Tuesday ∗ July 16, 2024

6:00 ***REGISTRATION OPEN – BALLROOM I: PENICHE AND SINTRA FOYER***

7:00 ***BREAKFAST – TEMPERA RESTAURANT AND OUTDOOR TERRACE***

OPENING CEREMONY – BALLROOM I: PENICHE AND SINTRA

8:00 **The 22q11.2 Society Welcomes a Parade of Nations on Our Silver Anniversary**
Donna M. McDonald-McGinn, 22q11.2 Society Chair

Bem Vindo a Portugal
Marta Sousa Santos

Program Synopsis and Acknowledgements
Beata A. Nowakowska, 22q11.2 Society Trustee and 2024 Program Chair

Presentation of the Angelo DiGeorge Memorial Medal of Honor
Ann Swillen, 22q11.2 Society Secretary

Presentation of the Clodagh Murphy Memorial Unsung Hero Award
Erik Boot, 22q11.2 Society Trustee

Opening Breath
Maria Mascarenhas

SESSION 1: GENOTYPES, PHENOTYPES, AND MECHANISMS

8:30 **001** ***Invited Presentation and Lifetime Achievement Award***
Presented by Bernice Morrow, 22q11.2 Society Trustee
Back to the Future – A Brief History of the 22q11.2 Deletion Syndrome
Beverly S. Emanuel, Philadelphia, PA, USA

8:50 **002** ***Peter Scambler Invited Lecture and Award***
Presented by Anne Bassett, 22q11.2 Society Treasurer
Genetics of CNVs Informing Human Disorders and the Prospects for Treatment
Stephen W. Scherer, Toronto, CA

9:15 **Q&A**

9:20 **003** **Size Matters: Nested Chromosome 22q11.2 Deletions**

Donna M. McDonald-McGinn, Victoria Giunta, Bekah Wang, Daniel E. McGinn, Audrey Green, Lydia Rockart, Oanh Tran, Ryan LaPointe, Sam Alperin, Vaneeta Bamba, Katherine Baum, Madeline Chadehumbe, Christopher Cielo, Malcolm Ecker, Lisa Elden, John Flynn, Brian Forbes, R. Sean Gallagher, Elizabeth Goldmuntz, Raquel E. Gur, Steven Handler, Sarah Hopkins, Oksana Jackson, Lorraine Katz, Thomas Kolon, Michele Lambert, Asim Maqbool, Maria Mascarenhas, Edward Moss, Hyun-Duck Nah, Michael Nance, Michelle Scott, Cynthia Solot, Kathleen E. Sullivan, Ian Campbell, Elaine H. Zackai, Beverly S. Emanuel, and T. Blaine Crowley

9:27 **004** **22q11.2 Deletion and Duplication Syndromes – Analysis and Refinement of Breakpoints by Array-CGH**

Joana B. Melo, Luís M. Pires, Mariana Val, Susana I. Ferreira, and Isabel M. Carreira

9:31 **005** **The Tiniest Piece Leading to a Big Picture: Nested Chromosome 22q11.2 LCR22C-LCR22D Deletions** *

Daniel E. McGinn, Victoria Giunta, Bekah Wang, T. Blaine Crowley, Audrey Green, Oanh Tran, Renee DiCicco Wright, Julie Moldenhauer, Elaine H. Zackai, Beverly S. Emanuel, and Donna M. McDonald-McGinn

9:38 **006** **22q11.2 Deletion Syndrome Parent of Origin by Sex and Race** *

Ryan Lapointe, Oanh Tran, Steven Pastor, Victoria Giunta, T. Blaine Crowley, Audrey Green, Lydia Rockart, Bekah Wang, Daniel E. McGinn, Elaine H. Zackai, Donna M. McDonald-McGinn, and Beverly S. Emanuel

9:42 **007** **Optical Mapping Reveals Significant Differences in 22q11.2 Genomic Structures Between African American and White Populations**

Steven Pastor, Oanh Tran, Ryan Lapointe, Victoria Giunta, Daniel E. McGinn, Bekah Wang, Audrey Green, T. Blaine Crowley, Elaine H. Zackai, Donna M. McDonald-McGinn, and Beverly S. Emanuel

9:52 **008** **Proximal Nested 22q11.2 Deletions and Ancestry**

Anne S. Bassett, Tracy Heung, Erik Boot, María Ángeles Mori, María de los Ángeles Gómez Cano, Cristina Digilio, Damien Heine-Suñer, Bruno Marino, Julián Nevado Blanco, Beata Nowakowska, Federica Pulvirenti, Carolina Puttoto, Emi Rikeros, T. Blaine Crowley, Lydia Rockart, Victoria Giunta, Bekah Wang, Beverly Emanuel, the International 22q11.2 Brain and Behavior Consortium, Bernice Morrow, and Donna M. McDonald-McGinn

9:56 **009** **22q11.2DS Embryonic Stem Cell Lines with and without the Flanking Low Copy Repeats***

Marta Sousa Santos and Joris Vermeesch

10:00 **010** **Dissecting the Clinical Complexity of 22q11 Deletion Syndrome by Deep Phenotyping and Functional Genomics**
Maciej Geremek, Victoria Giunta, T. Blaine Crowley, Daniel E. McGinn, Audrey Green, Bekah Wang, Oanh Tran, Ryan Lapointe, Beverly S. Emanuel, Elaine H. Zackai, Donna M. McDonald-McGinn, and Beata A. Nowakowska

10:10 **011** **Two Circuits, Thirty-Two Genes, One Copy: 22q11.2 Deletion Syndrome is a Polygenic Disorder of Neural Circuit Development**
Anthony S. LaMantia

10:20 **Q&A**

10:30 ***COFFEE BREAK AND POSTER VIEWING ON THE COVERED TERRACE***
Odd Authors Present

SESSION 2: INCIDENCE, PREVALENCE, DETECTION, AND MODIFIERS

11:00 **012** **Cell free DNA and the Promise of Novel Biomarkers**
Joris Robert Vermeesch, Leuven, Belgium

11:15 **013** **Performance of SNP-based Cell-Free DNA Prenatal Screening for 22q11.2 Deletion Syndrome in a Commercial Population**
Wendy DiNonno, Melissa K. Maisenbacher, Georgina Goldring, Melda Balcioglu, Kayla Turner, M. Caleb Meads, Kayla Ruiz, Priyanka Arya, Jeffrey Meltzer, Katherine Howard, Sheetal Parmar

11:25 **014** **Single cell Sequencing of Circulating Extravillous Trophoblasts for Non-Invasive Fetal Copy Number Variant Screening**
Francesca Romana Grati, Tamara Stampalija, Emma Bertucci, Claudia Izzi, Paolo Volpe, Isabella Fabietti, Antonio Novelli, Lucia Pasquini, Sara Ornaghi, Elisa Bevilacqua, Dario Paladini, Tullio Ghi, Debora Lattuada, Paolo Gasparin, Fabio Facchinetti, Chiara Dordoni, Valentina De Robertis, Elena Nicastri, Valentina Parisi, Marco Bonito, Grazia Di Gregorio, Maria Verderio, Francesco Danilo Tiziano, Daniela Orteschi, Antonio Brocco, Genny Buson, Arianna Casadei, Chiara Mangano, Chiara Maranta, Martina Dori, Lorenzo Monasta, Chiara Bolognesi, Claudio Forcato, Anna Doffini, Thomas J Musci, Enrico Ferrazzi

11:35 **015** **Identification of 22q11.2 Quadruplication in Mother and Son Through Prenatal Cell-Free DNA Screening**
Natalie Blagowidow, Amy Kimball, Antonie D. Kline

11:42 **016** **Investigating the Incidence of the 22q11.2 Deletion Syndrome in Miscarriages**
Melissa K. Maisenbacher, Katrina Merrion, Jeffrey Meltzer, Katherine L. Howard, Samantha Leonard

11:49 **017** **Prevalence of 22q11.2 Deletion Syndrome in Offspring Conceived Via Assisted Reproductive Technology Versus Spontaneously**

Jennifer Borowka, T. Blaine Crowley, Victoria Giunta, Daniel E. McGinn, Bekah Wang, Audrey Green, Lydia Rockart, Oanh Tran, Beverly S. Emanuel, Elaine H. Zackai, Lorraine Dugoff, Kathleen Valverde, Donna M. McDonald-McGinn

11:55 **Q&A**

12:10 **018** **22q and Two Squared** *

Victoria Giunta, T. Blaine Crowley, Daniel E. McGinn, Bekah Wang, Audrey Green, Lydia Rockart, Oanh Tran, Beverly S. Emanuel, Elaine H. Zackai, Victoria Vetter, Sarah Hopkins, Donna M. McDonald-McGinn

12:17 **019** **Unusual Cases of 22q11.2 Disorders: Phenotypes and Lessons**

Marina S. Clarke, Caroline Y. Kuo, Apisadaporn Thambundit, Gregory Perens, Chloe Rome, Sulagna C. Saitta

12:24 **020** **Hereditary Paraganglioma-Phaeochromocytoma Syndrome in a Patient With 22q11.2 Deletion Syndrome** *

Kate Rigney, TS Paran, J Pears, SM O'Connell, A James, L Bradley, W Mulcahy, S Kelleher

12:28 **021** **Influence of Polygenic Risk on Height In Individuals with a 22q11.2 Deletion** *

Shengjie Ying, Tracy Heung, Bernice Morrow, Bhooma Thiruvahindrapuram, Ryan K. C. Yuen, Anne S. Bassett

12:34 **022** **Influence of Polygenic Risk on BMI in Individuals with a 22q11.2 Deletion** *

Shengjie Ying, Tracy Heung, Bernice Morrow, Bhooma Thiruvahindrapuram, Ryan K. C. Yuen, Anne S. Bassett

12:41 **023** **Mortality and Age at Molecular Diagnosis in Adults with 22q11.2 Deletion Syndrome** *

Christina Blagojevic, Tracy Heung, Sarah L. Malecki, Sabrina Cancelliere, Maria Corral, Anne S. Bassett

12:51 **024** **Clinical Suspicion and Diagnostic Delay in a Cohort of Adults With 22q11.2 Deletion Syndrome: Are We Doing Enough?** *

Federica Pulvirenti, Eleonora Sculco, Patricia Quijada-Morales, Daniele Guadagnolo, Maddalena Sciannamea, Maria Elena Santaniello, Giulia Di Napoli, Bruno Marino, Carolina Putotto, Isabella Quinti

13:01 **025** **Rare Copy-Number Variants as Modulators of Clinical Phenotypes of Adults with 22q11.2 Deletion Syndrome** *

Federica Pulvirenti, Daniele Guadagnolo, Maddalena Sciannamea, Eleonora Sculco, Bruno Marino, Flaminia Pugnaloni, Isabella Quinti, Laura Bernardini, Carolina Putotto

13:10 **Q&A**

13:30 ***LUNCH – TEMPERA RESTAURANT AND OUTDOOR TERRACE***

SESSION 3: HEART, IMMUNE, AND BLOOD

14:30 **026** ***Invited Presentation*** - **Cardiac Organoids Provide a Platform for Complex Genetic Studies of 22q11 Deletion Syndrome**
Casey Gifford, Palo Alto, CA, USA

14:45 **027** ***Tbx1*** **Promotes Maturation and Restricts Atypical Fates in Multilineage Progenitor Cells Needed to Form the Cardiac Outflow Tract**
Bernice E. Morrow, Kevyn Jackson, Alexander Ferrena, Deyou Zheng

14:55 **028** **Cell Compensation Associated with Heart Defects in a Mouse Model of 22q11.2 Deletion Syndrome**
Bingruo Wu, Punit Bhattachan, Deyou Zheng, Bernice Morrow, Bin Zhou

15:05 **029** **Spectrum of Congenital Heart Disease in a National Cohort of Patients with 22q11.2 Deletion Syndrome**
Ciara Ryan, Wesley Mulcahy, Suzanne Kelleher, Colin J. McMahon

15:09 **030** **The "Hidden" Cardiac Anomalies in Patients Presenting Without Major Congenital Heart Disease in 22q11.2 Deletion Syndrome**
Elizabeth Goldmuntz, Michelle Litvak, Brande Latney, Kristen Reed, T. Blaine Crowley, Ian Campbell, Beverly S. Emanuel, Victoria Giunta, Audrey Green, Daniel E. McGinn, Bekah Wang, Elaine H. Zackai, Donna M. McDonald-McGinn

15:16 **031** **Cardiac Disease in Patients with Distal 22q11.21-3 Deletions (LCR22D-E, D-F, D-G, E-F)**
Tanner J. Nelson, Daniel E. McGinn, Bekah Wang, Victoria Giunta, Lydia Rockart, Audrey Green, Oanh Tran, Daniella Miller, Elaine H. Zackai, T. Blaine Crowley, Beverly S. Emanuel, Elizabeth Goldmuntz, Bernice Morrow, and Donna M. McDonald-McGinn

15:20 **032** **Chromatin Modifiers to Elucidate the Phenotypic Variability of Congenital Heart Disease in 22q11.2DS** *
Daniella Miller and Bernice E. Morrow

15:25 **Q&A**

15:40 **033** **Ipsc-Derived Thymic Epithelial Cells Promote Tcrab and Regulatory T Cell Reconstitution in Athymic NSG-Nude Mice**
Hanh Dan Nguyen, Abdulvasey Mohammed, Wenqing Wang, Kelsea M. Hubka, Martin Arreola, Zihao Zheng, Priscila Slepicka, Benjamin D. Solomon, Vittorio Sebastiano, Andrew Gentles, and Katja G. Weinacht

15:50 **034** **Single Cell RNA-Seq Identifies Significant Differences in CD4 T Cell Populations Between 22q11.2 Deletion Syndrome and Controls**
Nouf Alsaati, Montana Knight, Kelly Maurer, and Kathleen Sullivan

16:00 **035** **Elucidating Humoral Immunity and Vaccine Response in Pediatric Patients with 22q11.2 Deletion Syndrome: A Retrospective Case Series** *
Laura Gutierrez, Raul Escobar, Hanadys Ale

16:07 **036** **Four Patients with Severe Immune Related Complications Including Lymphoma and Interstitial Lung Disease**
Jenny Lingman Framme, Annika Malmgren, Vanda Friman, Sólveig Óskarsdóttir

16:14 **037** **A Cohort Study Demonstrating Atypical Characteristics of Iron Deficiency in Patients With 22q11.2DS**
T Blaine Crowley, Donna M McDonald-McGinn, and Michele P Lambert

16:20 **Q&A**

16:30 ***AFTERNOON TEA AND POSTER VIEWING ON THE COVERED TERRACE***
Even Poster Authors Present

SESSION 4: THYROID, PARATHYROID, AND GROWTH

17:00 **038** **Occurrence of Hypothyroidism and Hyperthyroidism in 22q11.2 Deletion Syndrome**
Vaneeta Bamba, Bekah Wang, T. Blaine Crowley, Victoria Giunta, Audrey Green, Elaine H. Zackai, Beverly S. Emanuel, Donna M. McDonald-McGinn, Edna E. Mancilla, Lorraine Katz

17:07 **039** **Association of thyroid and parathyroid disease in 22q11.2 deletion syndrome patients followed at Nationwide Children's Hospital** *
Bianca Zapanta, Cole Simons, Kathryn Stephens Obrynba, Scott E. Hickey

17:14 **040** **Outcomes of Provocative Growth Hormone Testing in Patients with 22q11.2 Deletion Syndrome and Short Stature** *
Rachel Brown, Ian Campbell, T. Blaine Crowley, Victoria Giunta, Daniel McGinn, Beverly S. Emanuel, Elaine H. Zackai, Donna M. McDonald-McGinn, Vaneeta Bamba, Lorraine E.L. Katz

17:21 **041** **Growth Hormone Treatment in Patients with 22q11.2 Deletion Syndrome and Short Stature**

Lorraine E. Levitt Katz, Rachel Brown, Ian Campbell, T. Blaine Crowley, Victoria Giunta, Audrey Green, Daniel E. McGinn, Bekah Wang, Edna Mancilla, Beverly S. Emanuel, Elaine H. Zackai, Donna M. McDonald-McGinn, Vaneeta Bamba

17:28 **042** **Association Between Congenital Heart Defects and Growth in the 22q11.2 Deletion Syndrome**

Lisa Briel, Cas L.J. Kruitwagen, Martijn G. Slieker, Hanneke M. van Santen, Michiel L. Houben

18:05 **043** **Children and Young Adults with 22q11.2 Deletion Syndrome and Fractures: A Case Series**

Lama Alzoebie, Bekah Wang, T. Blaine Crowley, Victoria Giunta, Audrey Green, Elaine H. Zackai, Beverly S. Emanuel, Donna M. McDonald-McGinn, Vaneeta Bamba, Lorraine Katz, Victor Ho-Fung, and Edna E. Mancilla

18:12 **044** **Obesity and Metabolic Syndrome in Adults with 22q11.2 Deletion Syndrome**

Hester Jaspers Faijer-Westerink, Emma N.M.M. von Scheibler, Elisabeth F.C. van Rossum, Thérèse A.M.J. van Amelsvoort, Agnies M. van Eeghen, Erik Boot

18:19 **045** **Non-Fasting Triglyceride-Glucose Index as a Marker of Metabolic Syndrome In 22q11.2 Deletion Syndrome**

Sabrina Cancelliere, Tracy Heung, Anne S. Bassett

18:26 **046** **Trajectory of Cardiometabolic Conditions in Adults with 22q11.2 Deletion Syndrome**

Sarah L. Malecki, Tracy Heung, Samantha Morais, Refik Saskin, Drew Wilton, Anne S. Bassett

18:35 **Q&A**

18:45 ***FAMILY VOICES – MARC AND BARBI WEINBERG***

19:00 **Adjourn**

20:00 POOLSIDE WELCOME RECEPTION

Signature Cocktail and Heavy Hors d'oeuvres

Wednesday ∗ July 17, 2024

6:00 ***REGISTRATION OPEN – BALLROOM I: PENICHE AND SINTRA FOYER***

7:00 ***BREAKFAST – TEMPERA RESTAURANT AND OUTDOOR TERRACE***

SESSION 5: HEENT, EARLY DEVELOPMENT, AND SLEEP

8:00 **047** **Ophthalmologic Manifestations Associated with 22q11.2 Copy Number Variants: An Update**

Brian J. Forbes, Lydia Rockart, Victoria Giunta, Daniel E. McGinn, Audrey Green, Bekah Wang, Oanh Tran, Elaine H. Zackai, Beverly S. Emanuel, Jane C. Edmond, Monte Mills, William Anninger, Gil Binenbaum, T. Blaine Crowley, and Donna M. McDonald-McGinn

8:07 **048** **Prevalence of Preterm Birth and Polyhydramnios in Association with 22q11.2 Deletion Syndrome**

Hayley Ron, T. Blaine Crowley, Jennifer Borowka, Victoria Giunta, Daniel E. McGinn, Bekah Wang, Audrey Green, Lydia Rockart, Oanh Tran, Beverly S. Emanuel, Oksana Jackson, Ian Jacobs, Karen Zur, Lisa Elden, Maria Mascarenhas, Julie Moldenhauer, Elaine H. Zackai, Ian Campbell, Kathleen Valverde, and Donna M. McDonald-McGinn

8:14 **049** **Tracheoesophageal Anomalies in Association with the Chromosome 22q11.2 Deletion Syndrome** *

Bekah Wang, Daniel E. McGinn, Victoria Giunta, T. Blaine Crowley, Audrey Green, Erica Schindewolf, Julie Moldenhauer, Lisa Elden, Ian Jacobs, Karen Zur, Beverly S. Emanuel, Elaine H. Zackai, Maria Cristina Digilio, and Donna M. McDonald-McGinn

8:21 **050** **Gastroesophageal Reflux Disease and Associated Comorbidities in Patients with 22q11.2 Deletion Syndrome**

Asim Maqbool, T. Blaine Crowley, Victoria Giunta, Daniel E. McGinn, Bekah Wang, Audrey Green, Lydia Rockart, Lauren Lairson, Oanh Tran, Beverly S. Emanuel, Elaine H. Zackai, Ian Campbell, Donna M. McDonald-McGinn, Prasanna Kapavarapu, Maria Mascarenhas

8:27 **051** **Prevalence of Enteral Feeding and Subsequent Intervention in Patients with 22q11.2 Deletion Syndrome**

Maria Mascarenhas, Victoria Giunta, T. Blaine Crowley, Daniel E. McGinn, Bekah Wang, Audrey Green, Lydia Rockart, Oanh Tran, Beverly S. Emanuel, Elaine H. Zackai, Asim Maqbool, Prasanna Kapavarapu, Donna M. McDonald-McGinn

8:34 **052** **Dysphagia in Children with 22q11.2 Deletion Syndrome and 22q Duplication Syndrome**
Jill Arganbright, Jana Ghulmiyyah, Srivats Narayanan, Lauren Bartik, Meghan Tracy, Hung-Wen Yeh, Janelle Noel-MacDonnell, Jake Schneider

8:41 **053** **Exploring the Presentation and Mechanism of Dysphagia in Adults with 22q11.2 Deletion Syndrome**
Samantha D'Arcy, Tracy Heung, Nikolai Reyes, Anne S. Bassett

8:45 **054** **Dental Caries and Malocclusion in Patients with 22q11.2 Deletion and Duplication Syndromes** *
Shalin N. Shah, Cynthia Solot, Oksana Jackson, Audrey Green, Bekah Wang, Victoria Giunta, T. Blaine Crowley, Beverly S. Emanuel, Elaine H. Zackai, Lorraine L. Katz, L. Yap, Donna M. McDonald-McGinn, Hyun Duck Nah

8:49 **055** **Guidelines on the Dental Management of Individuals With 22q11.2DS** *
Charlotte Lenes, Rohan Dasari, Michelle Scott

8:53 **056** **Surgical Decision Making for Patients with 22q11.2 Deletion Syndrome and Velopharyngeal Dysfunction: A Clinical Update**
Jill M Arganbright

9:00 **057** **Long-term Changes in Post-Operative Hypernasality Over Time in Patients with 22q11.2 Deletion Syndrome and Velopharyngeal Insufficiency**
Sylvie Render, Alexander Szymczak, Laura H. Swibel Rosenthal

9:07 **058** **Prevalence of Permanent Hearing Loss and Ear Malformations in Children with Distal LCR22D-LCR22E 22q11.2 Deletions**
Lisa Elden, Daniel E. McGinn, Victoria Giunta, Bekah Wang, Audrey Green, Lydia Rockart, Oanh Tran, Beverly S. Emanuel, Conor Devine, Scott P. Bartlett, Elaine H. Zackai, T. Blaine Crowley, and Donna M. McDonald-McGinn

9:13 **Q&A**

9:25 **059** **Factors Supporting Better Language Outcomes in Children With 22q11.2 Deletion Syndrome**
Cynthia Solot, Victoria Giunta, Daniel E. McGinn, Lydia Rockart, Bekah Wang, Audrey Green, Oanh Tran, Beverly S. Emanuel, Elaine H. Zackai, Oksana Jackson, Katherine Baum, Edward Moss, T. Blaine Crowley, and Donna M. McDonald-McGinn

9:32 **060** **Language Abilities of School-Aged Children With 22q11.2 Copy Number Variants: A Two-Site Study** *

Jente Verbesselt, Cynthia B. Solot, Ellen Van Den Heuvel, Jeroen Breckpot, Donna M. McDonald-McGinn, Inge Zink and Ann Swillen

9:36 **061** **Linguistic Profile in Children and Adolescents with 22q11 Syndrome** *

Noelia Santos-Muriel, Noelia Pulido García, Javiera Espinosa Villarroel, Patricia López Resa, Esther Moraleda Sepúlveda

9:42 **062** **Serial Order Short-Term Memory and Vocabulary In 22q11.2 Deletion Syndrome** *

Jantine Wignand, Tessel Boerma, Iris Selten, Emma Everaert, Michiel Houben & Frank Wijnen

9:46 **063** **Early Development and Longitudinal Data in Children With 22q11.2 Duplication** *

Jente Verbesselt, Jeroen Breckpot, Inge Zink and Ann Swillen

9:53 **064** **A Comprehensive Overview of Neurodevelopmental Symptoms in Adolescents with 22q11.2 Deletion Syndrome – A Dimensional Perspective** *

Iris Selten, Jill Blok, Tessel Boerma, A. A. A. Manik J. Djelantik, Michiel Houben, Frank Wijnen, Janneke Zinkstok, & Jacob A. S. Vorstman & Ania M. Fiksinski

9:57 **065** **Irritability in Young People With Copy Number Variants Associated With Neurodevelopmental Disorders (ND-Cnvs), Including 22q11.2 Deletion Syndrome** *

Jessica H. Hall, Samuel. J.R.A Chawner, IMAGINE-ID consortium, Jeanne Wolstencroft, David Skuse, Peter Holmans, Michael J. Owen, Marianne B.M. van den Bree

10:01 **066** **Polysomnography Findings in Patients With 22q11.2 Deletion Syndrome Related To Tonsillectomy**

Jill Arganbright, Bryan Hankey, Hannah Brown, Meghan Tracy, Janelle Noel-Macdonnell, Yaslam Balfaqih, Dave Ingram, Lisa Elden, Oksana Jackson, Blaine Crowley, Christopher Cielo, Donna McDonald-McGinn

10:05 **067** **Tonsillectomy in Children With 22q-Related Disorders**

Jill Arganbright, Hannah Brown, Bryan Hankey, Meghan Tracy, Janelle Noel-MacDonnell

10:09 **068** **Sleep Difficulties Related to Psychopathology and Neurocognition in People With 22q11.2 Deletion Syndrome**

Raquel E. Gur, Margaret C. Souders, Kosha Ruparel, Tyler M. Moore, T. Blaine Crowley, Elaine H. Zackai, Beverly S. Emanuel, Donna M. McDonald-McGinn, Ruben C. Gur

10:13 **069** **Sleep and Cognition in Individuals with 22q11.2 Deletion Syndrome** *

Kathleen P. O'Hora, Charles Schleifer, Jennifer Xu, Elizabeth Bondy, Hoki Fung, Leila Kushan-Wells, Jared M. Saletin, Carrie E. Bearden

10:20 **Q&A**

10:30 ***COFFEE BREAK AND POSTER VIEWING ON THE COVERED TERRACE***
Even Authors Present

SESSION 6: SPINE AND BRAIN

11:00 **070** **Prospective Natural History Study of Idiopathic-like Scoliosis in Patients with 22q11.2 Deletion Syndrome, Starting Before its Pathological Onset** *
Lafranca Peter, de Reuver S, Abdi A, Kruijt MC, Houben ML, Ito K, Castelein RM, Schlösser TPC

11:04 **071** **Tethered Spinal Cord in Association with 22q11.2 Deletion Syndrome**
Madeline Chadehumbe, Sarah E. Hopkins, Victoria Giunta, Daniel E. McGinn, Bekah Wang, Audrey Green, Lydia Rockart, Oanh Tran, Beverly S. Emanuel, Elaine H. Zackai, Sam Alperin, T. Blaine Crowley, and Donna M. McDonald-McGinn

11:11 **072** **Possible Causes of Lower Extremity Pain in Children with 22q11.2 Deletion Syndrome - A Retrospective Cohort Study**
Michiel L. Houben, Femke G.M. van den Helder

11:15 **073** **Occipital Frontal Circumference in 22q11.2 Deletion Versus 22q11.2 Duplication Syndromes**
Sarah Hopkins, Victoria Giunta, Daniel E. McGinn, Bekah Wang, Audrey Green, Lydia Rockart, Lauren Lairson, Oanh Tran, Beverly S. Emanuel, Elaine H. Zackai, T. Blaine Crowley, Ian Campbell, Sam Alperin, Madeline Chadehumbe, Donna M. McDonald-McGinn

11:22 **074** **Neuroradiological Findings in a Large, Unselected Clinical Sample of Individuals with 22q11.2 Deletion Syndrome (22q11.2DS)**
J. Eric Schmitt, Jenna Schabdach, Simon Smerconish, David Roalf, T. Blaine Crowley, Victoria Giunta, Daniel E. McGinn, Elaine H. Zackai, Beverly S. Emanuel, Sarah Hopkins, Madeline Chadehumbe, Raquel E. Gur, Donna M. McDonald-McGinn, and Aaron Alexander-Bloch

11:29 **075** **Neuroradiologic Findings in 22q11.2 Duplication Syndrome and Comparison to 22q11.2 Deletion Syndrome***
Samuel Alperin, Sarah E. Hopkins, T. Blaine Crowley, Daniel E. McGinn, Lauren Lairson, Victoria Giunta, Beverly S. Emanuel, Elaine H. Zackai, and Donna M. McDonald-McGinn

11:36 **076** **Local Cerebellar Dysplasia and Motor Learning Deficit in 22q11.2 Deletion Syndrome are Attributed to a Skeletal Deformity**
Tae-Yeon Eom, J Eric Schmitt, Yiran Li, Audrey Bonnan, Christopher M. Davenport, Khaled Khairy, David R. Roalf, Raquel E. Gur, Beverly S. Emanuel, Donna M. McDonald-McGinn, Jason M. Christie, Paul A. Northcott, Stanislav S. Zakharenko

11:45 **Q&A**

11:55 **077** **Presence, Severity, and Functional Associations of Incomplete Hippocampal Inversion in 22q11.2 Deletion Syndrome**

David Roalf, Donna M. McDonald-McGinn, J. Eric Schmitt, Sarah Hopkins, Adam Czernuszenko, Ally Atkins, Margaret Pecsok, Aaron Alexander-Bloch, T. Blaine Crowley, R. Sean Gallagher, Emily McClellan, Daniel E. McGinn, Paul J. Moberg, Kosha Ruparel, Bruce I. Turetsky, Lauren White, Elaine H. Zackai, Ruben C. Gur & Raquel. E. Gur

12:02 **078** **Synaptic-related Developmental Dysconnectivity in 22q11.2 Deletion Syndrome**

Filomena Grazia Alvino, Silvia Gini, Antea Minetti, David Sastre-Yagüe, Charles Schleifer, Alexia Stuefer, Marco Pagani, Caterina Montani, Alberto Galbusera, Francesco Papaleo, Michael Vincent Lombardo, Massimo Pasqualetti, Carrie E. Bearden, Alessandro Gozzi

12:09 **079** **22q11.2 Gene Dosage Effects on Cerebello-cortical Functional Connectivity**

Hoki Fung, Charles Schleifer, Kathleen O'Hora, Leila Kushan, Elizabeth Bondy, Carrie E. Bearden

12:16 **080** **Neurite Orientation Dispersion and Density Imaging in 22q11.2 Deletion and Duplication carriers** *

Rune Boen, Julio Villalon-Reina, Leila Kushan, Nadine Parker, Ibrahim A. Akkouh, Dag Alnæs, Sergiu Pasca, Ruth O'Hara, Matthew John Marzelli, Lara Foland-Ross5, Christina French Chick, Isabelle Cotto, Allan Reiss, Joachim Hallmayer, Ole A. Andreassen, Ida E. Sønderby, Carrie E. Bearden

12:20 **081** **Focal Volumetric Reductions in Subthalamic Nuclei in the 22.11.2 Deletion Syndrome (22q11DS)**

J. Eric Schmitt, David Roalf, Donna M. McDonald-McGinn, Simon Smerconish, Sarah Hopkins, Aaron Alexander-Bloch, T. Blaine Crowley, R. Sean Gallagher, Daniel E. McGinn, Kosha Ruparel, Lauren K. White, Elaine H. Zackai, Anne Bassett, Stanislav Zakharenko, Ruben C. Gur, and Raquel. E. Gur

12:27 **082** **Altered GABA-ergic Short-term Synaptic Plasticity in Prefrontal Cortex of a Mouse Model of 22q11DS**

Gregg W. Crabtree

12:31 **083** **7-Tesla in-vivo 1H-magnetic Resonance Spectroscopy of Glutamate and GABA in 22q11.2 Copy Number Variants.**

Claudia Vingerhoets, Amy Sylvester, Chaira Serrarens, Esther Steijvers-Peeters, Kim Brouwers, David E. J. Linden, Desmond HY Tse and Therese van Amelsvoort

12:35 **084** **Unique Functional Neuroimaging Signatures of Genetic Versus Clinical High Risk for Psychosis**

Charles H. Schleifer, Sarah E. Chang, Carolyn M. Amir, Kathleen P. O'Hora, Hoki Fung, Leila Kushan-Wells, Jee Won Kang, Eileen Daly, Fabio Di Fabio, Marianna Frascarelli, Maria Gudbrandsen, Wendy R. Kates, Declan Murphy, Jean Addington, Alan Anticevic, Kristin S. Cadenhead, Tyrone D. Cannon, Barbara A. Cornblatt, Matcheri Keshavan, Daniel H. Mathalon, Diana O. Perkins, William Stone, Ming Tsuang, Elaine Walker, Scott W. Woods, Lucina Q. Uddin, Kuldeep Kumar, Gil Hoftman, Carrie E. Bearden

12:42 **085** **Neuromelanin-sensitive Magnetic Resonance Imaging as a Proxy for Dopamine and Norepinephrine Dysfunction in 22q11.2 Deletion/Duplication Syndrome: Interim Results***

Amy Sylvester, Jeltje Spapens, Chaira Serrarens, Amée Wolters, Nikos Priovoulos, Desmond Tse, Dimo Ivanov, Benedikt Poser, David Linden, Thérèse van Amelsvoort & Claudia Vingerhoets

12:45 **Q&A**

13:00 ***LUNCH – TEMPERA RESTAURANT AND OUTDOOR TERRACE***

SESSION 7: TRANSITION, TRANSCRIPTION, AND iPSCS

14:00 **086** ***Invited Presentation*** **- Transition from Youth to Adult Care, Most Recent Insights and Future Challenges**

Therese van Amelsvoort, Maastricht, the Netherlands

14:10 **087** ***Invited Presentation*** **- Cognitive, Adaptive and Daily Life Functioning in Adults with 22q11.2 Deletion Syndrome**

Ann Swillen, Leuven, Belgium

14:20 **088** **Global Burden of Clinical Conditions in 405 Canadian Adults with 22q11.2 Deletion Syndrome ***

Christina Blagojevic, Tracy Heung, Sarah L. Malecki, Sabrina Cancelliere, Vikita Mehta, Brigid Conroy, Anne S. Bassett

14:27 **089** **Secular and other Trends in 22q11.2 Deletion Syndrome at Transition to Adult Care and Beyond**

Tracy Heung, Christina Blagojevic, Lisa Palmer, Samantha D'Arcy, Maria Corral, Anne S. Bassett

14:34 **090** **An Examination of Select Functional Outcomes in Adults with 22q11.2 Deletion Syndrome**

Lisa Palmer, Tracy Heung, Sierra McNulty, Maria Corral, Anne S. Bassett

14:40 **Q&A**

14:50 **091** ***Invited Presentation*** **- Systematic Genetic Dissection of 22q11.2-Linked Genes In Vivo With AAV-Perturb-Seq**
Randall J. Platt, Zurich, Switzerland

15:05 **092** **Premature Neurogenesis Prefigures Loss of Upper Layer Cortical Neurons in 22q Model Mice**
Thomas Maynard, Shah Rukh, Zachary Erwin, Anthony LaMantia, Daniel Meechan

15:15 **093** **Subtype-specific Alterations in Interneuron Activity During Learning in the 22q11.2 Deletion CA1**
Stephanie Herrlinger, Bovey Y Rao, Anna L Tuttman, Haroon Arain, Bert Vancura, Tristan Geiller, Erdem Varol, Joseph A Gogos, Attila Losonczy

15:25 **094** **Transcriptional Regulation of Basal Progenitor Cells in the Developing Cortex of 22q11DS Model** *
S. Rukh, D. W. Meechan, T. M. Maynard, Z. Erwin, C. Siggins, A.S. LaMantia

15:32 **095** **Investigating Convergent Cellular Phenotypes of 22q11 and 3q29 Deletions**
Ryan H. Purcell, Maxine I. Robinette, Erica J. Duncan, Joseph F. Cubells, Zhexing Wen, Jennifer G. Mulle, Victor Faundez, Gary J. Bassell

15:35 **Q&A**

15:50 **096** **Generation of Induced Pluripotent Stem Cells Carrying 22q11.2 CNVs as a Model System for Studying Neurodevelopmental Disorders**
Danijela Drakulic, Natasa Kovacevic-Grujicic, Olena Petter, Mina Peric, Goran Cuturilo, Ivana Simeunovic, Jovana Kostic, Danijela Stanisavljevic Ninkovic, Adrian J. Harwood, Milena Stevanovic

15:54 **097** **Protein and RNA levels in 22q11.2DS iPSC Derived Neurons Compared to Controls over a 100-day Time Course** *
Sabrina Burton, Gemma Wilkinson, Adrian Harwood, Lawrence Wilkinson

16:00 **098** **Multi-modality Functional Genomics Analysis of the Effects of the 22q11.2 Deletion in Multiple Cell Types, Obtained with Multiple Cell-reprogramming Methods**
Alexander E Urban

16:04 **099** **Generation and Characterization of Human Induced Pluripotent Stem Cell Derived Neuronal Models from Patients with Microdeletion Syndrome and Microduplication Syndrome 22q11.2**
Franziska Radtke, Deniz Gücsavas, Rhiannon V. McNeill, Matthias Nieberler, Georg C. Ziegler, Zora Schickardt, Carolin Kurth, Marcel Romanos, Sarah Kittel-Schneider

16:10 **Q&A**

16:20 ***FAMILY VOICES – CAROL CAVANA***

16:30 ***AFTERNOON TEA AND POSTER VIEWING ON THE COVERED TERRACE***
Odd Authors Present

17:00 ***SPECIAL COLLABORATIVE DISCUSSION***
HORIZON EUROPE – 22q11.2DS iPSC ANALYSIS
Open to all attendees

18:00 **Adjourn**

19:00 22q OLYMPICS AND SUNSET SOCIAL

Thursday ∗ July 18, 2024

6:00 ***REGISTRATION OPEN – BALLROOM I: PENICHE AND SINTRA FOYER***

7:00 ***BREAKFAST – TEMPERA RESTAURANT AND OUTDOOR TERRACE***

SESSION 8: COORDINATION, INTERVENTIONS, AND OUTCOMES

8:00 **100** **The 22q and You Center – A Model for Comprehensive Multidisciplinary Coordinated Care**

T. Blaine Crowley, Victoria Giunta, Daniel E. McGinn, Bekah Wang, Audrey Green, Lydia Rockart, Oanh Tran, Ryan LaPointe, Sam Alperin, Sarah Hopkins, Madeline Chadehumbe, Vaneeta Bamba, Lorraine Katz, Katherine Baum, Ed Moss, Malcolm Ecker, John Flynn, Lisa Elden, Steven Handler, Brian Forbes, Elizabeth Goldmuntz, Oksana Jackson, Cynthia Solot, Thomas Kolon, Asim Maqbool, Maria Mascarenhas, Michael Nance, Kathleen Sullivan, Beverly S. Emanuel, Ian Campbell, Elaine H. Zackai, and Donna M. McDonald-McGinn

8:07 **101** **Characterizing the Spectrum of Clinical Manifestations in 22q11.2 Duplication Syndrome: Insights from an Institutional Experience** *

Gail Budhu, Kristen Facey, Raghuram Reddy, Karla Santoyo, Raul Escobar, Brian Cauff, Hanadys Ale

8:11 **102** **Clinical Review of a Large 22q11.2 Cohort at a Tertiary Centre** *

Megan Dunlop, Alice Roueché, Julia Kenny

8:15 **103** **An Overview of 22q11.2 Diagnostic and Research Facilities in the Countries of The Western Balkan Region**

Goran Cuturilo, Danijela Drakulic, Natasa Kovacevic-Grujicic, Mina Peric, Milena Stevanovic

8:19 **104** **Overview of 22q11.2 Deletion Syndrome: A First Moroccan Pediatric Series**

Asmaa Gaadi, Said Trhanint, Laila Boughenouch, Karim Ouldim, Ahmed Aziz Bousfiha, Mouna Lehlimi

8:23 **105** **Establishing a Support Framework for Children with 22q11.2DS in School Settings in Japan**

Simon Elderton, Chiaki Kitamura, Ai Muro, Yuma Chino, Hiroki Ishiguro, Yasue Horiuchi, Nobuhiko Hayashi, Satoko Nakagomi

8:27 **106** **No-show Clinic Appointments and the Social Determinants of Health in Pediatric Patients with 22q11.2 Deletion Syndrome and 22q Duplication Syndrome**
Jill Arganbright, Meghan Tracy, Adrian Williamson

8:31 **107** **Parent Acceptability of a 22q Multidisciplinary Infant Assessment Clinic to Assess Motor and Cognition in Infants with 22q** *
Cindy Trevino, Tracy Brundage, Emily Gallagher

8:35 **108** **Animation Genetic Counselling Aid for Adults with 22q11.2DS and their Caregivers**
Lisa Palmer, Emily Tjan, Nicholas Woolridge, Samantha D'Arcy, Joanne Loo, Anne S. Bassett

8:40 **Q&A**

8:50 **109** ***Invited Presentation*** **- Neurobiological Insights from Human Cellular Models of the 22q11 Deletion Syndrome**
Ralda Nehme, Boston, MA, USA

9:05 **110** ***Invited Presentation*** **- A Report from the International 22q11.2 Brain and Behavior Consortium and Genes 2 Mental Health Network**
Raquel E. Gur, Philadelphia, PA, USA

9:15 **111** **CNVs Elucidate Rare-variant Associations and Genotype-phenotype Relationships across 6 Major Psychiatric Disorders**
Jonathan Sebat, Omar Shanta, Worrowat Engchuan, Marieke Klein, Adam Maihofer, Jeff MacDonald, Bhooma Thiruvahindrapuram, James Guevara, Oanh Hong, Guillaume Huguet, Maria Kalyuzhny, Caroline Nievergelt, Sandra Sanchez-Roige, Patrick Sullivan, Sebastien Jacquemont, Steve Scherer, and the Autism, ADHD, Bipolar Disorder, Major Depressive Disorder, PTSD, Schizophrenia, and CNV Working Groups of the Psychiatric Genomics Consortium.

9:22 **112** **The Clinical Course of Individuals with 22q11.2 Deletion Syndrome Converting to Psychotic Disorders: a Long-term Multi-center Retrospective Follow-up**
Katerina Kulikova, Maude Schneider, Donna M. McDonald McGinn, Shira Dar, Michal Taler, Stepan Eliez, Raquel E. Gur, Doron Gothelf

9:29 **113** **The Core PsychoPathology Summary (C2PS): A Novel Tool to Harmonize Large Scale Neuropsychiatric Phenotype Collection for Genomic Studies**
Danielle Baribeau, Ania Fiksinksi, Genes to Mental Health Network (G2MH), Carrie E. Bearden, Jacob A.S. Vorstman

9:36 **114** **Neuropsychiatric Presentation in de novo and Inherited 22q11.2 Deletion Syndrome**
Ruben C. Gur, R. Sean Gallagher, Emily J. McClellan, T. Blaine Crowley, Daniel E. McGinn, Elaine H. Zackai, Kosha Ruparel, Tyler M. Moore, Beverly S. Emanuel, Donna M. McDonald-McGinn, Raquel E. Gur

9:45 **Q&A**

9:57 **115** **Integrative Health and 22q11.2 Copy Number Variants: Results of a Patient-family Survey** *
Lydia Rockart, Robin Miccio, Lisa Squires, T. Blaine Crowley, Audrey Green, Victoria Giunta, Donna M. McDonald-McGinn, Maria Mascarenhas

10:04 **116** **Use of Auricular Acupressure for Symptom Control in a Child with 22q11.2 Deletion Syndrome**
Maria R. Mascarenhas, Lisa Squires, Christina L. Szperka, Melissa Crawford, and Donna M. McDonald-McGinn

10:08 **117** **Evaluating the Impact of an Online Coaching Intervention for Parents of Children Diagnosed with the 22q11.2 Deletion Syndrome** *
Holly Carbyn, Patricia Lingley-Pottie, Lisa D. Palmer, Andrea Shugar, Donna M. McDonald-McGinn, Patrick J. McGrath, Anne S. Bassett, Cheryl Cytrynbaum, Ann Swillen & Sandra Meier

10:15 **118** **Assessing the Dietary Impact of an Online Nutrition Program for Adults with 22q11.2 Deletion Syndrome**
Samantha D'Arcy, Lisa Palmer, Maria Corral, Anne S. Bassett

10:20 **Q&A**

10:30 ***COFFEE BREAK AND POSTER VIEWING ON THE COVERED TERRACE***
Odd Authors Present

SESSION 9: DIET, VITAMINS, AND TRADITIONAL THERAPEUTICS

11:00 **119** ***Invited Presentation*** **- Maternal Diet as a Modifier of Congenital Heart Disease in 22q11.2 Deletion Syndrome**
Irene Zohn, Washington, DC, USA

11:15 **120** **Tbx1 Haploinsufficiency Causes Brain Metabolic and Behavioral Anomalies in Adult Mice which are Corrected by Vitamin B12 Treatment**

Marianna Caterino, Debora Paris, Giulia Torromino, Michele Costanzo, Gemma Flore, Annabella Tramice, Elisabetta Golini, Silvia Mandillo, Diletta Cavezza, Claudia Angelini, Margherita Ruoppolo, Andrea Motta, Elvira De Leonibus, Antonio Baldini, Elizabeth Illingworth and Gabriella Lania

11:22 **121** **Exploring the Epigenetic Impact of Vitamin B12 Supplementation on Cardiac Phenotypes in a 22q11.2DS Mouse Model** *

Llull-Albertí, M.V, Ventayol-Guirado, M, Amengual-Cladera, E, Hernández-Rodríguez, J, Merkel, A, Asensio, VJ, Muncunill, J, Rocha, J, Torres-Juan, L, Santos-Simarro, F, Martínez, I, Baldini, A, Illingworth, E, Lania, G, Esteller, M, Heine-Suñer, D

11:29 **122** **Changes in Dietary Vitamin A Dosage Disrupt Compensatory Mechanisms in the Cardiac Phenotype of a 22q11.2 Deletion Syndrome Mouse Model** *

Llull-Albertí, M.V, Amengual-Cladera, E, Ventayol-Guirado, M, Hernández-Rodríguez, J, Asensio, VJ, Muncunill, J, Rocha, J, Torres-Juan, L, Santos-Simarro, F, Martínez, I, Baldini, A, Illingworth, E, Lania, G, Heine-Suñer, D

11:36 **123** **Real-world Treatment of Schizophrenia in Adults with a 22q11.2 Microdeletion**

Anne S. Bassett, Tracy Heung, Lily Van, Nikolai Gil D. Reyes, Erik Boot, Eva W. C. Chow, Maria Corral

11:46 **124** **Historical Perspective of the Role of Fasoracetam in Neurodevelopmental Disorders**

Hakon Hakonarson, Donna M. McDonald-McGinn

11:50 **125** **A Randomized, Double-blind, Placebo-controlled Phase 2 Clinical Trial of NB-001 (fasoracetam) for Neuropsychiatric Symptoms in Children and Adolescents with 22q11 Deletion Syndrome (22q11DS)**

Madeline Chadehumbe, Sarah Hopkins, Raquel E. Gur, Donna M. McDonald-McGinn, Emily R. Gallagher, Kerry D. Conant, Naomi J.L. Meeks, Hakon Hakonarson, Nancy J. Butcher, Danielle Baribeau, Jacob Vorstman

11:57 **126** **Riluzole as Cognitive Enhancer in 22q11.2 Deletion Syndrome?**

Claudia Vingerhoets, Amy Sylvester, Desmond Tse, Chaira Serrarens, Paddy Janssen, Ann Swillen, Elfi Vergaelen, Annick Vogels, Janneke Zinkstok & Therese van Amelsvoort

12:00 **Q&A**

12:15 **127** **TANGO2: What's it to You?**
Donna M. McDonald-McGinn, Victoria Giunta, Daniel E. McGinn, Bekah Wang, Lydia Rockart, Audrey Green, Oanh Tran, Ryan Lapointe, Elaine H. Zackai, Beverly S. Emanuel, T. Blaine Crowley, Madeline Chadehumbe, Sarah Hopkins, Jeroen Breckpot, Ann Swillen, Joris Vermeesch, Maciej Geremek, and Beata A. Nowakowska

12:20 **128** ***Invited Presentation*** **- Prevention, Recognition, and Life Saving Treatment: TANGO2 Deficiency Disorder and Life-Threatening Cardiac Risks among 22q11.2 Patients**
Christina Y. Miyake, Houston, TX, USA

12:30 **129** ***Invited Presentation*** **- Episodic Dystonia, Ataxia, and Weakness in Children with 22q11.21 Deletion Syndrome Should Prompt Consideration of Co-morbid TANGO2 Deficiency Disorder**
Samuel J. Mackenzie, Rochester, NY, USA

12:40 ***FAMILY VOICES- DEBBIE DELOACH***

12:50 **Q&A**

13:00 ***LUNCH – TEMPERA RESTAURANT AND OUTDOOR TERRACE***

SESSION 10: MOVEMENT AND MITOCHONDRIA

14:00 **130** ***Invited Presentation*** **- Mitochondrial Proteins and Neurodevelopment in 22q11.2 Deletion Syndrome**
Michael Granato, Philadelphia, PA, USA

14:15 **131** **Neurovascular Mitochondrial Susceptibility in the 22q11.2 Deletion Syndrome Impacts Blood-Brain Barrier Function and Behavior**
Crockett, A. M, Vélez Colón, M. C, Kebir, H, Iascone, D. M, Cielieski, B, Rossano, A, Sehgal, A, Anderson, S. A, Alvarez, J. I

14:25 **132** **Synaptic Energetics in Schizophrenia Risk and Treatment in the Context of 22q11.2 Deletion Syndrome**
Eleonora Stronati, Adam Rossano, Minna Kim, Raquel Gur, Donna McDonald-McGinn, Stewart Anderson

14:32 **133** **Transcriptional Response to Mitochondrial Dysfunction, and Treatment, in Developing Cortical Projection Neurons**
Daniel Meechan, Shah Rukh, Abra Roberts, Zachary Erwin, Thomas Maynard, Anthony LaMantia

14:38 **Q&A**

14:53 **134** **Prevalence of Parkinson's Disease in 22q11.2 Deletion Syndrome: A Multicenter Study**
Emma N.M.M. von Scheibler, Ann Swillen, Gabriela M. Repetto, Nikolai Gil D. Reyes, Anthony E. Lang, Connie Marras, Mark L. Kuijf, Rob P.W. Rouhl, Agnies M. van Eeghen, Carlos Juri, Annick Vogels, Thérèse A.M.J. van Amelsvoort, Anne S. Bassett, Erik Boot

15:03 **135** **Increased Striatal Dopamine Transporter Binding in 22q11Del versus 22q11Dup Individuals**
Therese van Amelsvoort, Carmen van Hooijdonk, Rik Schalbroeck, Erik Boot, Claudia Vingerhoets, en Jan Booij

15:10 **136** **Expanding the Phenotypic Spectrum of Movement Disorders in 22q11.2 Deletion Syndrome: a Retrospective Study** *
Nikolai Gil D. Reyes, Talyta Cortez-Grippe, Marcus Callister, Emilio Q. Villanueva, Tracy Heung, Anne S. Bassett, Anthony E. Lang

15:20 **Q&A**

15:30 ***AFTERNOON TEA AND POSTER VIEWING ON THE COVERED TERRACE***

SESSION 11: METABOLOMICS AND PROTEOMICS

16:00 **137** **Proteomic Analysis of Plasma in 22q11.2DS**
Kathleen Sullivan, Valentina Frusone, Kelly Maurer, Raquel Gur, T. Blaine Crowley, Beverly S. Emanuel, Donna McDonald-McGinn

16:10 **138** **A Follow-up Study Indicates Inflammatory Factors as Predictors to Cognitive Decline and Psychosis in Individuals With 22q11.2 Deletion Syndrome**
Katerina Kulikova, Shira Dar, Noam Matalon, Ehud Mekori, Ronnie Weinberger, Doron Gothelf, Michal Taler

16:17 **139** **Altered Metabolomic and Proteomic Profiles in Individuals with 22q11.2 Deletion Syndrome**
Marwa Zafarullah, Kathleen Angkustsiri, Hannah Culpepper, Austin Quach, Seungjun Yeo, Blythe P Durbin-Johnson, Heather Bowling, Flora Tassone

16:24 **140** **The Contribution of Genome-wide Tandem Repeat Expansions to Schizophrenia in 22q11.2 Deletion Syndrome**
Ryan K. C. Yuen, Muyang Cheng, Tracy Heung, Yue Yin, Anne S. Bassett

16:35 **Q&A**

16:45 **141** ***Invited Presentation*** **– The Future is Now**
Peter Scambler, London, UK

17:05 **Q&A**

17:10 ***FAMILY VOICES: AN INTERNATIONAL PERSPECTIVE - CAROL CAVANA, DEBBIE DELOACH, ANNE LAWLOR, KIM VAN BEKKUM, MARC AND BARBI WEINBERG, AND JULIE WOOTTON***

Junior Investigator Award
Beata A. Nowakowska, Daniel McGinn, and Daniella Miller

Closing Remarks and 2026 Meeting
Donna M. McDonald-McGinn

Wellness Close
Maria Mascarenhas

17:30 **2024 Meeting Adjourned**

19:30 Buses Depart Hotel Entrance

20:00 SILVER ANNIVERSARY GALA

Portuguese Traditional Ranch Show
Dinner and Dancing
Special Awards Presentation
West Cliffs Club House Restaurant
Obidos, Portugal

Poster Presentations

142 **Implementation of a 22q Tracker to Improve Efficiency of Clinic Visits**
Christina Parrish

143 **Implementation of RN Clinic Prep for Pediatrician Visits using a 22q Focused Assessment Checklist**
Christina Parrish

144 **Bridging the Care Gap: A Multidisciplinary Approach for Adults with 22q11.2 Deletion Syndrome in Florida** *
Hanadys Ale, Evana Valenzuela-Scheker, Roman Yusupov, Joshua Saef, Marea Kefalas, Courtney Laczko, Nancy Carranza, Todd Roth

145 **Co-producing a Healthcare Passport to Improve Quality of Care and Communication Engagement for Young People Living with 22q11DS**
Wesley Mulcahy, Suzanne Kelleher

146 **Transition to Adult Care from a Pediatric 22q Center: Midway Through a Pilot Project**
Hannah Berntson, Matthew Blessing, Emily Gallagher, Patricia Harriman, Anna Meehan, Maria Mills, Christina Parrish, Lex Powers

147 **Genetic Breakthroughs in Neonatal Medicine: Advancements Transforming Newborn Care**
Asmaa Gaadi, Hind Dehbi, Ahmed Aziz Bousfiha, Mouna Lehlimi

148 **Integrated Care for Young People with 22q.11.21 Deletion Syndrome – a Patient, Provider Initiative**
Wesley Mulcahy, Suzanne Kelleher, Marie Louise Healy, Anne Lawlor

149 **Surgical Needs of Patients with 22q11.2 Deletion or Velocardiofacial Syndrome**
Emma Martin, Alyssa Smith, Laura Rosenthal

150 **Prevalences of Comorbid Cardiovascular, Psychiatric, Orthopedic, Endocrinologic and Development-related Diseases in Patients with 22q11.2 Duplication Syndrome – a Systematic Review** *
Carina Sauter, Paula Franz, Laura Kettenstock, Klara Henrich, Matthias Linhardt, Marcel Romanos, Franziska Radtke

151 **Examining Parent of Origin in Patients with de novo 22q11.2 Duplication Syndrome** *
Oanh Tran, Ryan Lapointe, Victoria Guinta, T. Blaine Crowley, Audrey Green, Lydia Rockart, Bekah Wang, Daniel E. McGinn, Steven Pastor, Elaine H. Zackai, Donna M. McDonald-McGinn, and Beverly S. Emanuel

152 **Supernumerary Nipple in Association with 22q11.2 Deletion Syndrome** *
Audrey Green, Victoria Giunta, T. Blaine Crowley, Daniel E. McGinn, Bekah Wang, Lydia Rockart, Lauren Lairson, Oanh Tran, Beverly S. Emanuel, Elaine H. Zackai, Donna M. McDonald-McGinn

153 **Telangiectasia in the Distribution of the Superior Vena Cava – a Novel Phenotype in 22q11.2 Deletion Syndrome: A Case Report**
A. Hollywood, Kate Rigney, A. Irvine, S. Kelleher

154 **Development, Evaluation and Implementation of a Psychoeducation Program for Families Affected by 22q11DS** *
Ania M. Fiksinski, N. van Wijngaarden, K. van Bekkum, J.A.S. Vorstman, H. de Veye, M.L. Houben

155 **The Sleep Detectives: Co-Designing a Protocol for Longitudinal Tracking of Sleep Health and Cognition in Children and Young People Living with Copy Number Variants**
Matt W. Jones, Meg Attwood, Abiola Saka, Christopher Jarrold, Nicholas Donnelly, Julie Clayton, Jeremy Hall, Alexander D. Shaw and Marianne van den Bree

156 **Anxiety and Adaptive Function in Teens with Chromosome 22q11.2 Deletion Syndrome**
Aishworiya Kolli, Jonathan Bystrynski, Byrn Ritter, Flora Tassone, Kathleen Angkustsiri

157 **Talking to the Teacher: the Value of Behavioral Observations of Children with 22q11DS in the Classroom**
Jane Summers, Matisse Blundell, Sarah McGaughey, Katrina Palad, Jacob Vorstman

158 **Leveraging Early Genetic Diagnoses: The Exemplary Case of 22q11.2 Deletion Syndrome in Addressing Caregiver Needs in Neurodevelopmental Disorders**
Polina Perlman Danieli, Jacob Vorstman, Ny Hoang, Jane Summers, Danielle Baribeau, Jessie Cunningham, Benoit H. Mulsant

159 **Development and Delivery of Psychoeducational and Parenting Programmes for 22q11.2DS Families in Ireland**
Veselina Gadancheva, Ahmed Khan, Wesley Mulcahy, Suzanne Kelleher, Fiona McNicholas

160 **Semi-structured Interviews with Parents/Caregivers of Children with 22q11.2 Deletion Syndrome Highlight Areas Where Support is Needed**
Emily R. Gallagher, Brent Collett, Cindy Ola Trevino

161 **Evaluating the Relationship Between Parent Mental Health, Parenting Skills, and Child Behavioural Issues in Families Affected by 22q11.2 Deletion Syndrome** *
Holly Carbyn, Patricia Lingley-Pottie, Lisa D. Palmer, Andrea Shugar, Donna M. McDonald-McGinn, Patrick J. McGrath, Anne S. Bassett, Cheryl Cytrynbaum, Ann Swillen & Sandra Meier

162 **Co-Production: a Transition Clinic for Young People with 22q11.2 Deletion Syndrome**
Wesley Mulcahy, Suzanne Kelleher, Marie Louise Healy, Anne Lawlor

163 **BE-WEHL Wellness Education for Families of Children with Behavioral Health Challenges**
Robin Miccio, Lisa Squires, T. Blaine Crowley, Audrey Green, Lydia Rockart, Donna M. McDonald-McGinn, Maria Mascarenhas

164 **Characteristics of Motor Patterns in Infants with 22q11.2DS: Bayley Trends and Early Signs of Neuromotor Impairment**
Tracy Brundage, Cindy Ola Trevino, Emily Gallagher

165 **Investigating Convergence of Neurodevelopmental Mechanisms between 16p11.2 CNV and Cullin3 (Cul3) using Brain Cortical Organoids**
Luca Trovò, Aline Martins, Gimena Gomez, John Yates III, Alysson R. Muotri, and Lilia M. Iakoucheva

166 **Changes in Brain Structure and Associated Functions in Children with 22q11.2 Deletion Syndrome versus Controls: A six-year Longitudinal Study**
Stephen R. Hooper, Vandana Shashi, Kelly Schoch, & Matcheri S. Keshavan

167 **Neurodevelopmental Disorders and Executive Function in a Clinical Sample of Children and Adolescents with 22q11.2**
Rygvold TW, Midtlyng E, Michael B. Lensing

168 **Threat Sensitivity and Neuropsychiatric Disorders in Individuals with 22q11.2 Deletion Syndrome**
Lauren K. White, Tyler M. Moore, R. Sean Gallagher, Emily J. McClellan, David R. Roalf, T. Blaine Crowley, Victoria Giunta, Audrey Green, Daniel E. McGinn, Bekah Wang, Beverly S. Emanuel, Donna M. McDonald-McGinn, Ruben C. Gur, and Raquel E. Gur

169 **22q Microdeletion Predisposes iPSC derived Microglial like Cells to Increased Activity** *
Kieona Cook, Sonial Lomboroso, Daniel Iascone, Amita Seghal, F. Chris Bennett, Stewart Anderson

170 **Convergent Biology among Copy Number Variants Associated with Schizophrenia**
Mulle Jennifer G, Pollak RM, Pato M, Pato C, Pang Z, Hart R

171 **Analyzing Mitochondrial Deficits in 22q11.2 Deletion in Developing Neural Models** *
Maxine I. Robinette, Victor Faundez, Ryan H. Purcell, Gary J. Bassell

172 **The Role of Screening Tools in the Psychiatric Evaluation of Children with 22q11.2 DS**
Ahmed Khan, Veselina Gadancheva, Rae Lyz Yee, Suzanne Kelleher, Fiona McNicholas

173 **Psychophysiological Deficits in 22q11DS** *
David Parker, Sid Imes, Gabrielle Ruban, Brett Henshey, Nicholas Massa, Grace Lee, Bruce Cuthbert, Opal Ousley, Elaine Walker, Erica Duncan, Joseph Cubells

174 **Environmental Influence on the Patients with 22q11.2 and 16p11.2 Deletions and Duplications** *
Yelyzaveta Snihirova, Therese van Amelsvoort, Claudia Vingerhoets, Mieke van Haelst, David E.J. Linden & Dennis van der Meer

*Indicates Junior Investigator