

A Summary of the 14th Biennial International 22q11.2 Scientific Meeting – July 2026

We hope you had an enjoyable summer! We can assure you that the more than 200 attendees and accompanying persons representing a parade of 22 nations across 5 continents certainly did while gathered for the 14th Biennial International 22q11.2 Scientific Meeting held from July 13th through the 17th on the beautiful island of Crete, the Jewel of Greece. Focusing on education and collaboration, the conference, presented by the 22q11.2 Society (the international scientific body dedicated to understanding chromosome 22q11.2 differences and related conditions), which occurs every two years in summer and aims to deliver cutting edge scholarship with the goal to enhance clinical care while never losing sight of the main focus – patients and families – hit the mark and then some in 2026!

Led by a Program Committee made up of 22 Society Trustees and Advisors including Conference Director, Donna McDonald-McGinn, MS, LCGC (Philadelphia), Associate Conference Director, Anne Bassett, MD (Toronto), and Program Chair, Erik Boot, MD, PhD (Amsterdam), the four-day symposium was all-inclusive, providing unlimited opportunities for scientific exchange and discussion over coffee breaks, lunches, and social functions. The 144 oral and 20 poster presentations provided soup to nuts coverage of every important aspect of chromosome 22q11.2 differences, including presentations analyzing each system in the body, as well as 22q11.2 duplications, distal deletions and duplications, the importance of deletion and duplication size as well as the critical need to examine genes on the non-deleted chromosome 22, most especially *TANGO2*, a gene associated with hypoglycemia, sometimes leading to hypoglycemic seizures and arrhythmia associated with sudden death in a subset of individuals.

Invited speakers included a number of “first timers”, such as basic scientists, Spyros Petrakis, PhD, a local,

who is working tirelessly to better understand the brain and behavior with hopes to develop novel treatment strategies for psychiatric illness, and Sophie Astrof, PhD, from the USA, who like Bernice Morrow, PhD, one of the Society’s “Originals” (the 5 current attendees who have been participating in this meeting since the inaugural meeting in Strasbourg, France in 1998) is investigating cardiac development to better understand congenital heart disease, including ways to prevent it. Additional highlights included an entire morning dedicated to the importance of the immune system such as the hope of bringing laboratory derived thymic tissue to immune compromised patients and the immune system’s possible impact on psychiatric illness - having the potential to lead to an entirely new stream of treatment options! The conference continued with a host of exquisite presentations, including short talks which were some of the most impressive of the meeting. Attendees were regaled with the first ever study examining the gut microbiome in children with chromosome 22q11.2 differences, the significance of the blood-brain barrier, neural circuit dysfunction in relation to developing psychiatric illness in animal models and humans, the window to the brain provided by using high-level (7 Tesla) neuroimaging, the effectiveness and safety of current drugs for behavioral differences, and the need to continue to work collaboratively to move our understanding forward as quickly as possible to help patients and families now. No question, the audience was inspired by the presentation of first-time invited speaker, Ian Campbell, MD, PhD, from the USA, who demonstrated the vast promise of using AI-powered novel tools to “examine the whole elephant” in the electronic health record, allowing researchers to consider the collective impact of a person’s entire medical history on outcomes – in every area. Finally, the audience was fully engaged with the discussions

led by Marc van Hout and Kim van Bakkum from the Netherlands, Carol Cavana and Debbie DeLoach from the USA, Julie Wootton from the UK, and Vesna Vujicic from Serbia, as they represented their respective family organizations under the umbrella of Family Voices, where they posed the provocative questions, “What can we do for one another?” and “What do we both need” – which resoundingly led to a key dialog around the desperate need for fundraising on an international scale as the only definitive way to bring all of our mutual hopes and dreams, including improved quality of life from infancy through adulthood for all people living with chromosome 22q11.2 differences, to a reality. But the meeting did not end on a negative note, on the contrary, attendees were empowered to continue to strive to do even more work and as quickly as possible! This attitude was bolstered even further by the thought provoking and uplifting concluding invited presentation by Original, Peter Scambler, MD, entitled “Towards restoration of the lost folio.” Prof. Scambler cited amazing work that is happening both within and external to the 22q11.2 community which will surely lead to astonishing breakthroughs in the next two years. In fact, he was forecasting new and improved targeted therapies and predicted they will be in use before our next meeting in France in 2028!

To view the 2026 scientific program, please visit www.22qsociety.org

Beyond the science, and with an eye to always fostering opportunities for developing friendships which no doubt leads to both novel and continuing collaborations, the social program was an absolute highlight and a terrific way to celebrate our incredible international 22q community. The Welcome Reception, with local Cretan entertainment, kicked off the group bonding. The 22q Olympics, with 20 teams formed under the express intent of “Mixing It Up” so everyone could meet someone new, worked exactly as planned in solidifying new friendships – occasionally via skinned knees, unquestionably with perspiration-stained shirts, and most definitely with pizza to celebrate all the winners. Finally, the Closing

Dinner and Dancing brought everything home by providing an opportunity to celebrate top talks from junior and senior investigators alike, while sharing a meal with old and now new friends, dancing the night away, and fostering an environment of global teamwork.

In addition to the incredible science, recognition of the outstanding contributions of individuals working tirelessly on behalf of the 22q11.2 scientific and family community is *the* highlight of the meeting. The 2026 award recipients (in order of appearance) included:

- **The Angelo DiGeorge Memorial Medal of Honor** recognizing an outstanding contribution to our understanding and/or treatment of the 22q11.2 deletion syndrome. It was inaugurated in 2010 to commemorate the life and work of Dr. Angelo DiGeorge. The award may be presented to basic scientists working on the molecular, cellular, or developmental basis of the condition, clinical scientists working on the diagnosis and treatment of any aspect of the syndrome,^[SEP] allied health care professionals, educators or welfare workers who make major contributions to the wellbeing of patients and families, or exceptionally, organizations who fund or facilitate the above activities. The 2026 Angelo DiGeorge Medal was presented by “Original” Ann Swillen, PhD, to Sixto Garcia-Miñaur, MD, and Damian Heine-Suñer, PhD of Spain, for their exceptional and longstanding work and dedication to patients and families in both Madrid and Palma de Mallorca, including the family organizations across Spain and for their collegial relationship with colleagues within the 22q11.2 Society and beyond, including hosting the 2014 International 22q11.2 conference in Mallorca.
- **The Unsung Hero Award** is presented to honor an individual having demonstrated outstanding contributions to the 22q11.2 community. In 2022, the 22q11.2 Society renamed the Unsung Hero Award in honor of Founding Member, Clodagh

(Dot) Murphy, MD, PhD, who sadly passed in 2020. Qualities of Unsung Hero Awardees include commitment as evidenced by the number of years of service, hard work and devotion demonstrated by selfless hours of service, collaboration and inclusivity with sister organizations and the research and healthcare community at large, along with making a national and international impact on individuals with 22q11.2DS and/or their families and caregivers. The 2026 Clodagh Murphy Unsung Hero Award was presented by “Original” Donna McDonald-McGinn, MS, LCGC, to Carol Cavana, a Founding Board Member and current Administrative Director for the International 22q11.2 Foundation, USA. Carol, who is mother to 27 year old Louis with the 22q11.2 deletion syndrome, has been working on behalf of families for the International 22q11.2 Foundation since its inception in 2003, having led innumerable events most notably, 22q at The Zoo – Worldwide Awareness Day since 2011. More details about Carol and the Foundation can be found at 22q.org

- **The Peter Scambler Invited Lecture and Outstanding Achievement Award**, inaugurated in 2022, is a featured lecture at the Biennial International 22q11.2 Scientific Conference in honor of Founding Trustee and inaugural Chair (2013-2019), Prof. Peter Scambler. The lecture, awarded by Dr. Scambler and the Trustees of the 22q11.2 Society, kicks off the scientific program. The 2026 Peter Scambler Invited Lecture and Outstanding Achievement Award, was presented by “Original”, Anne Bassett, MD, to Joris Vermeesch, PhD of Belgium. Prof. Vermeesch, an Advisor for the 22q11.2 Society, heads the Laboratory for Cytogenetics and Genome Research at KU Leuven, pioneering genomic technologies to map the causes and mechanisms underlying rare developmental disorders, including chromosome 22q11.2 differences across species. Prof. Vermeesch is an outstanding

collaborator and advocate for personalized patient care.

- **The Special Service Award**, envisaged to commemorate the 20th anniversary of the International 22q11.2 Conference, was inaugurated to recognize the long-term dedication of a clinical/scientific Center that demonstrates outstanding, longstanding, exemplary and unwavering commitment, and contributions to the chromosome 22q11.2 community. Given the rarity of programs that have supported coordinated multidisciplinary patient care, the award has only been presented previously on two occasions. The 2026 Special Service Award was presented by “Original” Peter Scambler, MD, to the KU Leuven Clinical and Research Program from Leuven, Belgium! The Center at KU Leuven, like the previous two programs receiving recognition (the 22q and You Center from the Children’s Hospital of Philadelphia in 2018 and the Dalglish Family 22q Clinic team from the Center for Addiction and Mental Health in Toronto in 2024) KU Leuven has been operational since the mid-1990’s, has been continuously operational, and has contributed to the education of innumerable clinician scientists and countless publications – while remaining, first and foremost – collegial. The award was accepted by Ann Swillen, PhD, Joris Vermeesch, PhD, and Elfi Vergaelen, MD, PhD, on behalf of their expansive and impressive team.
- **Junior Investigator Awards.** All presenters identifying themselves as junior investigators are eligible to receive recognition during and at the conclusion of the meeting based on top oral presentations, top posters, and top session presentations. Junior Investigators are those researchers engaged in Pre-doctoral Studies (prior to having received an MD, PhD, DO, DOT, AUD, etc.) and those with a doctoral degree engaged in post graduate training but having

completed no more than 5 years post-doctoral degree.

Junior Investigators categories are as follows:

- Pre-doctoral Clinical
- Pre-doctoral Basic Science
- Post-doctoral Clinical
- Post-doctoral Basic Science

Top junior investigator presentations receive a certificate suitable for framing, a \$500 travel stipend, and complimentary registration for the subsequent International 22q11.2 Biennial Conference. Top poster presentations receive a poster ribbon.

- The 2026 Junior Investigator Awards were presented by Daniela Miller, 2024 Junior Investigator Award Recipient as follows:
 - Post-doctoral Clinical – Douglas Villarta, MD of Philadelphia, USA and Cristina Angellotto, MD, of Rome, Italy
 - Post-doctoral Basic Science – Christina Michalski, PhD of Atlanta, USA
 - Pre-doctoral Clinical – Wenna Chen, of New York, USA and Lydia Hamilton of Philadelphia, USA
 - Pre-doctoral Basic Science – Marta Sousa Santos of Leuven, Belgium
- In addition, thirty top session presentation awards (2 per session including one junior and one senior investigator) and three top poster presentations were presented by Prof. Peter Scambler

The 22q11.2 Society Trustees, Advisors, and organizers are incredibly grateful to all those who

traveled to Crete and contributed to making this meeting such a brilliant success. Not the least of whom include our volunteers and the sponsors who really made this meeting possible, most notably: The International 22q11.2 Foundation, Natera, Tolerance Bio, Chromosome 22 Central, and Synaptiq Therapeutics.

At the conclusion of the meeting, Dr. Astrof stated that it was “the best conference she had ever attended and plans to definitely be back for all future meetings!” We couldn’t agree more! Until we meet again in France in 2028 - *Yamas!*

Donna M. McDonald-McGinn, MS, LCGC

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