



12th Biennial International 22q11.2 Scientific Conference

Le Méridien Lav Hotel

Split, Croatia on the Dalmacija Coast

Wednesday, June 29 through Friday, July 01, 2022

www.22qsociety.org

Tuesday, June 28

15:00 – 17:00 Registration Open – Jim’s Foyer, Grand Dalmacija Ballroom
Speaker Ready Room Open – Hvar

Wednesday, June 29

6:00 Registration Open – Jim’s Foyer, Grand Dalmacija Ballroom
Speaker Ready Room Open – Hvar

6:30 Sunrise Yoga – Meet in Korcula at 6:25

7:00 Breakfast – Spalatum Restaurant

Session 1 Opening Ceremonies - Grand Dalmacija Ballroom

22q11.2 Society Trustees

Donna M. McDonald-McGinn, Anne Bassett, Bernice Morrow, Peter Scambler, Ann Swillen

8:00 Musical Prelude
Karen Ruckman-Lindsay

8:05 22q11.2 Society Welcome
22q11.2 Society Trustees
Donna M. McDonald-McGinn, Anne Bassett, Bernice Morrow, Peter Scambler, Ann Swillen

8:10 Croatia - Delighted to Greet You
Le Méridien Lav Hotel
Vana Kovacic

8:15 2020 and 2022 Inaugural Clodagh Murphy Unsung Hero Awards
22q11.2 Society
Ben Cresswell

- 8:20** **2020 and 2022 Angelo DiGeorge Memorial Medals of Honor**
 22q11.2 Society
Presented by 2018 Recipient Bernice Morrow
- 8:25** **Looking Back and Looking Forward – Two Lives Dedicated to Studying 22q11.2**
Angelo DiGeorge Memorial Medal of Honor Award Recipients
- 8:35** **1** **From Split to Philadelphia and Back Again – A Scientists Tale**
 Genetic Dissection of Complex Behaviors in Mice and Humans
Maja Bucan
Philadelphia, PA, USA
- 8:50** **2** **Peter Scambler Inaugural Invited Lecture**
 Exploring *TBX1* Function in the 2nd Heart Field and Cardiopharyngeal Mesoderm
Robert Kelly
Marseilles, France
- 9:05** **3** **Whole Genome Sequence Analysis Identifies Genetic Modifiers of Conotruncal Heart Defects**
Bernice Morrow, Yingjie Zhao, T. Blaine Crowley, Elizabeth Goldmuntz, Donna M. McDonald-McGinn, Beverly S. Emanuel
- 9:15** **4** **Generation and Characterization of an in Vitro Differentiation Platform to Generate Pharyngeal Endodermal Cells Starting from Human Embryonic Stem Cells**
Andrea Cipriano, Alessio Colantoni, Danielle Gomes, Mahdi Moqri, Alexander Parker, Francesca Briganti, Mariagrazia Roncarolo, Antonio Baldini, Giangetano Tartaglia, Vittorio Sebastiano
- 9:25** **5** **Neuroanatomical Abnormalities in the Cerebellum of Mouse Models and Human Subjects With 22q11.2 Deletion Syndrome**
Tae-Yeon Eom
- 9:35** **6** **Abnormalities of *TBX1* Result in Broad Overlapping Features of 22q11.2 Deletion Syndrome**
Donna M. McDonald-McGinn, Victoria Giunta, T. Blaine Crowley, Daniel E. McGinn, Lauren Lairson, Oanh Tran, Alice Bailey, Beth Keena, Beverly Emanuel, Rosemarie Smith, Ellen Moran, Elaine Zackai, and Beata Nowakowska
- 9:45** **Q&A**
- 10:00** **Coffee Break and Poster Viewing - Grand Dalmacija Terrace**
- Session 2** **Impact of Detection on Families - Grand Dalmacija Ballroom**
- 10:30** **7** **22q11.2 – A Global Health Challenge**
 Providing Comprehensive Care in Developing Nations
Gabriela Repetto
Santiago, Chile

- 10:45 8 22q11.2 – Challenges and Benefits of Prenatal Detection**
 Prenatal Screening and Preimplantation Genetic Diagnosis
Natalie Blagowidow
 Baltimore, MD, USA
- 11:00 9 22q11.2 – Population Based Detection**
 Results of the SMART Study
Hakon Hakonarson
 Philadelphia, PA, USA
- 11:15 Q&A**
- 11:25 10 Prenatal Diagnosis of 22q11.2 Deletion Syndrome Improves Perinatal and Infant Outcomes**
Lindsay Freud, Stephanie Galloway, T. Blaine Crowley, Julie Moldenhauer, Nicole Sarles-Philip Ann Swillen, Jeroen Breckpot, Antoni Borrell, MD, Neeta Vora, Bettina Cuneo, Beata Nowakowska, Trisha Vigneswaran, Jeffrey Dungan, Karl-Philipp Gloning, Maria Cristina Digilio, Gabriela Repetto, Sixto Garcia-Minaur, Anne Bassett, Ronald Wapner, Donna McDonald-McGinn
- 11:35 11 Prenatal Screening For 22q11.2DS: Does It Change Patient Care?**
Sheetal Parmar, Kimberly Martin, Mary Norton, Cora MacPherson, Bo Jacobsson, Sina Haeri, Melissa Egbert, Fergal Malone, Ronald Wapner, Ashley Roman, Asma Khalil, Revital Faro, Rajeevi Mandankumar, Lance Edwards, Noel Strong, Robert Silver, Nidhi Vohra, Jon Hyett, Zachary Demko, Charly Kao, Pe'er Dar
- 11:45 12 A Low-Cost Microsatellite Marker Method For Quick And Reliable Prenatal 22q11.2DS Screening and Detection in Fetuses With Suspected Congenital Heart Defects**
Laura Torres-Juan, Carmen Vidal, Rosa Martorell, Fernando Santos-Simarro, Victor Asensio-Landa, Iciar Martinez-Lopez, Damian Heine Suñer
- 11:50 13 Prenatal Screening for Pathogenic Submicroscopic Copy Number Variations in Circulating Fetal Cells From Maternal Blood**
Claudio Forcato, Anna Doffini, Chiara Mangano, Martina Dori, Roberta Aversa, Chiara Maranta, Emilia D. Giovannone, Genny Buson, Chiara Bolognesi, Rebecca Maiocchi, Debora Lattuada, Enrico Ferrazzi, Paola Ricciardi-Castagnoli, Francesca R. Grati
- 11:55 14 Dilated Cavum Septum Pellucidum: a Prenatal Soft Marker for Identifying Patients with 22q11.2 Deletion Syndrome**
Erica Schindewolf, T. Blaine Crowley, Daniel E. McGinn, Alice Bailey, Elaine H. Zackai, Sarah Hopkins, Madeline Chadehumbe, Larissa Bilaniuk, Ed Oliver, Donna M. McDonald-McGinn, Julie Moldenhauer
- 12:00 15 Reproductive Outcomes in Adults with 22q11.2 Deletion Syndrome ***
Lisa Palmer, Tracy Heung, Zoe McManus, Grace McAlpine, Christina Blagojevic, Maria Corral, Anne Bassett

12:05 **16** **Marital Status and Family Structure Within the Complex Care of Children With 22q11.2 Deletion Syndrome ***
Kimberly Gaiser, T. Blaine Crowley, Lauren Lairson, Victoria Giunta, Alice Bailey, Daniel McGinn, Oanh Tran, Beverly S. Emanuel, Elaine H. Zackai, Donna M. McDonald-McGinn

12:10 **17** **Adoption in Association with 22q11.2 Deletion Syndrome ***
Victoria Giunta, T. Blaine Crowley, Kimberly Gaiser, Daniel McGinn, Lauren Lairson, Oanh Tran, Alice Bailey, Beverly S. Emanuel, Elaine H. Zackai, Donna M. McDonald-McGinn

12:15 **Q&A**

12:30 **Group Photo and Lunch – Spalatum Restaurant**

Session 3 Mechanisms - Grand Dalmacija Ballroom

13:30 **18** **Exploring Low Copy Repeats in Humans**
Optical Maps of 22q11.2 Deletion Syndrome Affected Families Reveals Complex LCR22 Structures and Preferred Locations for Non-Allelic Homologous Recombination
Beverly S. Emanuel
Philadelphia, PA, USA

13:45 **19** **Exploring Low Copy Repeats in Apes**
Examining Optical Maps in the Great Apes (Chimpanzees, Bonobo, Gorillas, Orangutans, and Rhesus Monkeys) Provides Insights into the Unique LCR Expansion in Humans and Potential Associations with Phenotypic Variability in 22q11.2DS
Joris Vermeesch
Leuven, Belgium

14:00 **20** **Optical Mapping Reveals Significant Differences in 22q11.2 Genomic Structures Between an African American Population and Whites**
Steven Pastor, Beverly Emanuel, Oanh Tran, Andrea Jin, Danielle Carrado, Donna M. McDonald-McGinn, Mike Xie, Deanne Taylor, Ming Xiao

14:05 **21** **Is 22q11.2 Deletion Syndrome Truly Less Common in African American Patients?**
Donna M. McDonald-McGinn, T. Blaine Crowley, Ian Campbell, Alice Bailey, Daniel McGinn, Kim Gaiser, Victoria Giunta, Lauren Lairson, Oanh Tran, Matt Share, Kathleen Valverde, Steven Pastor, Beverly Emanuel, PhD, and Elaine H. Zackai, MD

14:15 **22** **22q11.2 Rearrangements Caused by Non-Allelic Homologous Recombination and Palindromic At-Rich Repeat-Mediated Pathways ***
Lisanne Vervoort, Nicolas Dierckxsens, Bo Zhou, Ruben Cools, Tracy Heung, Greet Peeters, Ann Swillen, Jeroen Breckpot, Steven Pastor, Donna M. McDonald-McGinn, Beverly Emanuel, Hilde Van Esch, Anne Bassett, Alexander Urban, Joris Vermeesch

14:20 23 **CTIR-Seq Enables Complete and Haplotype-Specific Sequence Resolution of the 22q11.2 A-D Rearrangements in Individual Patients and Mapping of the A-D Deletion Breakpoints**
Bo Zhou, GiWon Shin, Lianne Vervoort, Stephanie Greer, Tanmoy Roychowdhury, Reenal Pattni, Alexej Abyzov, Hanlee Ji, Joris Vermeesch, Alexander Urban

14:25 Q&A

Session 4 Duplications, Atypicals, and Mosaics – Oh My! - Grand Dalmacija Ballroom

14:35 24 **The importance of the Intact Chromosome 22q11.2 Allele in Humans**
Pathogenic Variants on the Remaining Allele in Patients with a Chromosome 22q11.2 Deletion Result in Unexpected Autosomal Recessive Conditions
Beata Nowakowska
Warsaw, Poland

14:50 25 **The Perplexing 22q11.2 Duplication Syndrome: An Important Cause of Congenital Anomalies, Medical Conditions, Cognitive Deficits and Behavioral Phenotypes – Or Nothing At All! ***
Daniel E. McGinn, T. Blaine Crowley, Alice Bailey, Kim Gaiser, Victoria Giunta, Lauren Lairson, Oanh Tran, Beverly S. Emanuel, Elaine H. Zackai, Sixto Garcia-Minaur, Damian Heine-Suner, Marta Unolt, M. Cristina Digilio, Nicole Sarles-Philip, Anne S. Bassett, Ann Swillen, Donna M. McDonald-McGinn

15:00 26 **Deep Phenotypic Characterization In Patients With Proximal 22q11.2 Duplication ***
Jente Verbesselt, Inge Zink, Jeroen Breckpot, Ann Swillen

15:05 27 **Unexpected 22q11.2 Laboratory Results: Parental CNVs, Somatic Mosaicism, and Presumed Germline Mosaicism ***
Sarah Donoghue, T. Blaine Crowley, Alice Bailey, Daniel E. McGinn, Lauren Lairson, Victoria Giunta, Oanh Tran, Elaine Zackai, Beverly S. Emanuel, Donna M. McDonald-McGinn

15:15 28 **22q11.2-22q11.23 Distal Deletions and Duplications: Exploring Atypical Diagnoses ***
Daniel E. McGinn, T. Blaine Crowley, Alice Bailey, Kim Gaiser, Victoria Giunta, Lauren Lairson, Oanh Tran, Beverly Emanuel, Elaine Zackai, Donna M. McDonald-McGinn

15:25 29 **Association of 22q11.2 Deletion/Duplication With Another Genetic Anomaly: Clinical and Genetic Implications ***
Carolina Putotto, Marta Unolt, Maria Cristina Digilio, Rossella Capolino, Francesca Romana Lepri, Viola Alesi, Maria Gnazzo, Maria Lisa Dentici, Paolo Versacci, Emanuele Agolini, Silvia Genovese, Lorenzo Sinibaldi, Alessandra Terracciano, Bruno Marino, Bruno Dallapiccola, Antonio Novelli

15:35 30 **Coexisting Genetic Findings in a Cohort of Patients with Laboratory Confirmation of 22q11.2 Deletion Syndrome ***
Flaminia Pugnaroni, Carolina Putotto, Gioia Mastromoro, Federica Pulvirenti, MD, Stella Maiolo, Marta Unolt, Maria Cristina Digilio, Agnese Giovannetti, Viviana Caputo, Paolo Versacci, Bruno Marino

15:40 **31** **Two Different Microdeletion Syndromes Responsible for Neonatal Hypotonia and Developmental Delay: Third Case Report of a Patient Diagnosed with Williams and 22q11.2DS***
Fanni Szumutku, Éva Pinti, Zaránd Némethi, Árpád Ferenc Kovács, Irén Haltrich

15:45 **Q&A**

16:00 **Afternoon Tea, Poster Viewing, and Wellness Breakout I – Grand Dalmacija Terrace, Grand Dalmacija Ballroom, and Vis**

16:00 **Wellness Breakout I – Vis**
Writing for Stress Release
MJ McGinn
Philadelphia, PA, USA

Session 5 **Care Across the Lifespan Including During COVID-19 - Grand Dalmacija Ballroom**

16:30 **32** **Updated Clinical Practice Guidelines for Managing Children With 22q11.2 Deletion Syndrome**
Sólveig Óskarsdóttir, Erik Boot, T. Blaine Crowley, Joanne C.Y. Loo, Jill M. Arganbright, Marco Armando, Adriane L. Baylis, Elemi J. Breetvelt, René M. Castelein, Madeline Chadehumbe, Christopher M. Cielo, Steven de Reuver, Stephan Eliez, Ania M. Fiksinski, Brian J. Forbes, Emily Gallagher, Sarah E. Hopkins, Oksana A. Jackson, Lorraine Levitz-Katz, Gunilla Klingberg, Michele P. Lambert, Bruno Marino, Maria R. Mascarenhas, Julie Moldenhauer, Edward M. Moss, Beata Anna Nowakowska, Ani Orchanian-Cheff, Carolina Putotto, Gabriela M. Repetto, Erica Schindewolf, Maude Schneider, Cynthia B. Solot, Kathleen E. Sullivan, Ann Swillen, Marta Unolt, Jason P. Van Batavia, Claudia Vingerhoets, Jacob Vorstman, Anne S. Bassett, Donna M. McDonald-McGinn

16:35 **33** **Updated Clinical Practice Guidelines for Managing Adults with 22q11.2 Deletion Syndrome**
Erik Boot, Sólveig Óskarsdóttir, Joanne C.Y. Loo, T. Blaine Crowley, Ani Orchanian-Cheff, Danielle M. Andrade, Jill M. Arganbright, René M. Castelein, Christine Cserti-Gazdewich, Steven de Reuver, Ania M. Fiksinski, Gunilla Klingberg, Anthony E. Lang, Maria R. Mascarenhas, Edward M. Moss, Beata Anna Nowakowska, Erwin Oechslin, Lisa Palmer, Gabriela M. Repetto, Nikolai Gil D. Reyes, Maude Schneider, Candice Silversides, Kathleen E. Sullivan, Ann Swillen, Therese A.M.J. van Amelsvoort, Jason P. Van Batavia, Claudia Vingerhoets, Donna M. McDonald-McGinn, and Anne S. Bassett

16:40 **34** **Lifetime Health Care Burden of Adults with 22q11.2 Deletion Syndrome**
Sarah Malecki, Tracy Heung, Maria Corral, Anne Bassett

16:45 **35** **Findings in Individuals with Chromosome 22q11.2 Deletion Syndrome Diagnosed In Adulthood**
Dierdre Kessler, T. Blaine Crowley, Alice Bailey, Kim Gaiser, Victoria Giunta, Lauren Lairson, Daniel E. McGinn, Oanh Tran, Kathleen Valverde, Beverly Emanuel, Elaine Zackai, Donna M. McDonald-McGinn

16:50 **36** **Features of 22q11.2 Deletion Syndrome and Transition to Adult Care ***
Lisa Palmer, Tracy Heung, Maria Corral, Anne Bassett

- 16:55 **37** **A Three-Year Experience of a New 22q Syndrome Multidisciplinary Clinic In Florida: From Inauguration, Through COVID-19 Pandemic and Beyond ***
Zaimary Meneses, Jeshira Reynoso, Sweeti Shah, Martha Taboada, Andres Jimenez-Gomez, Tara Popp, Carolina Miranda, Karezhe Mersha, Roman Yusupov, Kristen DeLuca, Diana Acevedo, Alesky Caffo, Diana Gelfald, Veronica Figallo, Ivy Fernandez-Fulchan, Hanadys Ale
- 17:00 **Q&A**
- 17:15 **38** **Genes To Mental Health Network Report: Stakeholders Perspectives On Research Participation**
T. Blaine Crowley, Lauren White, Brenda Finucane, Sarah Donoghue, Sixto Garcia-Minaur, Gabriela Repetto, Raquel Gur, Anne Maillard, Kirsty Donald, Anne Bassett, Ann Swillen, Donna M. McDonald-McGinn
- 17:20 **39** **Report from Individuals with 22q11.2 Deletion Syndrome and Their Caregivers During the COVID-19 Pandemic**
Lauren White, T. Blaine Crowley, Brenda Finucane, Sixto Garcia-Minaur, Gabriela Repetto, Marianne Van Den Bree, Matthias Fischer, Sebastien Jacquemont, Ran Barzilay, Anne Maillard, Kirsty Donald, Raquel Gur, Anne Bassett, Ann Swillen, Donna M. McDonald-McGinn
- 17:25 **40** **How Adults with 22q11.2 Deletion Syndrome Fared in the COVID-19 Pandemic in Canada**
Tracy Heung, Lisa Palmer, Samantha D'Arcy, Maria Corral, Anne S. Bassett
- 17:30 **41** **Immune Response Following Two and Three Doses of BNT162b2 mRNA COVID-19 Vaccine In Patients With 22q11.2 Deletion Syndrome ***
Federica Pulvirenti, Ane Fernandez Salins, Carolina Putotto, Sara Terreri, Eva Piano Mortari, Bianca Cinicola, Paolo Versacci, Bruno Marino, Rita Carsetti, Isabella Quinti
- 17:35 **Q&A**
- 17:50 **Wellness Close**
 Release Your Day Through a Moment of Mindfulness
Maria Mascarenhas
Philadelphia, PA, USA
- 18:00 **Adjourn**
- 19:00 Welcome Reception – 7 Palms Restaurant at The Beach**
- 21:00 **Integrative Health Experiential - Korcula**
 Release Your Day Through Body and Mind Practices
Greg Hedler
Maria Mascarenhas
Lisa Squires

Thursday, June 30

6:00 Registration Open – Jim’s Foyer, Grand Dalmacija Ballroom
Speaker Ready Room Open – Hvar

6:30 Sunrise Yoga – Meet in Korcula at 6:25

7:00 Breakfast – Spalatum Restaurant

Session 6 Of Mice and Men - Grand Dalmacija Ballroom

- 8:00 42 **The importance of the Intact Chromosome 22q11.2 Allele in Mice**
Snap29 Mutant Mice Recapitulate Neurological and Ophthalmological Abnormalities Associated with 22q11.2 Deletion Syndrome and CEDNIK Syndrome
Loydie Jerome-Majewska
Montreal, Canada
- 8:15 43 **Seizures and Epilepsy in Children with 22q11.2 Duplication Syndrome**
Sarah Hopkins, Chadehumbe Madeline, T. Blaine Crowley, Daniel E. McGinn, Lauren Lairson Lauren, Victoria Giunta, Alice Bailey, Beverly S. Emanuel Beverly, Elaine H. Zackai, Donna M. McDonald-McGinn Donna
- 8:25 44 **Clinical and Cognitive Correlates of Multivariate Patterns of Sleep Disturbance in 22q11.2 Deletion Syndrome ***
Niveetha Thillainathan, Natacha Reich, Corrado Sandini, Farnaz Delavari, Schneider Maude, Eliez Stephan
- 8:35 45 **RANBP1-Mediated Midline Signaling Anomalies Contribute to Craniofacial Defects in 22q11.2 Deletion Syndrome**
Thomas Maynard
- 8:45 46 **Computer-Aided Facial Dysmorphology Technology to Probe Links Between 22q11.2 Deletion Syndrome and Psychosis**
David Roalf, Donna McDonald-McGinn, Joelle Jee, Mckenna Krall, Christian Kohler, Andrew Crow, T. Blaine Crowley, Ian Campbell, Daniel McGinn, Alice Bailey, Russell Shinohara, Lauren White, Emily McClellan, R. Sean Gallagher, Virgil Gozenbach, Ruben Gur, Elaine Zackai, Paul Moberg, Raquel Gur
- 8:55 Q&A
- 9:10 47 **The Posterior Pharyngeal Flap for Operative Management of Velopharyngeal Dysfunction in 22q11.2 Deletion Syndrome**
Rotem Kimia, Cynthia Solot, Susan McCormack, Nisha Vora, Adam Hornbaker, Lisa Elden, Elaine Zackai, Donna M. McDonald-McGinn, Richard Kirschner, Oksana Jackson

- 9:20 **48** **Peri-Operative Bleeding in Patients with 22q11.2 Deletion Syndrome Undergoing Palatal Repair/Adenotonsillectomy**
Lisa Elden, Ryan Borek Ryan, Rotem Kimia, Julia Dailey, Terri Giordano, T. Blaine Crowley, Daniel E. McGinn, Alice Baily, Elaine H. Zackai, Michele Lambert, Donna M. McDonald-McGinn, Oksana Jackson
- 9:30 **49** **Magnetic Resonance Angiography in Preoperative Planning for Patients with 22q11.2 Deletion Syndrome Undergoing Craniofacial and Otorhinolaryngologic Procedures**
Rotem Kimia, Lisa Elden, Julia Dailey, Mallika Kodavatiganti, T. Blaine Crowley, Daniel E McGinn, Alice Bailey, Elaine H. Zackai, Donna M. McDonald-McGinn, Oksana A. Jackson
- 9:35 **50** **Airway Anomalies Present in 22q11.2 Deletion Syndrome, Systematic Review and Meta-Analysis**
Paul Hanky, Jana Ghulmiyyah, Hung-Wen Yeh, Meghan Tracy, Jill Arganbright
- 9:40 **51** **Dental Care of the Paediatric 22q11.2 Deletion Syndrome Population in the Republic of Ireland**
Rema Buzeid, Yvonne MacAuley, Kirsten FitzGerald, Suzanne Kelleher
- 9:45 **Q&A**
- 10:00** **Coffee Break and Poster Viewing - Grand Dalmacija Terrace**
- 10:30 **52** **Auditory Evoked-Potential Abnormalities in a Mouse Model of 22q11.2 Deletion Syndrome Correlate With Inter-Individual Variability in Hearing Impairment ***
Chen Lu, Jennifer Linden
- 10:40 **53** **Hearing Loss in Patients With 22q11.2 Deletion Syndrome**
Jill Arganbright, T. Blaine Crowley, Meghan Tracy, Janelle Noel-Macdonnell, Kimberly Gaiser, Lori Yaktine, Amanda Moore, Jaime Hamm, Bernice Morrow, Hansoo Song, Lisa Elden, Srivats Narayanan, Donna McDonald-McGinn
- 10:50 **54** **Age-Related Hearing Loss in Adults With Microdeletion 22q11.2 ***
Emma von Scheibler, Josine Widdershoven, Denise Breukels-van Barneveld, Nina Schröder, Thérèse van Amelsvoort, Agnies van Eeghen, Erik Boot
- 10:55 **55** **Tonsillectomy in Children With 22q11.2 Deletion Syndrome**
Jill Arganbright, Meghan Tracy, Srivats Narayanan, David Ingram
- 11:00 **56** **Ocular Findings in 22q11.2 Deletion Syndrome: A Systematic Literature Review and Results of a Dutch Multicenter Study ***
Emma von Scheibler, Emy van der Valk Bouman, Myrthe Nuijts, Noël Bauer, Tos Berendschot, Pit Vermeltfoort, Levinus Bok, Agnies van Eeghen, Michiel Houben, MD, Thérèse van Amelsvoort, Erik Boot, Michelle van Egmond-Ebbeling
- 11:05 **57** **Half of Patients with 22q11.2 Deletion Syndrome Develop Scoliosis and The Sagittal Spinal Shape at Early Age Predicts the Risk and Curve Type: A Prospective Cohort Study ***
Steven de Reuver, Tom Schlösser, Michiel Houben, Moyo Kruyt, René Castelein

- 11:10 Q&A
- 11:25 58 **Gastrointestinal Features of 22q11.2 Deletion Syndrome Across the Life Course**
Rebecca Kotcher, Daniel Chait, Jason Heckert, T.Blaine Crowley, Daniel McGinn, Elaine Zackai, Nitin Ahuja, Maria Mascarenhas, Donna M. McDonald-McGinn, James Reynolds
- 11:35 59 **Long-Term Growth Outcomes Following Gastrostomy Tube Feeding in Children With 22q11.2 Deletion Syndrome**
Lauren Lairson, Ian Campbell, T. Blaine Crowley, Alice Bailey, Daniel McGinn, Victoria Giunta, Oanh Tran, Beverly Emanuel, Elaine Zackai, Lorraine Katz, Maria Mascarenhas, Donna M. McDonald-McGinn
- 11:45 60 **Characteristics of Patients With 22q11.2DS Requiring Clinical and Instrumental Feeding Evaluation: A Retrospective Review**
Tracy Brundage, Thomas Randi, Emily Gallagher
- 11:50 61 **22q11.2 Deletion Syndrome: a New Model for the Study of Functional Gastrointestinal Diseases**
Rebecca Kotcher, Daniel Chait, Jason Heckert, T. Blaine Crowley, Daniel McGinn, Elaine Zackai, Nitin Ahuja, Maria Mascarenhas, Donna M. McDonald-McGinn, James Reynolds
- 12:00 62 **Nutrition-Related Issues and the Role of the Dietitian for Adults With 22q11.2 Deletion Syndrome ***
Samantha D'Arcy, Tracy Heung, Joanne Ha, Christina Blagojevic, Sarah Malecki, Lily Van, Maria Corral, Anne Bassett
- 12:10 63 **Very Early Onset Inflammatory Bowel Disease in Patients With 22q11.2 Deletion Syndrome**
Judith Kelsen, Soma Jyonouchi, Elizabeth Goldmuntz, Maria Mascarenhas, Donna M. McDonald-McGinn, Katheen Sullivan

12:15 Q&A

12:30 **Lunch – Spalatum Restaurant**

Session 7 *The Triad of DiGeorge Phenotypes - Grand Dalmacija Ballroom*

- 13:30 64 **Omic Approaches to the Analysis of the Dietary Modulation of Congenital Heart Defects by Vitamins A, Folic (B9) and B12, in a 22q11.2 Mouse Model**
Emi Amengual-Cladera, M.V. Llluc-Albertí, M. Solivellas-Pieras, J. Hernández-Rodríguez, V.J. Asensio, J. Muncunill, J. Rocha, A. Tubau, M. Juan, G. Lania, A. Baldini, C. Vives-Bauzá, Damian Heine-Suñer
- 13:40 65 **FGF8 Dosage Regulates Severity and Asymmetry of Pharyngeal and Cardiac Defects**
Jennifer Fish

- 13:50 **66** **Chromatin Regulatory Genes Identified in Individuals With 22q11.2DS are Implicated in Genetic Syndromes of Congenital Heart Disease and Developmental Abnormalities ***
Daniella Miller, Yingjie Zhao, T. Blaine Crowley, Elizabeth Goldmuntz, Donna M. McDonald- McGinn, Beverly Emanuel
- 14:00 **67** **Improved Outcomes in Patients With 22q11.2 Deletion Syndrome and a Perinatal Diagnosis of Interrupted Aortic Arch ***
Haley Ron, T. Blaine Crowley, Yichuan Liu, Marta Unolt, Erica Schindewolf, Julie Moldenhauer, Jack Rychik, Elizabeth Goldmuntz, Beverly Emanuel, Douglas Ryba, J. William Gaynor, Elaine Zackai, Hakon Hakonarson, Donna M. McDonald-McGinn
- 14:10 **68** **Aortic Root Dilation in Adolescents and Adults With 22q11.2 Deletion Syndrome ***
Carolina Putotto, Marta Unolt, Federica Pulvirenti, Ina Isufi, Gioia Mastromoro, Silvia Anaclerio, Luigi Tarani, Corrado Moretti, Bruno Marino, Paolo Versacci
- 14:20 **69** **Chromosome 22q11 Copy Number Variants and Single Ventricle Congenital Heart Disease**
Tracy Geoffrion, David Goldberg, T. Blaine Crowley, Jonathan Chen, Donna M. McDonald-McGinn, J. William Gaynor
- 14:25 **70** **Mild to Moderate Hypertriglyceridemia in Young Adults With a 22q11.2 Microdeletion ***
Christina Blagojevic, Tracy Heung, Sarah Malecki, Shengjie Ying, Sabrina Cancelliere, Robert A. Hegele, Anne S. Bassett
- 14:35 **71** **Influence of Rare and Common Genetic Variation on Dyslipidemia Risk in 22q11.2 Deletion Syndrome ***
Shengjie Ying, Tracy Heung, Bhooma Thiruvahindrapuram, Worrawat Engchuan, Christina Blagojevic, Robert Hegele, Zhaolei Zhang, Ryan Yuen, Anne Bassett
- 14:40 **72** **A Case Control Study of Cardiac Surgical Bleeding in Patients With 22q11.2DS**
T. Blaine Crowley, Ian Campbell, Abinaya Arulselvan, David Friedman, Char Witmer, Donna M. McDonald-McGinn, Michele Lambert
- 14:50 **Q&A**
- 16:00 **Afternoon Tea, Poster Viewing, and Wellness Breakout II – Grand Dalmacija Terrace, Grand Dalmacija Ballroom, and Korcula**
- 16:00 **Wellness Breakout II – Korcula**
Chair Massage
Greg Hedler
Maria Mascarenhas
Lisa Squires

- 16:30 73** **IPSc-Derived Thymic Epithelial Progenitor Cells From Patients with 22q11 Deletion Syndrome Display Cell Cycle Defects and Proinflammatory Polarization in a Cellular Model of Thymic Development**
Kelsea Hubka, Han Dan Nguyen, Abdulvasey Mohammed, Hui Gai, Georg Hollander, Maria Grazia Roncarolo, Vittorio Sebastiano, Katja Weinacht
- 16:40 74** **Humoral Immunity and Vaccine Responses in Children With 22q11.2 Deletion Syndrome**
Veronica Santilli, Carmela Giancotta, Donato Amodio, Lucia Pacillo, Emma Concetta Manno, Paola Zangari, Nicola Cotugno, Andrea Rotulo, Beatrice Rivalta, Silvia Di Cesare, Paolo Rossi, Andrea Finocchi, Paolo Palma, Caterina Cancrini
- 16:45 75** **Platelet Counts in Patients With 22q11.2DS Demonstrate Accelerated Decline in the First Two Decades of Life Compared to Non-Deleted Populations**
Ian Campbell, T Blaine Crowley, Donna M. McDonald-McGinn, Michele Lambert
- 16:55 76** **Pineoblastoma in a Nine Year-Old Boy With 22q11.2 Deletion Syndrome ***
Emi Rikeros Orozco, Diego Plaza Lopez de Sabando, Gloria Lopez Sobrino, Javier Saceda Gutierrez, Julio Guerrero Fernandez, Rebeca Rodriguez Peña, Sixto Garcia-Miñaur
- 17:00** **Q&A**
- 17:10 77** **Postoperative Hypocalcemia in Children With 22q11.2 Deletion Syndrome**
Jill Arqanbright, Meghan Tracy, Ashna Mahadev, Srivats Narayanan, Max Feldt
- 17:15 78** **Association Between Congenital Heart Defect and Growth in the 22q11.2 Deletion Syndrome**
Michiel Houben, Lisa Briel, Martijn Sliker
- 17:20 79** **Long-Term Growth Trajectories in 22q11.2 Deletion Syndrome**
Ian Campbell, T. Blaine Crowley, Rebecca Kotcher, Alice Bailey, Daniel E. McGinn, Oksana Jackson, Elizabeth Goldmuntz, Maria Mascarenhas, Lorraine Katz, Elaine H. Zackai, Donna M. McDonald-McGinn
- 17:25 80** **Results of Provocative Growth Hormone Testing in Patients With 22q11.2 Deletion Syndrome at The Children’s Hospital of Philadelphia**
Hamza Nasir, Ian Campbell, T. Blaine Crowley, Elaine H. Zackai, Donna M. McDonald-McGinn, Vaneeta Bamba, Lorraine Katz
- 17:30 81** **Adult Height, Deletion Extent and Short Stature in 22q11.2 Deletion Syndrome***
Brigid Conroy, Tracy Heung, Sarah Malecki, Joanne Ha, Erik Boot, Maria Corral, Anne Bassett
- 17:35 82** **Severe Hypothyroidism In 22q11.2 Deletion Syndrome**
Vaneeta Bamba, Ian Campbell, T. Blaine Crowley, Donna M. McDonald-McGinn, Edna Mancilla, Lorraine Katz
- 17:45** **Q&A**

- 17:55** **Wellness Close**
 Release Your Day Through a Moment of Mindfulness
Maria Mascarenhas
Philadelphia, PA, USA
- 18:00** **Adjourn**
- 19:30** **Poolside Dinner – Grand Dalmacija Terrace and Pool**
-

Friday, July 1

- 6:00** **Registration Open – Jim’s Foyer, Grand Dalmacija Ballroom**
Speaker Ready Room Open – Hvar

- 6:30** **Sunrise Yoga – Meet in Korcula at 6:25**

- 7:00** **Breakfast – Spalatum Restaurant**

Session 8 **From Genes to Brains and Back Again – Grand Dalmacija Ballroom**

- 8:00** **83** **Three Hits and Multiple Errors**
 Disrupted Development of Association Cortico-Cortical Circuits Due to 22q11.2DS
Anthony LaMantia
Blacksburg, Virginia
- 8:15** **84** **The Cerebellum Has It All**
 Cerebellar Dysfunction is Related to the Development of Psychiatric Symptoms - Lessons from Mouse Models
Stanislaw Zakharenko
Memphis, TN
- 8:30** **85** **An Alteration of Excitatory Inhibitory Balance Underlies Hippocampal Atrophy in Individuals with 22q11.2 Deletion Syndrome with Psychotic Symptoms ***
Valentina Mancini, Muhammad Saleh, Farnaz Delavari, Joëlle Bagaudtinova, Stephan Eliez
- 8:40** **86** **In Vivo Plasticity Between Ventral Hippocampal Inputs and Medial Prefrontal Cortex Microcircuits in a Mouse Model of 22q11.2 Deletion Syndrome**
David Kupferschmidt, Thomas Clarity, Rachel Mikofsky, Maxym Myroshnychenko, Madeline Hsiang, Sapphire Bowen-Kauth, Joshua Gordon
- 8:50** **Q&A**

- 9:05 **87** **Impaired Amygdala Sub-Field Functional Connectivity at Rest, Provides Insight for Pathways of Impaired Tolerance to Stress and Psychosis Vulnerability in Patients With 22q11DS ***
Farnaz Delavari, Corrado Sandini, Halima Rafi, Dimitri Van De Ville, Stephan Eliez
- 9:10 **88** **Explainable Artificial Intelligence Reveals Brain Fingerprints of Psychosis in 22q11.2 Deletion Syndrome Resembling Idiopathic Early Psychosis**
Kaustubh Supekar, Carlos de los Angeles, Srikanth Ryali, Leila Kushan, Charlie Schleifer, Tony Simon, Carrie Bearden, Vinod Menon
- 9:20 **89** **Quantitative Susceptibility Mapping of Deep Brain Nuclei in 22q11.2 Deletion Syndrome**
Nestor Muñoz, Marisleidys García, Analía Cuiza, Javiera Vasquez, Angeles Tepper, Juan Pablo Ramirez-Mahaluf, Barbara Iruretagoyena, Juan Aguirre, Daniella Barbagelata, Elisa Maldonado, Claudia Ornstein, Rosemarie Fritsch, Gabriela Repetto, Julio Acosta-Cabronero, Cristián Tejos, Nicolas Crossley
- 9:30 **90** **Transcriptional Response To Mitochondrial Dysfunction In Developing Cortical Projection Neurons ***
Abra Roberts, Daniel Meechan, Anthony LaMantia
- 9:40 **91** **Can Mitochondrial Measures Predict Schizophrenia Risk In 22q11.2 Deletion Syndrome?**
Jianping Li, Donna M. McDonald-McGinn, Beverly Emanuel, Raquel Gur, Stewart Anderson
- 9:45 **92** **Association Between Cortical Thickness and Executive Function in 22q11.2 Deletion Syndrome***
Maria Gudbrandsen, Clodagh M Murphy, Eileen Daly, Charlotte Blackmore, Maria Rogdaki, Leila Kushan, Declan G M Murphy, Carrie E Bearden
- 9:50 **Q&A**
- 10:00** **Coffee Break and Poster Viewing - Grand Dalmacija Terrace**
- 10:30 **93** **Transcriptional State and Epigenetic Regulation of Genes in Cortical Progenitors of the LgDel Model of 22q11.2DS**
Shah Rukh, Connor Siggins, Zachary Erwin, Anthony LaMantia, Thomas Maynard, Daniel Meechan
- 10:40 **94** **MicroRNA in 22q11.2 Deletion Syndrome and Risk to Develop Schizophrenia ***
Chuan Jiao, Qin HE, Anton Iftimovici, Oussama Kebir, Marie-Odile Krebs, Boris Chaumette
- 10:50 **95** **Interactions of Polygenic Risk Scores with Rare CNVs in Psychiatric Disorders and Quantitative Traits ***
Marieke Klein, Omar Shanta, Oanh Hong, Jeffrey MacDonald, Bhooma Thiruvahindrapuram, Agathe de Pins, Alexander Charney, Stan Letovsky, Jake Humphrey, Elise Douard, Sebastien Jacquemont, PGC ADHD working group, PGC ASD working group, PGC Bipolar Disorder working group, PGC Post-Traumatic-Stress Disorder Working Group, PGC Schizophrenia Working Group, PGC CNV working group, The Genes 2 Mental Health Network, Stephen Scherer, Jonathan Sebat

- 10:55 **96** **A Cumulative Phenotypic Impact of Sequence Variants in the 22q11.2 Region in Cohorts Without the Deletion**
Elemi Breetvelt, Anne Bassett, Peter Szatmari, Stephen Scherer, Jacob Vorstman
- 11:05 **97** **Identification of Biomarkers Predictive of Medical and Psychiatric Risk in Chromosome 22q11.2 Deletion Syndrome**
Flora Tassone, Marwa Zafarullah, Kathleen Angkustsiri, Lilith Marinho-Davis, Austin Quach, Seungjun Yeo, Blythe Durbin-Johnson, Heather Bowling
- 11:10 **Q&A**
- 11:20 **98** **Involvement of Inflammation and Blood-Brain Barrier Permeability in Cognitive Deficits and Psychosis in 22q11.2 Deletion Syndrome**
Michal Taler, Ehud Mekori-Domachevsky, Shira Dar, Elfi Vergaelen, Stephan Claes, Yaffa Serur, Yael Levy-Shraga, Ronnie Weinberger, Abraham Weizman, Ann Swillen, Doron Gothelf
- 11:30 **99** **Mechanisms Regulating The Neurovascular-Immune Interplay In the 22q11.2DS**
Alexis Crockett, Ryan Sean, Adriana Hernandez, Caroline Canning, Nicole Kanyuch, Hania Kebir, Guadalupe, Elaine Zackai, Donna M. McDonald-McGinn, Angela Viaene, Nail Benallegue, Raquel Gur, Stewart Anderson, Jorge Alvarez
- 11:40 **100** **Neuroinflammation in 22q11.2 Deletion Syndrome and Healthy Controls: A 7-Tesla MRS Study***
Chaira Serrarens, Claudia Vingerhoets, Desmond Hy Tse, Therese van Amelsvoort
- 11:45 **101** **Hypothyroidism and Risk of Schizophrenia in Individuals with 22q11.2 Deletion Syndrome***
Brigid Conroy, Tracy Heung, Eric Boot, Maria Corral, Lily Van, Sarah Malecki, Susan George, Anne Bassett
- 11:55 **102** **Dopamine, Glutamate and the Relation with Cognition in 22q11.2 Deletion Syndrome: A [18F]Fallypride Pet and 7-Tesla 1H-MRS Study ***
Claudia Vingerhoets, Desmond Hy Tse, Jenny Ceccarini, Esther van Duin, Jan Booij, MD, Therese van Amelsvoort
- 12:05 **103** **Neural Function in 22q11.2DS: Insights from Electrophysiological Studies**
Ana Alves Francisco, Michaela Cooke, Douwe J Horsthuis, John J Foxe, Sophie Molholm
- 12:10 **104** **Retinal Vessel Geometry: A Biomarker for Major Neurocognitive Decline in 22q11.2 Deletion Syndrome? ***
Emma von Scheibler, Abhishek Appaji, Tos Berendschot, Noël Bauer, MD, Naren Rao, Thérèse van Amelsvoort, MD, Agnies van Eeghen, Erik Boot
- 12:15 **Q&A**
- 12:30 Lunch – Spalatum Restaurant**

Session 9 Neuropsychological Profiles – Grand Dalmacija Ballroom

- 13:30 105 Longitudinal Follow-Up with Deep Phenotyping of Individuals with 22q11.2DS**
Raquel Gur, R. Sean Gallagher, Emily McClellan, Lauren White, Tyler Moore, Noah Hillman, Victoria Giunta, Kim Gaiser, Alice Bailey, Daniel E. McGinn, T. Blaine Crowley, Kosha Ruparel, Elaine H. Zackai, Ruben Gur, Donna M. McDonald-McGinn
- 13:40 106 Stress Sensitivity in Youth with 22q11.2 Deletion Syndrome: Associations with Psychopathology and Coping Strategies ***
Laura Ilen, Clémence Feller, Stephan Eliez, Eva Micol, Farnaz Delavari, Carmen Sandi, Olivia Zanoletti, Maude Schneider
- 13:45 107 Post-Traumatic Stress in Adults with 22q11.2 Deletion Syndrome ***
Emma von Scheibler, Thérèse van Amelsvoort, Claudia Vingerhoets, Agnies van Eeghen, Erik Boot
- 13:55 108 In-Person Compared to Remote Neurocognitive Assessments of Individuals with 22q11.2DS**
Ruben Gur, Raquel Gur, MD, Tyler Moore, Donna M. McDonald-McGinn, Lauren White, R. Sean Gallagher, Noah Hillman, Emily McClellan, T. Blaine Crowley
- 14:00 109 Neurodevelopmental Outcome in 1½-3½ Year Old's with 22q11DS: Effect of Birth Weight, Gestational Age, Head Circumference, Gender and Congenital Heart Disease**
Sasja Duijff, Elemi Breetvelt, Michiel Houben, Ania Fiksinski, Jeroen Breckpot, Jacoba Louw, Ann Swillen
- 14:10 110 Executive Functioning In Preschool Children With 22q11.2 Deletion Syndrome and the Effect of Congenital Heart Defects ***
Emma Everaert, Tessel Boerma, Iris Selten, Ellen Gerrits, Michiel Houben, Jacob Vorstman, Frank Wijnen
- 14:15 111 The Complete IQ-Profile Over the Lifespan in Individuals with 22q11.2DS: An IBBC Study ***
Ania Fiksinski, Rielle Wierenga, Iris Selten, International 22q11.2DS Brain and Behavior Consortium, Jacob Vorstman, Therese van Amelsvoort
- 14:25 112 Cognitive Profiles of Individuals with 22q11.2 Deletion Syndrome Using Latent Profile Analysis and Their Relationship to Psychotic Symptoms**
Stephen Hooper, Vandana Shashi
- 14:35 113 Cognitive, Adaptive and Daily Life Functioning in Adults with 22q11.2 Deletion Syndrome**
Claudia Vingerhoets, Elfi Vergaelen, Emma von Scheibler, Annick Vogels, Erik Boot, MD, Therese van Amelsvoort, Ann Swillen
- 14:45 Q&A**

- 15:00 **114** **Factors Influencing Trajectory of Language Development in Children with 22q11.2 Deletion Syndrome**
Cynthia Solot, Jordan Armstrong, T Blaine Crowley, Kimberly Gaiser, Victoria Giunta, Alice Bailey, Daniel McGinn, Oanh Tran, Tyler Moore, Emily McClellan, R. Sean Gallagher, Beverly Emanuel, Elaine Zackai, Oksana Jackson, Sarah Hopkins, Madeline Chadehumbe, Edward Moss, Reuben Gur, Raquel Gur, Donna M. McDonald-McGinn
- 15:10 **115** **Comparison of Language Skills Between Children with 22q11.2 Duplication and Children with 22q11.2 Deletion Syndrome ***
Jente Verbesselt, Ellen Van Den Heuvel, Inge Zink, Jeroen Breckpot, Ann Swillen
- 15:15 **116** **Assessment of Speech and Language in 61 Children with 22q11.2 Duplication Syndrome**
Cynthia Solot, T. Blaine Crowley, Victoria Giunta, Kimberly Gaiser, Alice Bailey, Daniel E. McGinn, Jordan Armstrong, Oanh Tran, Emanuel S. Beverly, Elaine H. Zackai, Oksana Jackson, Donna M. McDonald-McGinn
- 15:20 **117** **Autism-Related Behaviors in Preschool Children with 22q11.2 Deletion Syndrome: The Impact of Language Abilities ***
Iris Selten, Tessel Boerma, Emma Everaert, Ania Fiksinski, Ellen Gerrits, Michiel Houben, Frank Wijnen, Jacob Vorstman
- 15:25 **118** **Weighted Gene Co-Expression Network Analysis Identifies Networks Related to Autism Spectrum Disorder and Psychosis in 22q11.2 Deletion Syndrome**
Michiel Houben, Alexandra de Reus, Janneke Zinkstok, Marco Boks
- 15:30 **119** **ASD-Associated Symptom Profiles in Dutch Individuals with 22q11.2DS and Subsequent Psychosis ***
Tessel Boerma, Nadia van Wijngaarden, Manik Djelantik, Iris Selten, Jacob Vorstman, Janneke Zinkstok, Ania Fiksinski
- 15:35 **120** **A Dimensional Perspective on Neurodevelopmental Phenotypic Expression in Adolescents with 22q11.2 Deletion Syndrome**
Iris Selten, Jill Blok, Manik Djelantik, Michiel Houben, Jacob Vorstman, Frank Wijnen, Janneke Zinkstok, Ania Fiksinski
- 15:40 **121** **Psycho-Educational Parent Groups to Optimise Communication Skills of Young People with 22q11.2 Deletion Syndrome ***
Marie Fahy
- 15:45 **Q&A**
- 16:00 **Afternoon Tea, Poster Viewing, and Wellness Breakout III - Grand Dalmacija Terrace, Grand Dalmacija Ballroom, and Korcula**

Wellness Breakout III - Korcula

Aromatherapy

Greg Hedler

Maria Mascarenhas

Lisa Squires

Session 10 Bench to Bedside – Grand Dalmacija Ballroom

- 16:30 122 Translational Medicine**
Clinical Trials Should Not Overlook the Multidisciplinary Nature of 22q11.2DS
Madeline Chadehumbe
Philadelphia, PA, USA
- 16:45 123 A Randomized Phase 2 Clinical Trial of Fasoracetam in Children with 22q11.2DS**
Madeline Chadehumbe, Nancy Butcher, Emily Gallagher, Raquel Gur, Hakon Hakonarson, Donna M. McDonald-McGinn, Naomi Meeks, Jacob Vorstman
- 16:50 124 Selective GSK3A and ? Inhibition During Early Postnatal Development Alter Working Memory-Related Behavior in a Sex-Biased Manner in a Mouse Model of 22q11.2 Deletion Syndrome**
Johannes Passecker, Chloe Aloimonos, Chia-Yuang Chang, Aleksandra Dagunts, Maxym Myroshnychenko, David Kupferschmidt, Joseph Gogos, Joshua Gordon
- 17:00 125 The Odds of Small Things: Phenotypic Variation as a Biological Consequence of 22q11.2 Gene Deletion During Neural Circuit Differentiation**
Anthony Lamantia
- 17:10 126 Treatment of Schizophrenia in 22q11.2 Deletion Syndrome ***
Lily Van, Anne Bassett
- 17:20 127 Effects of Risperidone Treatment on Psychotic Symptomatology and Cognitive Functions in 22q11.2 Deletion Syndrome: Preliminary Results from a Clinical Trial ***
Caren Latreche, Johanna Maeder, Valentina Mancini, Maude Schneider, Stephan Eliez
- 17:35 128 Which Environmental Factors are Associated with Prodromal Psychotic Symptoms in Individuals with 22q11.2 Deletion Syndrome? ***
Seline Ismail-Sutton, Sinead Morrison
- 17:40 129 Piloting The Peers© Programme With Young Adult Women With 22q11.2Ds In Ireland**
Lorna Kerin, Fiona McNicholas
- 17:45 Q&A**
- 18:00 Future Considerations**
Peter Scambler
London, UK

- 18:15 Junior Investigator Awards**
Presented by 2018 Award Recipient Lisanne Vervoort
- 18:20 Closing Remarks and Announcement of Future Meeting**
22q11.2 Society Trustees
Donna M. McDonald-McGinn, Anne Bassett, Bernice Morrow, Peter Scambler, Ann Swillen
- 18:25 Wellness Close**
Release Your Week Through a Moment of Mindfulness
Maria Mascarenhas
Philadelphia, PA, USA
- 18:30 Meeting Adjourned**
-

Poster Presentations:

* *Junior Investigator*

Top Scoring Posters

- ❖ *Odd numbered posters – authors present on Wednesday (June 29th)*
- ❖ *Even numbered posters – authors present on Thursday (June 30th)*

- 130 A Novel Non-Allelic Homologous Recombination Event in a Parent with the Recurrent T(11;22) Leading to 22q11.2DS *
*Oanh Tran, Steven Pastor, Donna M. McDonald-McGinn, Daniel McGinn, Alice Bailey, Elaine Zackai, Beverly S. Emanuel***
- 131 Genotype-First Mental Health Care: Opportunities, Challenges and How 22q11.2DS Shows The Way *
*Danielle Baribeau, Polina Perlman, Jane Summers, Jacob Vorstman***
- 132 Coincidence of Two Microdeletion Syndromes Associated with Congenital Heart Disease: First Case Report of a Patient Diagnosed with Kleeftstra and Digeorge Syndromes *
*Zarand Nemethi, Fanni Szumutku, Irén Haltrich, Árpád Ferenc Kovács***
- 133 Case Study: Deep Phenotyping of a Case with Paternally Inherited 22q11.2 Duplication and 16p11.2 Duplication *
*Jente Verbesselt, Inge Zink, Jeroen Breckpot, Ann Swillen***
- 134 Successful Treatment with Mycophenolate Mophetile of Relapsing/Refractory Immune Thrombocytopenia in Four Patients with Del22q11.2 Syndrome * #
*Lucia Pacillo Carmela Giancotta, Donato Amodio, Silvia Di Cesare, Cristina Cifaldi, Beatrice Rivalta, Emma Concetta Manno, Veronica Santilli, Giuseppe Palumbo, Paolo Rossi, Paolo Palma, MD, Andrea Finocchi, Caterina Cancrini***

- 135 Intraocular Manifestations in 22q11.2 Deletion Syndrome**
Joseph Martel, Samyuktha Melachuri, Marjan Imani Fooladi
- 136 The Validation Of The Medical Burden Scale in Individuals with 22q11.2 Deletion Syndrome ***
Noam Matalon, Ronnie Weinberger, Michal Taler, Doron Gothelf
- 137 Granulomatous-Lymphocytic Interstitial Lung Disease (Glild) vs Sarcoidosis in a Patient with 22q11.2 Deletion Syndrome: A Case Report**
Matthew Blessing
- 138 Use Of Social Media Amongst The 22q11.2Ds Community #**
Kaylee Paulsgrove, Emily Gallagher
- 139 Multilayer Network Signatures of Daily-Life Psychopathology Dynamics Differentiate 22q11.2 Deletion Syndrome from ASD and Healthy Controls ***
Corrado Sandini, Andrea Imparato, Clemence Feller, Maude Schneider
- 140 Decoding Intentions From Movement: An Embedded Approach of Social Cognition In Neurodevelopmental Disorders ***
Clemence Feller, Novella Pretti, Laura Ilen, Stephan Eliez, Cristina Becchio, Maude Schneider
- 141 Physiologically Relevant Levels of Reactive Oxygen Species Acutely Disrupt Neuronal Network Function. #**
Gregg Crabtree
- 142 EEG Microstate Dynamics in Individuals with 22q11.2Ds with and without Psychotic Symptoms**
Douwe J. Horsthuis, Michaela Cooke, Sophie Molholm, Ana Alves Francisco
- 143 Moodstratification: Psychiatric Symptoms, the Immune System and Physical Fitness In 22q11.2DS***
Kris Van de Woestyne, Stephan Claes, Ann Swillen, Elske Vrieze
- 144 Neurocognitive and Sensorimotor Performance in 22q11DS and their Relationship to Psychiatric Clinical Symptoms ***
David Parker, Opal Ousley, Sid Imes, Bruce Cuthbert, Evelt Henshey, Nicholas Massa, MPH6 Elaine Walker, Erica Duncan, Joseph Cubells
- 145 A Case Study of Pediatric Catatonia in 22q11.2DS**
Kerry Conant
- 146 Grammatical Skills of Dutch Children with 22q11.2 Deletion Syndrome in Comparison with Children with Developmental Language Disorder ***
Tessel Boerma, Maaïke Steggink, Dinte Vlieger, Emma Everaert, Iris Selten, Ellen Gerrits, PhD, Michiel Houben, Jacob Vorstman, Frank Wijnen
- 147 Developing a Multidisciplinary 22q Infant Assessment Clinic to Assess Motor and Cognition in Infants with 22q11.2DS ***
Cindy Ola, Tracy Brundage, Emily Gallagher

148 Evaluating a Remote Dialogic Reading Intervention Delivered to Caregivers of Children with 22q11.2 Deletion Syndrome *#

Kimberly Barajas, Cindy Ola, Yesenia Garcia, Jenny Thach, Emily Ghallagher, Emily Myers, Brent Collett

149 Care4carers: Raising a Child with Extra Challenges: A Mindful Parenting Training for Parents with a Child with 22q11.2DS #

Sasja Duijff, Linda Campbell

150 Applied Behavior Analysis: A Promising Treatment for Behavior Problems and Functional Deficits in 22q11.2Ds #

Yaffa Serur, Eitan Eldar, Keren Englander, Rachely Mazor, Doron Gothelf

