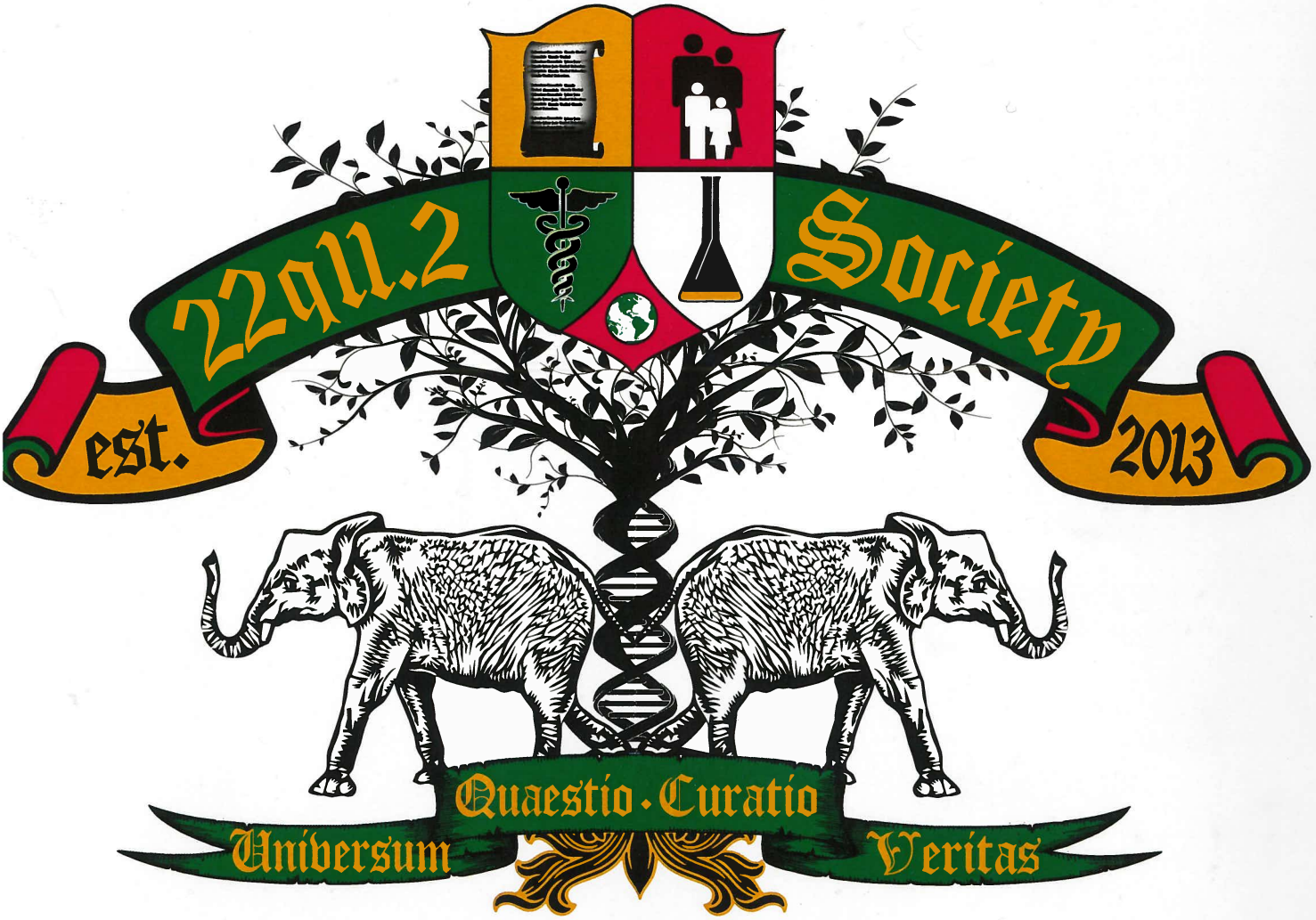
31. ****Gudbrandsen***
    - *Cortical Thickness and Gender Differences in 22q11.2 Deletion Syndrome*

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*



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