



14th Biennial International 22q11.2 Conference

Zeus Eleva Mirabello Bay Hotel
Crete, Greece

Tuesday, July 14 through Friday, July 17, 2026

www.22qsociety.org

Tuesday, July 14

6:00 *REGISTRATION OPEN – ZEUS BALLROOM COURTYARD*

7:00 *BREAKFAST – AMALTHEA RESTAURANT AND TERRACE
MIRABELLO BAY RESORT GUESTS ONLY*

SESSION I OPENING CEREMONIES - ZEUS BALLROOM

Chairs: Donna M. McDonald-McGinn and Erik Boot

8:00 **The 22q11.2 Society Welcomes a Parade of Nations to the Jewel of Greece**
Donna M. McDonald-McGinn, 22q11.2 Society Chair

Kalos Irthes stin Ellada!

Spyros Petrakis

Program Synopsis and Acknowledgements

Erik Boot, 22q11.2 Society Trustee and 2026 Program Chair

Presentation of the Angelo DiGeorge Memorial Medal of Honor

Ann Swillen, 22q11.2 Society Trustee and Secretary

Presentation of the Clodagh Murphy Memorial Unsung Hero Award

Donna McDonald-McGinn, 22q11.2 Society Chair

Opening Breath

Maria Mascarenhas, Philadelphia, PA, USA

SESSION II GENOTYPES, PHENOTYPES, AND MECHANISMS

Chairs: Peter Scambler and Joris R. Vermeesch

- 8:25 **001** **PETER SCAMBLER INVITED LECTURE AND OUTSTANDING ACHIEVEMENT AWARD**
Unraveling the role of segmental duplicons in predisposition and phenotypic variability of the 22q11.2 deletion syndrome
Joris R. Vermeesch, Leuven, Belgium
Award presented by Anne Bassett, 22q11.2 Society Trustee and Treasurer
- 8:50 **002** **Haplotype-resolved iso-seq reveals allele-specific isoform remodeling in 22q11.2 deletion patient-parent duos ***
Marta Sousa Santos, Erika Souche, David Porubsky, Natalia Olszewska, Evan E. Eichler, and Joris R. Vermeesch
- 8:57 **003** **Isogenic LCR22 deletions in H9 define an isoform-resolved transcript catalogue and LCR-specific regulatory programs across early development ***
Marta Sousa Santos, Erika Souche, Natalia Olszewska, and Joris R. Vermeesch
- 9:05 **004** **Resolving the exact breakpoints of the 22q11.2 deletion using CRISPR-Catch and long-read sequencing reveals chromosomal rearrangement mechanisms and individual variance in breakpoints**
Alexander E. Urban, Bo Zhou, Carolin Purmann, Hanmin Guo, GiWon Shin, Yiling Huang, Reenal Pattni, Qingxi Meng, Stephanie U Greer, Tanmoy Roychowdhury, Raegan N Wood, Marcus Ho, Heinrich Zu Dohna, Alexej Abyzov, Joachim F Hallmayer, Wing H Wong, and Hanlee P Ji
- 9:14 **005** **Optical mapping of nested 22q11.2 deletions reveals conserved structural architectures**
Steven Pastor, Oanh Tran, T. Blaine Crowley, Audrey Olson, Lydia Rockart, Daniel E. McGinn, Elaine H. Zackai, Donna M. McDonald-McGinn, and Beverly S. Emanuel
- 9:20 **Q&A**
- 9:30 **006** **INVITED PRESENTATION**
Setting the stage: The importance of deletion size, variants on the intact chromosome 22q11.2 allele, dual diagnoses, and genome-wide modifiers
Donna M. McDonald-McGinn, Philadelphia, PA, USA
- 9:45 **007** **Do proximal nested 22q11.2 deletions have a modified adult phenotype?**
Anne S. Bassett, Tracy Heung, Lisa Palmer, Samantha D'Arcy, Sarah Malecki, and Nikolai Reyes
- 9:55 **008** **Clinical phenotypes of 22q11.2 typical and atypical deletions and duplications**
Thérèse van Amelsvoort, Emma von Scheibler, Rianne van Roij, Winde Mercken, Tjitske Kleefstra, and David Linden

10:02 **009** **Genotype-phenotype correlations in 22q11.2 deletion syndrome: unravelling the clinical heterogeneity**
Deepthi Jyothish, Elizabeth Brettle, Hannah Titheradge, Asma Hamad, Neha Gupta, Hakim-Moulay Debhi, and Charlotte Clarke

10:10 **010** **22q11.2-22q11.23 distal deletions and duplications: An emerging important population ***
Daniel E. McGinn, T. Blaine Crowley, Audrey Olson, Lily N. Elman, Evie Andrewes, Lydia Rockart, Oanh Tran, Beverly S. Emanuel, Elaine H. Zackai, and Donna M. McDonald-McGinn

10:20 **Q&A**

10:30 **COFFEE BREAK AND POSTER VIEWING – COVERED TERRACE**

SESSION III EXPRESSION, METHYLATION, POLYGENIC RISK SCORES, AND EXPOSOME

Chairs: Anne Bassett and Beata Nowakowska

11:10 **011** **INVITED PRESENTATION**
Expression and methylation studies in 22q11.2 deletion syndrome
Beata A. Nowakowska, Warsaw, Poland

11:25 **012** **The 22q11.2 methylome ***
Senne Meynants, Erika Souche, Marta Sousa Santos, Nicolas Dierckxsens, Ann Swillen, Elfi Vergaelen, Jeroen Breckpot, and Joris R. Vermeesch

11:35 **013** **Impact of ancestry on prevalence of 22q11.2 deletion syndrome ***
Lydia Rockart, T. Blaine Crowley, Daniel E. McGinn, Audrey Olson, Oanh Tran, Steven Pastor, Laura Schultz, Laura Almasy, Raquel Gur, Kai Wang, Ian M. Campbell, Kathleen Valverde, Elaine H. Zackai, Sulagna C. Saitta, Loydie A. Jerome-Majewska, Beverly S. Emanuel, and Donna M. McDonald-McGinn

11:45 **014** **Demographic, socioeconomic, and clinical profiles of individuals with 22q11.2 deletion and duplication syndromes: a large multi-institutional EHR study**
Hanadys Ale, Marta Telatin, Brianna Santiago, and Jennifer Keelin

11:55 **015** **An exposome-wide analysis of environmental predictors of IQ in 22q11.2 and 16p11.2 CNV carriers ***
Yelyzaveta Snihirova, Sinan Guloksuz, David EJ Linden, Ann Swillen, Marianne BM van den Bree, Raquel E Gur, Ruben C Gur, Carrie E Bearden, Jacob AS Vorstman, Jonathan Sebat, Sébastien Jacquemont, Donna McDonald-McGinn, Anne S Bassett, Mieke van Haelst, Claudia Vingerhoets, and Thérèse van Amelsvoort

- 12:05 **016** **Characterizing environment and its relationship to psychopathology in 22q11.2 deletion syndrome**
Arielle Ered, Tyler M. Moore, Austin T. Moran, Claudine B. Van Arman, Alyssa M. Neppach, Mia Schwartzberg, R. Sean Gallagher, Simon Smerconish, Kosha Ruparel, Scott Troyan, Amy Cassidy, Lauren K. White, T. Blaine Crowley, Daniel E. McGinn, Audrey Olson, Oanh Tran, Elaine H. Zackai, Beverly S. Emanuel, Ruben C. Gur, Donna M. McDonald-McGinn, and Raquel E. Gur
- 12:05 **017** **Combined and differential effects of polygenic, socioeconomic, and developmental markers in individuals with 22q11.2 and 16p11.2 copy number variants**
Vincent-Raphaël Bourque, Anjali Srinivasan, Samuel J R A Chawner, Laura Almasy, Laura Schultz, Ruben Gur, Marianne B M van den Bree, Donna M McDonald-McGinn, Therese van Amelsvoort, Ann Swillen, Carrie E Bearden, Raquel E Gur, Genes to Mental Health Network (G2MH), Jonathan Sebat, Jacob A S Vorstman, and Sébastien Jacquemont
- 12:25 **Q&A**
- 12:35 **018** **Investigating the non-deleted chromosome 22q11.2 allele ***
Lily N. Elman, Audrey Olsen, Evie Andrewes, Lydia Rockart, Lydia Hamilton, T. Blaine Crowley, Bekah Wang, Daniel E. McGinn, Beverly S. Emanuel, Elaine H. Zackai, Beata Nowakowska, and Donna M. McDonald-McGinn
- 12:42 **019** **Autosomal recessive conditions unmasked by 22q11.2 deletion: a population-level analysis using a national EHR network**
Hanadys Ale, Jessica Colon, Jorge Haddad Gomez, Emily Woolhiser, Andres Wong, Virna Kneller, and Shwetha Maharaj
- 12:49 **020** **Screening for *TANGO2* and other autosomal recessive conditions in individuals with 22q11.2 deletion syndrome**
Valerie J. Allegretti, Jaime C. Duncan, Laura H. Swibel Rosenthal, and Kelly E. Regan-Fendt
- 12:56 **021** ***TANGO2* deficiency with 22q11.21 microdeletion: metabolic and neuropsychiatric considerations**
Valerie J. Allegretti, Renee J. Wakulski, Joshua J. Baker, Laura H. Swibel Rosenthal, and Kelly E. Regan-Fendt
- 13:00 **022** **22q and *TANGO2* - Understanding *TANGO2* deficiency disorder via genotype-phenotype correlations in 22q11.2 deletion syndrome**
Beata A. Nowakowska, Maciej Geremek, Theresa A. Grebe, Robert Śmigiel, T. Blaine Crowley, Daniel E. McGinn, Audrey Olson, Raquel Gur, Bernice Morrow, Anne Bassett, and Donna M. McDonald-McGinn

13:10 023 Clinical consensus guidelines for acute management of TANGO2 deficiency disorder
Samuel J. Mackenzie, Ashton B. Ariola, Deena L. Chisholm, Jonathan Scaccia, Mike Morris, Nourhan Suleiman, Sebastian C. Tume, Svjetlana Tisma-Dupanovic, Miriam Hull, Donna M. McDonald-McGinn, Scott Ceresnak, Angela L. Hewitt, Elizabeth G. Ames, Kimberly M. Houck, Alyssa Smith, Seema R. Lalani, Megha K. Kanjia, Georgia Sarquella-Brugada, Christina Y. Miyake, Divakar S. Mithal, Kuntal Sen, Joshua K. Meisner, and Lina Ghaloul-Gonzalez

13:20 Q&A

13:30 LUNCH – AMALTHEA RESTAURANT AND TERRACE
ALL CONFERENCE ATTENDEES AND ACCOMPANYING PERSONS

SESSION IV CARDIAC DEVELOPMENT, PRENATAL DIAGNOSIS, & THE IMPORTANCE OF TBX1
Chairs: Natalie Blagowidow and Bernice Morrow

14:30 024 INVITED PRESENTATION
Multifactorial role of Tbx1 in the second heart field
Sophie Astrof, Newark, NJ, USA

14:45 025 INVITED PRESENTATION
Tbx1 acts upstream of distal enhancers in the embryonic mesoderm for cardiac outflow tract formation
Bernice E. Morrow, Bronx, NY, USA

15:00 Q&A

15:05 026 Broad phenotype of congenital heart disease in fetuses and infants with 22q11.2 deletion syndrome: an international cohort study *
Marco Masci, Stephanie Galloway, Ronald Wapner, Alessandra Toscano, T. Blaine Crowley, Elizabeth Goldmuntz, Anne S. Bassett, Donna M. McDonald McGinn, and Lindsay R. Freud

15:15 027 Primary pulmonary artery anomalies in 22q11.2 deletion syndrome *
Cristina Angellotto, T. Blaine Crowley, Daniel E. McGinn, Marta Unolt, Carolina Putotto, Elaine H. Zackai, Cristina Digilio, Paolo Versacci, Jill Savla, Robert Kelly, Bernice Morrow, Bruno Marino, and Donna M. McDonald-McGinn

15:25 028 Branch pulmonary artery anomalies in 22q11 deletion-diagnosis, management and medium-term outcomes – a case series
Sanket S. Shah, Manbir Sanghera, Matthew M. Feldt, Julie Martin, Maria Kiaffas, and Nikita Raje

15:30 Q&A

15:40 **029** Prenatal diagnosis of 22q11.2 deletion syndrome: What can we learn about pregnancy options from the international 22q11.2 PVPOS study?

Natalie Blagowidow, T. Blaine Crowley, Stephanie Galloway, Daniel E. McGinn, Kimberly Gaiser, Oahn Tran, Elaine H. Zackai, Beverly S. Emanuel, Erica Schindewolf, Julie Moldenhauer, PVPOS Consortium, Qi Yan, Ron Wapner, Anne S. Basset, Lindsay R. Freud, and Donna M. McDonald-McGinn

15:50 **030** Prenatal diagnosis and outcomes in a large cohort of patients with 22q11.2 copy number variants *

Daniel E. McGinn, T. Blaine Crowley, Renee Wright, Audrey Olson, Oanh Tran, Beverly S. Emanuel, Elaine H. Zackai, Natalie Blagowidow, Nahla Kahlek, and Donna M. McDonald-McGinn

16:00 **031** The impact of non-invasive prenatal testing (NIPT) for 22q11.2 copy number variants

Donna M. McDonald-McGinn, T. Blaine Crowley, Daniel E. McGinn, Audrey Olson, Oanh Tran, Beverly S. Emanuel, Elaine H. Zackai, Anne Bassett, and Natalie Blagowidow

16:10 **032** Cell-free DNA screening for the 22q11.2 microdeletion: disparities in access to testing

Katelyn Hashimoto, Patrick Kelly, Brittany Prigmore, Kathleen D. Kueck, Thea N. Golden, and Sheetal Parmar

16:20 Q&A

16:30 AFTERNOON TEA AND POSTER VIEWING – COVERED TERRACE

SESSION V GENOMICS, DIET, VITAMINS, AND THE MICROBIOME

Chairs: Maria Mascarenhas and Irene Zohn

17:00 **033** Reduced dosage of *Kmt2d* modifies *Tbx1* haploinsufficiency towards phenotypes of 22q11.2DS *

Daniella Miller, Kevyn Jackson, Timothy C. Cox, and Bernice E. Morrow

17:10 **034** Mechanisms underlying variability in 22q11.2 deletion syndrome

Tatiana Ferebee, Pooja Chauhan, Elias Oxman, and Irene Zohn

17:20 **035** Nutritional implications for genetic counseling in 22q11.2 deletion syndrome: evidence from a mouse model *

Maria Victòria Llull-Albertí, Emilia Amengual-Cladera, Angelika Merkel, Jessica Hernandez-Rodriguez, Marc Ventayol-Guirado, Juan Antonio Jimenez-Barceló, Jairo Enrique Rocha, Aina Vaquer-Picó, Víctor José Asensio-Landa, Laura Torres-Juan, Fernando Santos-Simarro, María García de Paso-Mora, Iciar Martínez-López, and Damian Heine-Suñer

* Junior Investigator

- 17:30 036 Studying vitamin A signaling in human heart models of 22q11.2DS**
Conchi M. Estaras, Emily Megill, Eleonora Stronati, Elizabeth Abraham, Aidan Douglas, and Mikel Zubillaga
- 17:40 037 Dietary intake patterns in children with 22q11.2 deletion syndrome: widespread micronutrient imbalances revealed following 72-hour dietary recall analysis**
Luc Santilli, Alain J. Benitez, Audrey Olson, Lydia Rockart, Robin Miccio, Lauren White, Donna McDonald-McGinn, and Maria Mascarenhas
- 17:50 038 Vitamin B12 status in adults with 22q11.2 deletion syndrome**
Samantha D'Arcy, Tracy Heung, Lisa Palmer, Nikolai Reyes, and Anne S. Bassett
- 18:00 039 First characterization of the gut microbiome in children with 22q11.2 deletion syndrome**
Alain J. Benitez, Luc Santilli, Scott G. Daniel, Audrey Olson, Lydia Rockart, Robin Miccio, Lauren White, Donna McDonald-McGinn, and Maria Mascarenhas
- 18:10 040 Microbiota–Metabolite Remodeling Links *Tbx1* Haploinsufficiency to Brain Dysfunction in 22q11.2 Deletion Syndrome**
Stefano Leo, Sabrina Bianco, Sara Cioffi, Gemma Flore, Sara Veneziano, Michele Costanzo, Marianna Caterino, Margherita Ruoppolo, Lucia Mele, Lucia Marra, Marchesa Bilio, Rosa Ferrentino, Elizabeth Illingworth, Antonio Baldini, and Gabriella Lania
- 18:20 Q&A**
- 18:30 Adjourn – Day 1**
- 20:00 *WELCOME RECEPTION - GRECIAN CAVE TERRACE***
GROUP PHOTO, SIGNATURE COCKTAIL, AND HEAVY HORS D'OEUVRES
ALL CONFERENCE ATTENDEES AND ACCOMPANYING PERSONS



Wednesday, July 15

6:00 REGISTRATION OPEN – ZEUS BALLROOM COURTYARD

7:00 BREAKFAST – AMALTHEA RESTAURANT AND TERRACE
MIRABELLO BAY RESORT GUESTS ONLY

SESSION VI IMMUNE ACTIVATION, DYSREGULATION & INFLAMMATION: A NEW PARADIGM

Chairs: Hanadys Ale and Elfi Vergaelen

- 8:00 **041** **INVITED PRESENTATION**
Primate-specific RNA dysregulation and innate immune activation in 22q11.2 deletion syndrome
Sara Heras, Granada, Spain
- 8:15 **042** **Immunologic correlates of psychotic illness in 22q11.2 deletion syndrome**
Anne S. Bassett, Katherine Beigel, Kelly Maurer, Nouf Alsaati, Raquel Gur, T. Blaine Crowley, Donna McDonald McGinn and Kathleen E. Sullivan
- 8:25 **043** **Haploinsufficiency-Driven RNA Metabolism is Associated with Immune Dysregulation and Psychosis Risk in 22q11.2 Deletion Syndrome ***
Christina Michalski, David A. Parker, Leah Cheng, Mojahidul Islam, Sidney Imes, Gabrielle Ruban, Heath Dunlop, Bruce Cuthbert, Brett Henshey, Grace Lee, Nicholas M. Massa, Opal Y. Ousley, Elaine F. Walker, David R. Goldsmith, Joseph F. Cubells, Erica Duncan, Bradley D. Pearce, Ashish A. Sharma and Zhexing Wen
- 8:32 **044** **Discriminating psychosis in 22q11.2 deletion syndrome using monocyte inflammation-related gene expression ***
Emma Saillart, Elfi Vergaelen, Stephan Claes, Elske Vrieze and Ann Swillen
- 8:39 **045** **Immune dysregulation in chromosome 22q11.2 related disorders: insights from the USIDNet registry**
Nikita Raje, Vaibhavi Vichare, Roshini Abraham, Ivan Chinn, Megan A. Cooper, Ramsay Fuleihan, Avni Y. Joshi, Monica G. Lawrence, Rebecca Marsh, Luigi Notarangelo, Jennifer M. Puck, Charlotte Cunningham Rundles, Kelly Walkovich and Kathleen Sullivan
- 8:46 **046** **Immune system activation following trauma in 22q11.2 deletion syndrome**
Adi Drapisz, Yana Paltavets, Shira Dar, Doron Gothelf and Michal Taler
- 8:50 **Q&A**

- 9:00 **047** **Identification of whole gene deletion of *TBX1* with autoimmune cytopenias in a large pediatric cohort**
Emily M Harris, Amanda B Grimes, Yi-Lee Ting, Taylor Kim, Rachael F Grace, Kristin Shimano and Michele P. Lambert
- 9:08 **048** **Immune cytopenias in a large cohort of patients with 22q11.2DS: management, outcomes and unique considerations**
T. Blaine Crowley, Praharsha Konde, Samantha Gaerian, Donna McDonald-McGinn and Michele P. Lambert
- 9:15 **049** **Proteome wide autoantibody profiling in the 22q11.2 deletion syndrome**
Jenny Lingman Framme, Viktoria Hennings, Anna-Carin Lundell, Karolina Thörn, Christina Lundqvist, Susanne Lindgren, Vanja Lundberg, Esbjörn Telemo, Anders Fasth, Sólveig Óskarsdóttir and Olov Ekwall
- 9:22 **050** **Spatial multiomic studies of the thymus in individuals with 22q11.2 deletion syndrome**
Viktoria Hennings, Jenny Lingman Framme, Sólveig Óskarsdóttir and Olov Ekwall
- 9:28 **051** **Investigation of autoimmune alterations in 22q11.2 deletion syndrome in a single Brazilian center: preliminary results from 17 individuals ***
João Pedro Moretti and Vera Lúcia Gil-da-Silva-Lopes
- 9:32 **052** **Cardiac complexity predicts increased immune diagnostic burden in 22q11.2 deletion syndrome ***
Jasmyn Atalla, Blaine Crowley, Kathleen Sullivan, Jennifer Heimall Donna M. McDonald-McGinn and Timothy Buckey
- 9:39 **053** **Platelet function and bleeding risk in adults with 22q11.2 deletion syndrome: a cross-sectional investigational study ***
Emma N.M.M. von Scheibler, Jeltje C.C. Spapens, Magdolna Nagy, Irene M.L.W. Keularts-Körver, Floor C.J.I. Heubel-Moenen, Johan W.M. Heemskerk, Agnies M. van Eeghen, Thérèse A.M.J. van Amelsvoort and Erik Boot
- 9:45 **Q&A**
- 9:55 **054** **Immunologic outcomes following thymectomy in patients with 22q11.2 deletion syndrome**
Anne Jennings, T. Blaine Crowley, Audrey Olson, Lydia Rockart, Daniel E. McGinn, Oanh Tran, Elaine H. Zackai, Beverly S. Emanuel, J. William Gaynor, Kathleen E. Sullivan, Kathleen Valverde, and Donna M. McDonald-McGinn
- 10:02 **055** **TLB-101, a GMP allogeneic iPSC-derived thymic epithelial cell product for the potential treatment of congenital thymic insufficiency**
Francisco Leon, Holger A. Russ, Jessie M. Barra, Alexander S. Baker, Antonio Jimeno, J Jason Morton, Nathaniel Alzofon, Philip Ball, Lara Silverman, Paul Dunford, Christian Blount, and Yeh-Chuin Poh

10:12 056 **Therapeutic restoration of immune function through iPSC-derived human thymic epithelial cells (iTECs) - An update on bringing iTECs to the clinic**
Wenqing Wang, Abdulvasey Mohammed, Martin Arreola, Hanh Dan Nguyen, Zihao Zheng, Yujin Moon, Kelsea M. Hubka, Humza Ali Khan, Mark Krampf, Agnieszka Czechowicz, Anca Pasca, Li Li, Virginia Winn, Sean Bendall, Rosa Bacchetta, Vittorio Sebastiano, and Katja G. Weinacht

10:21 **Q&A**

10:30 ***COFFEE BREAK AND POSTER VIEWING – COVERED TERRACE***

SESSION VII HEAD, SHOULDERS, KNEES AND TOES

Chairs: Jill Arganbright and Sulagna Saitta

11:10 057 **Postoperative hypocalcemia in children with 22q11.2 deletion syndrome: results following initiation of an institution-wide evidence-based clinical pathway**
Jill Arganbright, Meghan Tracy, Max Feldt, Nikita Raje, Janelle Noel-MacDonnell

11:20 058 **Peri-operative pathway for hypocalcemia management in youth with 22q11.2 deletion/duplication syndrome: preliminary outcomes following palate repair ***
Douglas Villalta, Donna M. McDonald-McGinn, T. Blaine Crowley, Audrey Olsen, Daniel E. McGinn, Elaine H. Zackai, Lisa Elden, Oksana Jackson, Edna E. Mancilla, Vaneeta Bamba, Ian Campbell, and Lorraine E. Levitt Katz

11:27 059 **Recurrent postpartum hypocalcemia in association with 22q11.2 deletion syndrome ***
Evie Andrewes, Audrey Olson, T. Blaine Crowley, Daniel E. McGinn, Oanh Tran, Beverly S. Emanuel, Elaine H. Zackai, Michele P. Lambert, Vaneeta Bamba, Lorraine L. Katz, Natalie Blagowidow, Lorraine Dugoff, and Donna M. McDonald-McGinn

11:31 060 **Thyroid carcinoma and 22q11.2 deletion syndrome – A critical association**
Andrew Bauer, T. Blaine Crowley, Audrey Olson, Lily Elman, Evie Andrewes, Daniel E. McGinn, Elaine H. Zackai, Beverly Emanuel, Michele P. Lambert¹, Kathleen E. Sullivan, Vaneeta Bamba, Lorraine L. Katz, Julio Ricarte-Filho, Jenny Li and Donna M. McDonald-McGinn

11:41 061 **Testosterone deficiency in males with 22q11.2 deletion syndrome: a case series**
Evana Valenzuela, Hanadys Ale, Lauren Waidner, Lauryn Bachman, and Jeremy Purrow

11:45 062 **Delineating the incidence, accrual, and pathways to development of cardiovascular disease for 22q11.2 microdeletion using population-based and clinical data ***
Sarah L. Malecki, Tracy Heung, Samantha Morais, Refik Saskin, Drew Wilton, Therese A. Stukel, Eyal Cohen, Amol A Verma, and Anne S. Bassett

- 11:55 063 Use of GLP-1 agonists and associated outcomes in adults with 22q11.2 deletion syndrome**
Samantha D'Arcy, Tracy Heung, Lisa Palmer, Nikolai Reyes, and Anne S. Bassett
- 12:02 064 Body composition and bone density in children with 22q11.2 deletion syndrome: evidence for reduced lean mass ***
Helena-Jamin Ly, Anna Svedlund, Annika Malmgren, and Solveig Oskarsdottir
- 12:09 065 Vertebral fractures in youth with 22q11.2 deletion syndrome: a case series ***
Amanda Spatz, Edna Mancilla, Donna M. McDonald-McGinn, Vaneeta Bamba, and Lorraine E. Levitt Katz
- 12:15 Q&A**
- 12:25 066 Cervical spinal cord compression in association with 22q11.2 deletion syndrome ***
Jette A. Boxem, Michiel L. Houben, Malcolm L. Ecker, Gregory Heuer, Erin S. Schwartz, Tom Schlosser, Gregory Heuer, Elaine H. Zackai, T. Blaine Crowley, Donna M. McDonald-McGinn, Aebele B. Mink van der Molen, and Alexander M. Tucker
- 12:32 067 Early vertebral rotation before the adolescent growth spurt in children with 22q11.2DS: Implications for development of scoliosis ***
Annemie J. Sluijter, Peter P.G. Lafranca, René M. Castelein, Moyo C. Kruyt, Keita Ito, Michiel L. Houben, and Tom P.C. Schlösser
- 12:36 068 The role of orthopedic conditions in lower extremity pain and exercise intolerance in children with 22q11.2 deletion syndrome**
Femke G.M. van den Helder and Michiel L. Houben
- 12:43 069 Incidental neuroradiological findings in 22q11DS: Update in an expanded sample**
J. Eric Schmitt, Simon Smerconish, David Roalf, T. Blaine Crowley, Victoria Giunta, Daniel E. McGinn, Elaine H. Zackai, Beverly S. Emanuel, Sarah Hopkins, Madeline Chadhumbe, Raquel E. Gur, Donna M. McDonald-McGinn, and Aaron Alexander-Bloch
- 12:53 070 Handgrip strength in adults with 22q11.2 deletion syndrome**
Erik Boot and Claudia Vingerhoets
- 13:03 071 The cutaneous spectrum of 22q11.2 deletion syndrome: a retrospective cohort analysis across age groups**
Hanadys Ale, Aditi Chokshi, Lucia Martinez, Victoria Rosali Gonzalez, Lucila Mesa, Victoria Griffith, Sarah Asbek, Camille Robinson, and Raul Escobar
- 13:13 072 22q through the back door ***
Lily N. Elman, Audrey Olsen, T. Blaine Crowley, Daniel E. McGinn, Evie Andrewes, Lydia Rockart, Lydia Hamilton, Oanh Tran, Jennifer Borowka, Natalie Ginn, Abraham Haimed, Michele P. Lambert, Ethan Goldberg, Beverly S. Emanuel, Elaine H. Zackai, and Donna M. McDonald-McGinn

13:20 Q&A

13:30 LUNCH – AMALTHEA RESTAURANT AND TERRACE
ALL CONFERENCE ATTENDEES AND ACCOMPANYING PERSONS

SESSION VIII EYES AND EARS AND MOUTH AND NOSE

Chairs: Oksana Jackson and Emily Gallagher

14:30 **073** *INVITED PRESENTATION*

Outcomes in patients with 22q11.2 deletion syndrome following speech surgery

Oksana A. Jackson, Philadelphia, PA, USA

14:45 **074** **Timing of surgery for velopharyngeal insufficiency in patients with 22q11.2 deletion syndrome: a retrospective chart analysis ***

Jette A Boxem, Cynthia B. Solot, Michiel L. Houben, T. Blaine Crowley, Donna M. McDonald-McGinn, Aebele B. Mink van der Molen, and Oksana A. Jackson

14:52 **075** **Bilateral buccal myomucosal flap palatoplasty for the management of velopharyngeal dysfunction in children with 22q11.2 deletion syndrome: case series of 4 patients**

Jill Arganbright, Meghan Tracy, Janelle Noel-MacDonnell, Kathryn Dent, Susan Parsons, and Nikita Raje

14:59 **076** **Presentations of congenital anterior glottic webs and laryngeal clefts in individuals with 22q11.2 deletion syndrome: patterns, outcomes, and clinical pathways**

Lisa Elden, Karen Zur, Ian Jacobs, Luv Javia, Steve Sobol, Donna McDonald-McGinn, T. Blaine Crowley, Elaine Zackai, and Ryan Ruiz

15:06 **077** **Altered Sleep Architecture and NREM Physiology in 22q11.2 Deletion Syndrome ***

Kathleen P. O'Hora, Rune Bøen, Hoki Fung, Charles Schleifer, Leila Kushan-Wells, Jennifer Xu, Natasha Reddivalam, Jared M. Saletin, and Carrie E. Bearden

15:13 **078** **Gastrointestinal symptom burden is associated with anxiety, stress, and sleep disturbance in children with 22q11.2 deletion syndrome**

Alain J. Benitez, Luc Santilli, Kush Patel, Audrey Olson, Lydia Rockart, Robin Miccio, Lauren White, Donna McDonald-McGinn, and Maria Mascarenhas

15:20 Q&A

15:30 **079** *INVITED PRESENTATION*

Speech and language in 22q11.2 deletion as a window into understanding long term outcome

Cynthia Solot, Philadelphia, PA, USA

- 15:45 **080** Predictors of persistent expressive language impairment in 22q11.2 deletion syndrome *
Gracie Trulove, T. Blaine Crowley, Daniel E. McGinn, Audrey Olson, Sarah Hopkins, Donna M. McDonald-McGinn, Cynthia Solot, and Samuel Alperin
- 15:52 **081** Language development in 22q11.2 deletion syndrome from preschool to school-age and in adolescence: evidence from two longitudinal studies
Tessel Boerma, Jantine Wignand, Manik Djelantik, Emma Everaert, Ania Fiksinski, Michiel Houben, Iris Selten, and Frank Wijnen
- 15:59 **082** Memory, attention and vocabulary in 22q11.2 deletion syndrome: a longitudinal study *
Jantine R. Wignand, Tessel D. Boerma, Emma Everaert, Iris S. Selten, Michiel L. Houben and Frank N.K. Wijnen
- 16:03 **083** Additional diagnoses in selected patients with autism and 22q11.2 deletion syndrome: casting an even wider net
Jennifer Rodstein, Caroline Y. Kuo, Apisadaporn Thambundit, Reina Capati and Sulagna C. Saitta
- 16:13 **084** Cognitive and emotional profiles in young adults with chromosome 22q11.2 deletion syndrome
Kathleen Angkustsiri, Andrea Schneider, Hannah Culpepper, Marwa Zafarullah, and Flora Tassone
- 16:20 Q&A
- 16:30 **AFTERNOON TEA AND POSTER SESSION – COVERED TERRACE
 ALL AUTHORS PRESENT**
- 18:00 Adjourn – Day 2
- 19:30 **22q OLYMPICS AND SUNSET SOCIAL – BAHIA MAR AND STONE THEATER
 FOLLOWED BY PIZZA AND BEVERAGES
 ALL CONFERENCE ATTENDEES AND ACCOMPANYING PERSONS
 PLEASE WEAR YOUR OLYMPIC VEST**



Thursday, July 16

6:00 **REGISTRATION OPEN – ZEUS BALLROOM COURTYARD**

7:00 **BREAKFAST – AMALTHEA RESTAURANT AND TERRACE**
MIRABELLO BAY RESORT GUESTS ONLY

SESSION IX COGNITION AND PSYCHIATRIC ILLNESS

Chairs: Ann Swillen and Ania Fiksinski

- 8:00 **085 30 years later: IQ profile among individuals with 22q11.2 deletion syndrome**
Edward M. Moss, Katherine Baum, Michael F. Woodin, T. Blaine Crowley, Elaine H. Zackai, and Donna M. McDonald-McGinn
- 8:10 **086 Neurocognitive profiles of individuals with 22q11.2DS: effects of deletion size, intellectual disability, and overall psychopathology burden**
Ruben C. Gur, Donna M. McDonald-McGinn, Kosha Ruparel, Carrie Bearden, Ann Swillen, Thérèse van Amelsvoort, Jonathan Sebat, Marianne van den Bree, Sébastien Jacquemont, Anne Bassett, Jacob Vorstman, and Raquel E. Gur
- 8:20 **087 Educational best practices for school-age children with 22q11.2 deletion syndrome: recommendations for educators**
Stephen R. Hooper, Edward M. Moss, Jane Summers, Katherine Baum, Ania M. Fiksinski, Maude Schneider, Cheryl Cytrynbaum, Donna M. McDonald-McGinn, Andrea Shugar, and Ann Swillen on behalf of the International Brain Behavior Consortium Study Investigators
- 8:30 **088 Interactive digital psychoeducation for 22q11.2 deletion syndrome: iterative, user-centred development of an app to support understanding of genetic complexity**
Maxwell Szymanski, Robin De Croon, Ann Swillen, Marie Meuris, and Katrien Verbert
- 8:40 **089 Psychoeducation across the lifespan in 22q11.2 deletion syndrome: enhancing communication and tailored support in home, school, and work contexts**
Ann Swillen, Marie Meuris, Robin De Croon, Maxwell Szymanski, and Katrien Verbert
- 8:47 **090 Development and evaluation of an evidence-based group-based psychoeducation program for families affected by 22q11.2 deletion syndrome**
Ania Fiksinski, Lieke Reijn, and Michiel Houben
- 8:54 **091 Rare mutations implicate CGE interneurons as a vulnerable axis of cognitive deficits across psychiatric disorders**
Stephanie A. Herrlinger, Jiayao Wang, Bovey Y Rao, Jonathan Chang, Joseph A. Gogos, Attila Losonczy and Dennis Vitkup

- 9:05 Q&A
- 9:15 **092** **INVITED PRESENTATION**
22q11.2 deletion as a window into understanding the genomics of psychiatric illness
Anne Bassett, Toronto, ON, Canada
- 9:30 **093** **Medical burden, psychopathology and neurocognition in 22q11.2 deletion syndrome**
Raquel E. Gur, Donna M. McDonald-McGinn, Ruben C. Gur, Arielle Ered, Austin T. Moran, Claudine B. Van Arman, Alyssa M. Neppach, Mia Schwartzberg, R Sean Gallagher, Simon Smerconish, Kosha Ruparel, Scott Troyan, Tyler M. Moore, Amy Cassidy, Lauren K. White, T. Blaine Crowley, Daniel E. McGinn, Audrey Olson, Oanh Tran, Elaine H. Zackai, Beverly S. Emanuel, Yana Poltavets, Ronnie Weinberger, Michal Taler, and Doron Gothelf
- 9:40 **094** **Developmental trajectories of irritability and associations with a psychosis-prone state in youth with 22q11.2 deletion syndrome and other neurodevelopmental copy number variants**
Jessica H. Hall, Josephine E. Haddon, Samuel J. R. A. Chawner, Jeremy Hall, Peter Holmans, Michael J. Owen and Marianne B. M. van den Bree
- 9:50 **095** **Exposure to trauma and post-traumatic stress disorder in 22q11.2 deletion syndrome**
Adi Drapisz, Yana Poltavets, Ronnie Weinberg, Naama De La Fonataine, Nitza Nakash-Axelrod, Michal Taler, and Doron Gothelf
- 10:00 **096** **Copy Number Variants in Spanish Psychiatric Care: Clinical Characteristics and Findings a Multicenter Cohort**
Damià Heine-Suñer, Maria Victoria Lull Albertí, Javier Labad, Jesús Herrera-Imbroda, Vicent Balanzá-Martínez, Amaya Hervas, Anna Maria Cueto-González, Antoni Ramos-Quiroga, Rosa Ayesa-Arriola, Iñaki Zorrilla, Ana García-Blanco, Paz García-Portilla, Edith Pomarol-Clotet, Bàrbara Arias, Covadonga Martínez, Olga Rivero, the CESPED group, and Elisabet Vilella
- 10:10 **097** **Association between sluggish cognitive tempo symptoms and psychiatric diagnosis in a cohort of children with 22q11.2 deletion syndrome**
Veselina Gadancheva, and Fiona McNicholas
- 10:16 Q&A
- 10:30 **COFFEE BREAK – COVERED TERRACE**
- SESSION X NEUROANATOMY, METABOLISM, AND CNS FUNCTION**
Chairs: Jacob Vorstman and Stanislav Zakharenko
- 11:10 **098** **INVITED PRESENTATION**
Emerging Discoveries on 22q11.2 and the Blood–Brain Barrier
Francesco Papaleo, Genova, Italy

- 11:25 **099** **Biochemical alterations in tryptophan metabolism in 22q11.2 deletion syndrome**
Cassandra J. Hatzipantelis, David E. Olson, Kathleen Angkustsiri, Andrea Schneider, and Flora Tassone
- 11:32 **100** **Systemic energy metabolism disruption as a biomarker of psychosis risk in 22q11.2 deletion syndrome**
Marwa Zafarullah, Hannah Culpepper, Blythe Durbin-Johnson, Andrea Schneider, Kathleen Angkustsiri, and Flora Tassone
- 11:40 **101** **The making and un-making of association cortico-cortical connections in 22q11DS**
Anthony -S. LaMantia, Thomas M. Maynard, Zachary D. Erwin, and Shah Rukh
- 11:50 **Q&A**
- 12:00 **102** ***INVITED PRESENTATION***
Neural circuit dysfunctions in animal and human models of schizophrenia risks
Stanislav S. Zakharenko, Memphis, TN, USA
- 12:15 **103** **Cerebellar regulation of top-down auditory cortical circuits and its potential role in 22q11.2 deletion syndrome**
Tae-Yeon Eom, Christopher M. Davenport, Jay Blundon, Feng Feng, Troy A. Hackett, and Stanislav S. Zakharenko
- 12:25 **104** **Associations between psychotic symptoms and subthalamic nuclei volumes in the 22.11.2 deletion syndrome (22q11DS) using Ultra High Field (UHF) MRI**
J. Eric Schmitt, Simon Smerconish, David Roalf, Sarah Hopkins, Aaron Alexander-Bloch, T. Blaine Crowley, R. Sean Gallagher, Daniel E. McGinn, Kosha Ruparel, Lauren K. White, Elaine H. Zackai, Anne Bassett, Ruben C. Gur, Raquel. E. Gur, Stanislav Zakharenko, and Donna M. McDonald-McGinn
- 12:34 **105** **Cerebellar differences in 22q11.2 duplication syndrome ***
Samuel Alperin, Madeline Chadehumbe, T. Blaine Crowley, Donna M. McDonald-McGinn, and Sarah Hopkins
- 12:40 **Q&A**
- 12:50 **106** **Neurodevelopmental perturbations in 22q11.2DS using mouse and human functional genomic approaches ***
Wenna Chen, Lijie Shi, Elisha J. Fogel, Bernice E. Morrow, and Deyou Zheng

- 13:00 **107** **Disorganized inhibitory dynamics in hippocampal area CA1 of 22q11.2 deletion mutant mice**
Stephanie A. Herrlinger, Bovey Y Rao, Margaret E. Conde Paredes, Anna L. Tuttman, Haroon Arain, Erdem Varol, Joseph A. Gogos, and Attila Losonczy
- 13:10 **108** **Olfactory deficits and structural anomalies of the olfactory system in 22q11.2 deletion syndrome**
David R. Roalf, Donna M. McDonald-McGinn, Bruce I. Turetsky, Paul J. Moberg, T. Blaine Crowley, Phoebe Freedman, Monica E. Calkins, R. Sean Gallagher, Audrey Olson, Daniel E. McGinn, Lydia Rockart, Oanh Tran, Elaine H. Zackai, Beverly S. Emanuel, Kosha Ruparel, Russell T. Shinohara, Lauren White, and Raquel E. Gur

13:20 **Q&A**

13:30 **LUNCH – AMALTHEA RESTAURANT AND TERRACE**
ALL CONFERENCE ATTENDEES AND ACCOMPANYING PERSONS

SESSION XI BIOMARKERS AND MODIFIERS

Chairs: Carrie Bearden and Erik Boot

- 14:30 **109** **INVITED PRESENTATION**
Impact of the 22q11.2 Deletion on Brain Structure and Function: Insights from the ENIGMA Consortium
Carrie E. Bearden, Los Angeles, CA, USA
- 14:45 **110** **Network-specific deviations in functional brain development in 22q11.2DS linked to psychosis**
Kaustubh Supekar, Leila Kushan, Charles Schleifer, Gabriela Repetto, Nicolas A Crossley, and Carrie Bearden
- 14:55 **111** **Striatal dopamine transporter binding in adults with 22q11.2 copy number variants: An [123I]FP-CIT SPECT study ***
Jeltje C.C. Spapens, Carmen F. M. van Hooijdonk, Rik Schalbroeck, Amy L. Sylvester, Erik Boot, Therese A.M.J. van Amelsvoort, Claudia Vingerhoets, and Jan Booij
- 15:05 **112** **Cortical myoclonus as a distinct motor phenotype of 22q11.2 deletion syndrome**
Nikolai Gil D. Reyes, Talyta Grippe, Benedetta Angeloni, Victor Lira, Connie Marras, Erik Boot, Ryan K.C. Yuen, Anthony E. Lang, Danielle M. Andrade, Robert Chen, and Anne S. Bassett
- 15:15 **113** **Early-onset Parkinson's disease with 22q11.2 microdeletion and pathogenic GBA1 variant**
Nikolai Gil D. Reyes, Daniel G. Di Luca, Ivan Martinez-Valbuena, Erik Boot, Maria Corral, Ryan K.C. Yuen, Connie Marras, Anthony E. Lang, and Anne S. Bassett

- 15:23 **114** **Assessing a polygenic risk score for Parkinson's disease in 22q11.2 deletion syndrome**
Nikolai Gil D. Reyes, Shengjie Ying, Adrian I. Espiritu, Tracy Heung, Yue Yin, Erik Boot, Anthony E. Lang, Connie Marras, Ryan K.C. Yuen, and Anne S. Bassett
- 15:30 **115** **Biomarkers in tear fluid: examining dopamine and p-tau in participants with 22q11.2 deletion syndrome**
Thérèse van Amelsvoort, Emma von Scheibler, Rianne van Roij, Erik Boot, Claudia Vingerhoets, David Linden, and Marlies Gijs
- 15:40 **116** **Dopamine-associated oxidative stress differentially compromises cortical and striatal neurons and circuits in the *LgDel* 22q11DS mouse model**
Anthony-S. LaMantia, Zachary D. Erwin, Thomas M. Maynard, Seth R. Batten, Terrence M. Lohrenz, Leonardo Barbosa, Paul Sands III, and P. Read Montague
- 15:50 **Q&A**
- 16:00 **AFTERNOON TEA – COVERED TERRACE**

SESSION XII MODELING TOWARDS DISCOVERY

Chairs: Spyros Petrakis and Thérèse van Amelsvoort

- 16:30 **117** **INVITED PRESENTATION**
Stem cell-based drug discovery for neurological disorders
Spyros Petrakis, Thessaloniki, Greece
- 16:45 **118** **Aberrant glutamatergic transmission and homeostatic plasticity in a human neuronal model of 22q11.2 deletion syndrome**
Weibo Niu, Jidong Guo, Ximing Ran, David Parker, Xiangru Li, Shaojun Yu, Sidney Imes, Ryan Purcell, Eric Guo, Sophia Y. Jiang, Gary J Bassell, Jennifer G Mulle, Opal Y. Ousley, Bradley Pearce, Yue Feng, Elaine F. Walker, Joseph F. Cubells, Zhaohui Qin, Erica Duncan, and Zhexing Wen
- 16:55 **119** **Altered neurophysiological activity of iPSC-derived neurons of 22q11.2 deletion syndrome**
Adrian J Harwood, Jeremy Hall, and Gemma Wilkinson
- 17:02 **120** **Mapping genes differentially expressed in 22q deletion syndrome to sub-populations of control cells defined using scRNA-seq at developmental timepoints during neuro-differentiation**
Janet C. Harwood, Karolina Dec, Jamie Wood, David Linden, Thérèse van Amelsvoort, and Adrian J. Harwood

- 17:09 **121** **Transcriptomic signature of iPSC-derived neural progenitor cells from 22q11.2 microduplication carriers**
Natasa Kovacevic-Grujicic, Jovana Kostic, Mina Peric, Luka Bojic, Ivana Hadzi Makuncevic, Goran Cuturilo, Adrian J. Harwood, Danijela Drakulic, Milena Stevanovic
- 17:16 **122** **Neural lineage-specific transcriptomic signatures of intrafamilial clinical variability in a Mother-child pair with 22q11.2 microdeletion**
Danijela Drakulic, Ivana Hadzi Makuncevic, Natasa Kovacevic-Grujicic, Mina Peric, Friederike Ehrhart, Jovana Kostic, Stefan Lazic, Goran Cuturilo, Spyros Petrakis, David E. J. Linden, Adrian J. Harwood, and Milena Stevanovic
- 17:23 **Q&A**
- 17:30 **Adjourn – Day 3**
- 20:00 ***DINNER AND DANCING – ELIA TERRACE***
TOP SESSION PRESENTATION AWARDS
ALL CONFERENCE ATTENDEES AND ACCOMPANYING PERSONS
PLEASE WEAR YOUR DANCING SHOES



Friday, July 17

7:00 **BREAKFAST – AMALTHEA RESTAURANT AND TERRACE**
MIRABELLO BAY RESORT GUESTS ONLY

8:00 **REGISTRATION OPEN – ZEUS BALLROOM COURTYARD**

SESSION XIII TREATING WHAT IS TREATABLE

Chairs: Doron Gothelf and Raquel Gur

8:30 **123 INVITED PRESENTATION**

The effectiveness and safety of psychopharmacotherapy in 22q11.2 deletion syndrome

Doron Gothelf, Tel Aviv, Israel

8:45 **124 Targeting prefrontal chloride transport for cognitive remediation in 22q11.2 deletion syndrome and substance use disorders**

Arghya Mukherjee

8:55 **125 Effects of riluzole on psychiatric symptoms, excitation-inhibition balance and functional connectivity in 22q11.2 deletion syndrome**

Claudia Vingerhoets, Amy Sylvester, Desmond Tse, Jeltje Spapens, Sophie Kappert, Nele Soons, David Linden, Dimo Ivanov, and Therese van Amelsvoort

9:05 **126 Ten-year retrospective review of psychotropic medication use in individuals with 22q11.2 deletion syndrome ***

Samuel Alperin, Sarah Hopkins, Madeline Chadehumbe, T. Blaine Crowley, Daniel E. McGinn, Audrey Olson, Oanh Tran, R. Sean Gallagher, Kosha Ruparel, Ruben Gur, Elaine H. Zackai, Beverly S. Emanuel, Raquel Gur, and Donna M. McDonald-McGinn

9:12 **127 Exploring the relationship between ADHD medication and psychotic symptoms in 22q11.2 deletion syndrome**

Claudine B. Van Arman, Arielle Ered, Tyler M. Moore, Austin T. Moran, Alyssa M. Neppach, Mia Schwartzberg, R Sean Gallagher, Simon Smerconish, Kosha Ruparel, Scott Troyan, Amy Cassidy, Lauren K. White, T. Blaine Crowley, Daniel E. McGinn, Audrey Olson, Oanh Tran, Elaine H. Zackai, Beverly S. Emanuel, Ruben C. Gur, Donna M. McDonald-McGinn, and Raquel E. Gur

9:20 **128 Patients with 22q11.2 deletion syndrome - Too challenging to treat?**

Anne S. Bassett

9:30 **Q&A**

- 9:40 **129** Resolution of dysautonomic "seizure-like" episodes following antipsychotic withdrawal in an adult with 22q11.2 deletion syndrome and prior scoliosis surgery
Madeline A Chadehumbe, Donna M. McDonald-McGinn, and Raquel E Gur
- 9:47 **130** Psychiatric management concerns in the transition from pediatrics to adult 22q providers
Kerry D. Conant, Ashmita Banerjee, Matt Blessing, Gary Stobbe, and Emily Gallagher
- 9:54 **131** Feasibility and acceptability of BE-WEHL-22q: a virtual wellness program for children with 22q11.2 deletion syndrome and their families
Luc Santilli, Alain J. Benitez, Audrey Olson, Lydia Rockart, Robin Miccio, Lauren White, Donna McDonald-McGinn, and Maria Mascarenhas
- 10:01 **132** **INVITED PRESENTATION**
Current findings and future directions from the IBBC and G2MH Networks
Raquel E. Gur, Philadelphia, PA, USA
- 10:15 **Q&A**
- 10:30 **COFFEE BREAK – COVERED TERRACE**

SESSION XIV MULTIDISCIPLINARY CARE

Chairs: Anne Bassett and Donna McDonald-McGinn

- 11:10 **133** **INVITED PRESENTATION**
Seeing the whole elephant: AI-powered discovery across a referral center's 22q experience
Ian Campbell, Philadelphia, PA, USA
- 11:25 **134** An exploratory study using machine learning to detect clinical patterns for 22q11.2 deletion syndrome *
Lais A. Soares, Guilherme P. Coelho, João Pedro Moretti, Vera L. Gil-da-Silva-Lopes, and Ana E. A. Silva
- 11:35 **135** Multisystem medical burden in 22q11.2 deletion syndrome – managing complex care
T. Blaine Crowley, Evie Andrewes, Audrey Olsen, Daniel E. McGinn, Lily Elman, Lydia Rockart, Lydia Hamilton, Elaine H. Zackai, and Donna M. McDonald-McGinn
- 11:45 **Q&A**

- 11:55 **136** **Stakeholder survey in 240 members of and other relevant parties related to the Dutch 22q11 foundation**
Kim van Bekkum, Erik Boot, Sasja Duijff, Ania Fiksinski, Michiel Houben, Petra Oostema, and Claudia Vingerhoets
- 12:05 **137** **Caregiver burden and support needs in caregivers of adults with 22q11.2 deletion syndrome**
Lisa Palmer, Tracy Heung, Nikolai Reyes, Samantha D'Arcy, and Anne S. Bassett
- 12:15 **138** **Optimizing 22q care: the impact of a single-day multidisciplinary clinic model**
Jessica Goldberg, Valerie J Allegretti, Kelly Regan-Fendt, Jalen Clemmons, Sylvie Render, Mitch Bastillo, Elizabeth Lippner, Jennifer Miller, Naiomi Gunaratne, Margaret Ryan, Edward Jones, Chanel Howard, Ryan Cyzman, and Laura Swibel Rosenthal
- 12:22 **139** **Improving efficiency in the 22q deletion clinic ***
Eme Ani, Felmy Ajayi, Hui Yann, Hui Yann Yap, Chayanuch O. Charoenrat, and Alice Roueche
- 12:29 **140** **Geographic impacts of healthcare utilization for patients with 22q11.2 deletion/duplication in the pacific northwest**
Emily R. Gallagher, Jade Singleton, and Kristy Carlin
- 12:36 **141** **Shorter distance to main academic center associated with better longitudinal care and follow up in adult patients with 22q11.2 deletion syndrome ***
Jasmyn Atalla, Blaine Crowley, Kathleen Sullivan, Jennifer Heimall, Donna M. McDonald-McGinn, and Timothy Buckley
- 12:43 **142** **Where do we go from here? Strategies to support successful transition to adult interdisciplinary care from a pediatric 22q center**
Matthew Blessing, Kerry Conant, Gary Stobbe, and Emily Gallagher
- 12:50 **Q&A**

SESSION XV LAST WORDS

- 13:00 **143** **INVITED PRESENTATION**
Family Voices
- 13:15 **144** **INVITED PRESENTATION**
22q11.2DS - towards restoration of the lost folio
Peter Scambler, London, UK
- 13:35 **Q&A**

13:40 JUNIOR INVESTIGATOR AND TOP FRIDAY PRESENTATION AWARDS

Peter Scambler, 22q11.2 Society Trustee and Vice-Treasurer

13:50 CLOSING REMARKS AND ANNOUNCEMENT OF 2028 MEETING LOCATION

Donna M. McDonald-McGinn, 22q11.2 Society Chair

13:55 Closing Breath

Maria Mascarenhas, Philadelphia, PA, USA

14:00 Meeting Adjourned – See you in 2028!

FAREWELL FARE – COVERED TERRACE

ALL CONFERENCE ATTENDEES AND ACCOMPANYING PERSONS



POSTER PRESENTATIONS:

Top Scoring Posters

- 145** Implementation of digital 22q care plan in electronic medical record to improve interdisciplinary coordination and visibility
Lindsey H. Benedict, Hannah Berntson, Matthew Blessing, Laura Cline, Leslie Cronk, Emily Gallagher, and Suzanne Siegel
- 146** Engaging children with 22q11.2-related disorders in their interdisciplinary care
Hannah Berntson, Skyler Leonard, Maria Mills, Lex Powers, Cindy Trevino, and Matthew Blessing
- 147** Nonword repetition skills of both preschoolers with 22q11.2 deletion syndrome and peers with developmental language disorder are weak, but differently associated with vocabulary
Tessel Boerma, Marieke Huls, Emma Everaert, Iris Selten, Ellen Gerrits, and Frank Wijnen
- 148** Oxycephaly associated with 22q11.2 deletion syndrome: a first reported case *
Jette A. Boxem, Aebele B. Mink van der Molen, Elaine H. Zackai, T. Blaine Crowley, Donna M. McDonald-McGinn, and Scott P. Bartlett
- 149** Sleep disruption in a mouse model of 22q11.2 deletion syndrome * #
Lara M. Boyle, Srdjan Jaksimovic, Christopher M. Cielo, Donna M. McDonald-McGinn, Raquel E. Gur, Ashlea Choi, and Shinjae Chung
- 150** Importance of timely screening for mental health concerns within a multidisciplinary 22q clinic setting and the barriers surrounding identification
Jalen D. Clemmons and Carter A. Puckett
- 151** Altered germ layer specification during gastrulation in 22q11.2 deletion syndrome * #
Aidan Douglas, Eleonora Stronati, Elizabeth Abraham, Emily Megill, Olivia Mae Pericak, and Conchi Estarás
- 152** Establishing novel partnerships to create a comprehensive program for adults with 22q
Emily R. Gallagher, Matt Blessing, Kerry Conant, and Gary Stobbe
- 153** Impact of chromosome 22q11.2 CNVs on height in two unrelated patients with achondroplasia *
Audrey Olson, T. Blaine Crowley, Lydia Hamilton, Evie Andrewes, Lily Elman, Lydia Rockart, Oanh Tran, Daniel E. McGinn, Beverly S. Emanuel, Elaine H. Zackai, Vaneeta Bamba, Lorraine L. Katz, and Donna M. McDonald-McGinn
- 154** Enhanced serological protection after pneumococcal polysaccharide vaccine in a child with 22q11.2 deletion syndrome and recurrent sinopulmonary infection *
Annika Malmgren, Jenny Lingman Framme, and Solveig Oskarsdottir
- 155** Cardiopulmonary findings in teenagers with right circumflex aortic arch in 22q11 deletion syndrome
Julie Martin, Manbir Sanghera, Matthew M. Feldt, Nikita Rajee, Maria Kiaffas, and Sanket Shah

- 156** Very early onset inflammatory bowel disease in children with 22q11.2 deletion syndrome
Maire Conrad, Judith Kelsen, Kathleen Sullivan, Soma Jyonouchi, Audrey Olson, Donna M. McDonald-McGinn, and Maria Mascarenhas
- 157** Studying the interplay between vitamin A and hippo signaling pathways in human models of 22q11.2 deletion syndrome * #
Emily Megill, Eleonora Stronati, Elizabeth Abraham, Aidan Douglas, Mikel Zubillaga, and Conchi Estaras
- 158** Development of a screening protocol for *TANGO2* deficiency in pediatric patients with 22q11.2 deletion syndrome: initial findings and insights
Maria Mills, Lex Powers, Matthew Blessing, Kimberly Liekweg, Lindsey Benedict, Suzanne Siegel, and Emily Gallagher
- 159** Diagnosing and managing complex neuropsychiatric challenges in 22q11.2 deletion syndrome
Lisa Palmer, Tracy Heung, Nikolai Reyes, Samantha D'Arcy, and Anne S. Bassett
- 160** Binary predictive modeling for 22q11.2 deletion using structured clinical data: an experimental ensemble study with linear SVM and complement naive bayes
Giulia T. Barcellos, Vera L. Gil-da-Silva-Lopes, João Pedro Moretti, Ana E. A. Silva, and Guilherme P. Coelho
- 161** Immunodeficiency presentation in a patient with a distal 22q11.23 (LCR F-H) duplication *
Kelly E. Regan-Fendt, Valerie J. Allegretti and Elizabeth A. Lippner
- 162** Parent perspectives on educational practices for students with 22q11.2 deletion syndrome in Ontario, Canada
Jane Summers, Katrina Palad, Sana Emami, and Jacob Vorstman
- 163** Trauma-related symptoms in adults with 22q11.2 deletion syndrome: exploring the efficacy of eye movement desensitization and reprocessing (EMDR) therapy
Claudia Vingerhoets, Sasja Duijff, Sanne Vrinzen, Jeltje Spapens, Erik Boot, and Therese van Amelsvoort
- 164** Words in context: measuring the vocabulary network in children with 22q11.2 deletion syndrome *
Jantine R. Wignand, Naomi Oppeneer, Tessel D. Boerma, Michiel L. Houben and Frank N.K. Wijnen

Top Scoring Posters