International 22q11.2 Foundation

Newsletter September 2024

Successful Conference in Portugal

The International 22q11.2 Foundation was delighted to provide the lead sponsorship for the 13th Biennial International 22q11.2 Conference in Óbidos, Portugal. The conference supports scientific collaboration across the globe, welcoming 200 scientists, leading healthcare providers, family foundation organizers, patient representatives, and accompanying persons from 22 countries and five continents.

Speakers representing the International 22q11.2 Foundation included Board Members Marc Weinberg, Carol Cavana, and Debbie DeLoach, Founding Board Member and Conference Director Donna McDonald-McGinn, Medical Advisory Board Director Anne Bassett, and Medical Advisory Board Members Ann Swillen, Jacob Vorstman, Madeline Chadehumbe, Michele Lambert, Maria Mascarenhas, Bernice Morrow, Edward Moss, Sólveig Óskarsdóttir, Peter Scambler, Cynthia Solot, and Kathleen Sullivan.

Beyond the extensive scientific program, which included 141 platform presentations and 36 posters, the conference's social program encouraged friend raising and the emergence of new collaborative efforts across international sites.

Please check out pages 2 to 5 to revisit the excitement from Portugal. We look forward to the next conference in 2026!

In this newsletter:

Our Board Members Shared Their Experiences

Marc Weinberg

International 22q11.2 Foundation Founding Board Member and Chair, Marc Weinberg, spoke at the 13th Biennial International 22q11.2 conference in Óbidos, Portugal about his experience as a parent and leader of a family support organization.

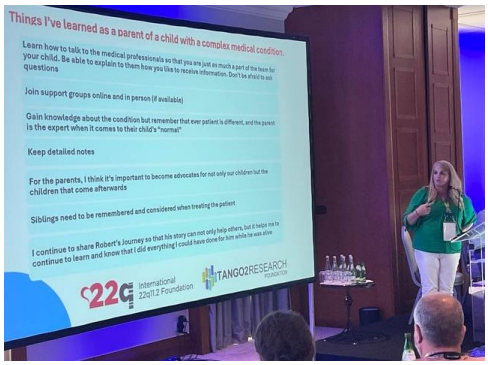
Marc discussed the challenges parents face in advocating for informed care and awareness and opportunities for parents and professionals to collaborate to guide the 22q community into the future.

Carol Cavana

Carol Cavana, parent of Louis and Founding Board Member of the International 22q11.2 Foundation, spoke at the 13th Biennial International 22q11.2 Conference in Óbidos, Portugal. She shared her personal experience raising Louis, who has 22q11.2 deletion syndrome.

Carol presented the medical obstacles that Louis faced in his early years and highlighted his progress with the support of the medical community. She touched on the importance of his active engagement with family, sports, art, and, most importantly, the community. Carol further discussed the challenges that families face with transitioning care from pediatric to adult providers who are knowledgeable about their care needs.

Debbie Deloach



Debbie DeLoach, a Board Member of the International 22q11.2 Foundation and a parent of a child with 22q11.2 deletion syndrome, spoke alongside the TANGO2 Research Foundation at the 13th Biennial International 22q11.2 conference in Óbidos, Portugal. She shared her personal experience with her son, Robert, who had 22q11.2 deletion syndrome and TANGO2 deficiency and sadly passed away in 2015.

Debbie's efforts to share her story and bring awareness to TANGO2 deficiency in association with 22q11.2 deletion syndrome were profoundly impactful to the medical professionals in attendance. The presentations from Debbie and Drs. Samuel Mackenzie and Cristina Miyake from the TANGO2 Research Foundation will undoubtedly create waves in clinics across the globe, promoting better recognition of the symptoms of TANGO2 deficiency in 22q patients as well as novel B vitamin treatment for these patients.

Family Voices Panel at the Conference

At the 13th Biennial International 22q11.2 conference in Óbidos, Portugal, International 22q11.2 Foundation Board Members Marc Weinberg, Carol Cavana, and Debbie DeLoach, 22q11 Ireland founder and Chair Anne Lawlor, Max Appeal Chair Julie Wooton, and Stichting Steun 22q11 Founder and Director Kim Van Bekkum and Chair Mark Van Hout led a panel about their experiences as parents and leaders of charity organizations.

The "Family Voices" discussion emphasized an international perspective on how parents and nonprofits can collaborate with professionals to advance understanding and support for individuals with 22q differences.

Dutch Advocate Received Unsung Hero Award

On behalf of the International 22q11.2 Foundation, we extend our congratulations to **Kim van Bekkum**, Founder and Director of Stichting Steun 22q11, on receiving the prestigious ***"Unsung Hero Award"*** at the 13th Biennial International 22q11.2 Conference in Óbidos, Portugal.

This award, presented biennially by the 22q11.2 Society, honors outstanding contributions to the 22q11.2 community from an individual who demonstrates extraordinary commitment, hard work, devotion, collaboration, and inclusivity through research, awareness, and global collaboration. Kim's tireless efforts raising awareness and supporting families in The Netherlands and beyond through Stichting Steun 22q11 have been undeniably heroic. Congratulations, Kim!

The 22q11.2 Society hosts the International 22q11.2 conference once every 2 years. It is a gathering for healthcare providers, scientists, junior investigators, and family support organizations. In the most recent conference in Portugal, individuals from 22 countries across 5 continents came together to share their respective work and mutli-center collaborations. Please visit the Meetings page of the Society to see photos and programs.

Awards Presented at the Conference

The Angelo DiGeorge Medal of Honor

Sólveig Óskarsdóttir

From left to right: Prof. Ann Swillen, Dr. Sólveig Óskarsdóttir (award recipient), and Prof. Donna McDonald-McGinn. All three are Trustees of the 22q11.2 Society and Members of the Medical Advisory Board of the International 22q11.2 Foundation.

The Unsung Hero Award

Kim van Bekkum

From left to right: Kim van Bekkum (award recipient), Dr. Erik Boot (Trustee of the 22q11.2 Society), and Prof. Donna McDonald-McGinn (Trustee of the 22q11.2 Society and Member of the Medical Advisory Board of the International 22q11.2 Foundation).

The Peter Scambler Invited Lecture Award

Stephen Scherer

From left to right: Dr. Stephen Scherer (award recipient) and Dr. Anne Bassett (22q11.2 Society Trustee and Member of the Medical Advisory Board of the International 22q11.2 Foundation)

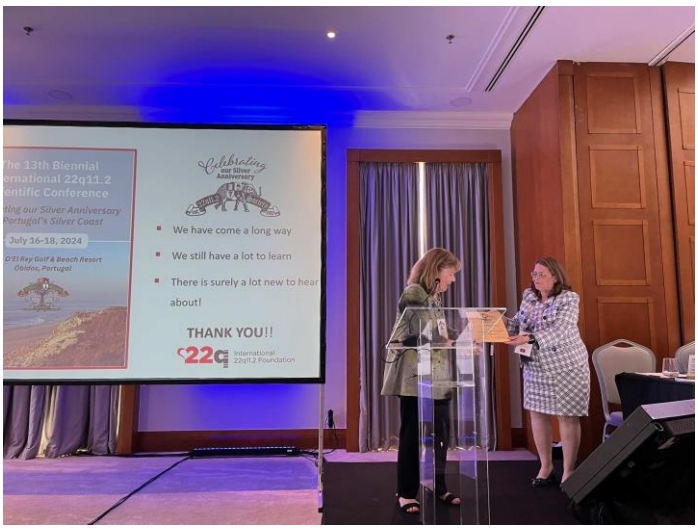
For information about the major awards, please visit the **Awards** page of the 22q11.2 Society website.

Awards Presented at the Conference (continued)

The Special Service Award

The Dalglish Family 22q Clinic, Directed by Anne Bassett

Representatives of the Dalglish Family 22q Clinic Team (award recipient). From left to right: Dr. Erik Boot (Trustee of the 22q11.2 Society), Samantha D'Arcy, Lisa Palmer, and Dr. Anne Bassett (Trustee of the 22q11.2 Society and Member of the Medical Advisory Board of the International 22q11.2 Foundation).



The Lifetime Achievement Award

Beverly Emanuel

From left to right: Prof. Beverly Emanuel (award recipient) and Prof. Donna McDonald-McGinn (Trustee of the 22q11.2 Society and Member of the Medical Advisory Board of the International 22q11.2 Foundation).

The Junior Investigator Award

Daniella Miller

Jente Verbesselt

Christina Blagojevic

From left to right: Beata Nowakowska (Trustee of the 22q11.2 Society); current award recipients Christina Blagojevic (2024), Daniella Miller (2022 & 2024), and Jente Verbesselt (2024); and 2022 recipient Daniel McGinn

Congratulations to the recipients of these major awards as well as the participants who won **Top Presenter Awards** in their sessions!

Light up the Night for 22q11.2!

Greetings from the International 22q11.2 Foundation, Inc.

November is 22q11.2 Awareness Month!

22q11.2 deletion and duplication syndromes are under-recognized genetic conditions that affect about 1 in 1000 pregnancies and 1 in 2000 live births. These conditions occur when a very small piece of chromosome 22 is missing or extra, resulting in the loss or gain of about 50 genes that help direct how the body is formed and functions. The most common associated features include birth defects (such as heart, palate and kidney problems), multiple medical conditions (including low calcium, difficulty fighting infection, feeding and swallowing differences, and seizures), developmental delay, learning differences, and behavioral health problems, such as ADHD, anxiety, autism, and psychiatric illness. Many individuals require care from multiple specialists across the lifespan, but there is also very broad variability.

Most deletions and duplications are the same size with a subset being a bit smaller. All can occur randomly for the first time in the person who has the chromosome difference and nothing that the parents did or did not do cause it to occur. However, once a person has the deletion or duplication there is a 50% chance of passing it on in every pregnancy.

Chromosome 22q11.2 differences are the most common chromosomal conditions after Down Syndrome. However, most people have never heard of chromosome 22, and some people with these differences spend years searching for a diagnosis. To increase public awareness, in 2018, La Asociación Síndrome 22q11 in Spain launched an awareness campaign entitled “Luces por el 22q” (Lighting the Night for 22q). Buildings and monuments were illuminated in **red** on **November 22** (22/11 in many parts of the world but 11/22 in North America) as a play on the name of the chromosome difference. Cities in Belgium, Poland, Canada, Finland, Germany, and the USA soon followed. At the bottom of this page are example photos from November 22 of previous years, where buildings and even the Niagara Falls were lit up in **red** to support 22q awareness. Please consider joining this international endeavor so that no child or adult struggles to find a diagnosis in a timely fashion. Therefore, we respectfully request that _____ in _____ be lit up in **red** on **November 22, 2024** to raise awareness for **chromosome 22q11.2 deletion and duplication syndromes**. Questions? Please contact the International 22q11.2 Foundation, Inc. at 001.877.739.1849 or info@22q.org

Thank you in advance for your kind consideration.

The International 22q11.2 Foundation, Inc.
PO Box 532
Matawan, NJ 07747, USA
https://www.22q.org/

22q
International
22q11.2 Foundation

22q Glossary

Third edition now available!

Click this link to access the glossary

22q GLOSSARY (3rd Edition)

To help families understand terms that they may encounter, we have been developing a glossary for 22q differences. We have just published the third edition, which contains **269** terms with explanations plus **29** additional terms with redirections ("See ___"). The latest version had a big increase in the number of terms related to speech, brain/nerves, and the digestive system, and is now available on the **Health Conditions Explained** section of our website. We will add more terms in future editions. On the right are some of the newest entries.

The International 22q11.2 Foundation would like to thank everyone who has contributed to the development of the 22q Glossary.

Vocal Nodules – Vocal nodules are little bumps, or callouses, on the inner edge of the **vocal cords**. They affect the way the vocal cords contact each other and vibrate, leading to a voice that is hoarse, breathy, and strained. They result from overuse or straining the voice. [See "Voice Disorders" in the Speech Series of the Health Conditions Explained page of our website for more information.]

Inferior Pharyngeal Constrictor (Cricopharyngeus Muscle) – The inferior pharyngeal constrictor is a muscle at the top of the esophagus. When it relaxes, food and drinks can go from the throat into the esophagus. See "Swallowing and Dysphagia" in the Gastrointestinal (GI) Series of the Health Conditions Explained page of our website for more information.

Myoclonus – Myoclonus are uncontrolled, sudden, startle-like movements that occur in one part of the body or all over the body. Some types of myoclonus happen in healthy people and do not cause concern – these include hiccups and jerking that happen just before falling asleep. However, problems with the brain and nerves can lead to myoclonus, and these phenomena have been reported in adults with 22q11.2DS. Treatment or the strategy for symptom control for myoclonus depends on the underlying cause.

Health Conditions Explained

For information developed for individuals and families affected by 22q differences, please visit the "**Health Conditions Explained**" section of our website. The information sheets are generated by individuals who are highly involved in 22q-related education and communication, and the content is carefully reviewed by 22q experts. We will add more contents on various topics as they become available.

The topics published so far are:

- **Heart** Series – 5 sheets
- **Dental** Series – 2 sheets
- **Palate** Series – 6 sheets
- **Mental Health** Series – 9 sheets
- **Gastrointestinal** (GI) Series – 13 sheets
- **Speech** Series – 8 sheets
- 22q **Glossary** (Third Edition)

SPEECH SERIES – June 2024

- Introduction to Speech
- Consonants and Vowels in English
- Speech Disorders in Individuals with 22q11.2 Deletion Syndrome – an Overview
- Speech Disorders in Individuals with 22q11.2 Duplication Syndrome – an Overview
- Voice Disorders
- Speech Sound Disorders
- Motor Speech Disorders
- How Parents and Professionals Can Help

Mpox – Quick Facts

The World Health Organization (WHO) recently declared Mpox a public health emergency of international concern, which is the highest level of alarm under international health law. Here is some important information about this disease.

Basic Facts

- Mpox is a **highly infectious disease** caused by the Mpox virus.
- The most common Mpox symptoms include **lesions, fever, rash, and swollen lymph nodes**. It can lead to death.
- Individuals with weakened immune systems may get seriously ill from Mpox.

The Spread of Mpox

- The Clade I Mpox that is spreading in Africa now causes **disease that is more serious** than the type in 2022 (Clade II).
- The current Mpox outbreaks is severely affecting a number of African countries, but has not spread widely across the globe.
- As of September 6th, 2024, there have been no reported cases of Clade I Mpox in USA. Clade II Mpox (from 2022) continues to circulate at low levels.
- Mpox spreads when a person **touches** the rashes or sores of an infected patient, their secretions, and the items they use.
- **Anyone can catch Mpox**. Many children in Africa have been affected in 2024, likely due to crowded households, contacts with infected animals, and lack of information about the spread. Since the environmental and medical situations are different in USA, the CDC does not expect a similar elevated risk for American children.

Treating Mpox

- International health authorities are **prioritizing** the prevention of Mpox transmission as well as its treatment.
- Mpox and smallpox are very similar, so some **antiviral drugs and vaccines that target smallpox** can be used to treat Mpox.

Avoiding Mpox

- **Avoid skin-to-skin contact** with people who have a rash that looks like Mpox.
- **Avoid touching things** that a person with Mpox has used.
- **Wash your hands often** with soap and water or use an alcohol-based hand sanitizer, especially before eating or touching your face and after you use the bathroom.

Vaccination Against Mpox

According to the **CDC's recommendations**:

- **Vaccination** is an important tool in stopping the spread of Mpox.
- **JYNNEOS** is a 2-dose vaccine developed to protect against Mpox and smallpox for **adults 18 years and older**.
- Individuals with immune deficiencies **can receive the JYNNEOS vaccine**, which contain the live virus that cannot replicate.
- Individuals who had a severe allergic reactions with JYNNEOS should not receive this vaccine again.

On Sept 13th, 2024, the WHO prequalified the MVA-BN vaccine, which contains the live MVA virus that cannot replicate. This vaccine can help protect against Mpox and should be available in the near future.

Trusted Information About Mpox

- Mpox – World Health Organization (WHO)
- Clade I Mpox Outbreak Originating in Central Africa – Centers for Disease Control and Prevention (CDC)
- JYNNEOS vaccine – US Food and Drug Administration (FDA)

Disclaimer: The information is provided for educational purposes only. It is not intended to be taken as medical advice. The needs for everyone are different, and not all information is applicable to everyone. If you have questions or concerns, please discuss them with your doctor or healthcare provider.

22q11.2DS Featured in Dutch Medical Journal

Dutch title: **Het 22q11.2-deletie-syndroom: onbekend maakt onderbehandeld**

English translation: **22q11.2DS: under-recognized and under-treated**

22q11.2 deletion syndrome (22q11.2DS) is not a well-known condition, even among medical professionals. When doctors do not know enough of the condition, patients may not receive the necessary assessments and care.

A group of doctors in the Netherlands were determined to raise awareness for 22q11.2DS among medical professionals. They published their article in Medisch Contact, a Dutch journal that reaches almost 50.000 medical doctors. Their article was also featured on the cover of the journal (bottom center in the image on the right).

The authors highlight some of the main problems. Each affected individual is unique, and the diverse range of features make it difficult for doctors to realize the genetic cause. Medical professionals often only focus on the problems that are within their specialties, and they may not collaborate with other specialists to provide coordinated care as a team for the patient. Moreover, there are not nearly enough clinics to provide specialized care for affected individuals. Once patients transition into adulthood, their medical care often becomes even more fragmented.

In addition, the authors presented the most common features of the syndrome and the stories of some affected individuals. They also provided the links to both sets of international clinical practice recommendations (for children and adults) for the readers.

The article about 22q11.2DS was highlighted on the front cover (bottom center) of the May 30th, 2024 edition of Medisch Contact.

The article is available online for free. Some browsers can automatically translate the Dutch text into English.

The 22q11.2 experts who wrote this article:

- Dr Erik Boot, physician specialized in intellectual disability medicine
- Dr. Lenneke van Hooijdonk, medical biologist on behalf of the patient association Stichting Steun 22q11
- Dr. Michiel Houben, pediatrician
- Prof. Dr. Thérèse van Amelsvoort, professor of transitional psychiatry

The lead author of this article, **Dr. Erik Boot**, is a trustee of the 22q11.2 Society and the lead author of the clinical recommendations document for adults with 22q11.2DS. The chair of the patient association Stichting Steun 22q11, **Kim van Bekkum**, who was quoted in the article, was the recipient of the Unsung Hero Award at The 13th Biennial International 22q11.2 Conference in July, 2024 in Portugal.

Save the Date for 22q & Boo!

An educational symposium and Halloween party for children

Join the 22q and You Center at Children's Hospital of Philadelphia (CHOP) for some Halloween fun on Sunday, Oct. 27!

Activities for this year's in-person event will include arts & crafts, games, and refreshments, and an educational component.

Costumes are strongly encouraged!

https://www.chop.edu/events/22q-boo

Sunday Oct 27, 2024
12 p.m. - 3 p.m. (EDT)

Children's Hospital of Philadelphia
Abramson Pediatric Research Center
3615 Civic Center Blvd, Philadelphia, PA 19104

Questions?
Please email **22q@chop.edu**.

Faces of Sunshine Calendar 2025

The **2025 Faces of Sunshine Calendar** is now available for pre-order! These calendars can be a great gift for teachers, bus drivers, professionals and family members who help our loved ones along this journey in raising awareness for the 22q11.2 deletion and duplication syndromes. All proceeds benefit the International 22q11.2 Foundation, Inc. so please feel free to order early & often!! Shipping will be begin the first week of December via USPS Priority Mail.

Pre-order your calendar

Photo Submission

To enter an individual in the calendar:

- Email a maximum of two **high quality unedited** photo(s) to **info@22q.org by November 1st, 2024**. **AND**
- Sign the **consent** form online.

The Foundation will make every effort to include each child or adult somewhere in the calendar. This is subject to the number of submissions and space available.

Buy a Birthday

You can also celebrate your loved one's special day by **buying a birthday by November 1st**. When you buy a birthday, the Foundation will include an acknowledgment (first name only) and a photo of your child or adult on their birthdate on the 2025 **Faces of Sunshine** calendar. The placement of a person's photo on a specific day happens on a first come first serve basis. If multiple individuals share a birthday, the Foundation will work with the families to find a suitable arrangement.

Donate Now

Our mission: to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.

International 22q11.2 Foundation

What would we do with the funds:

- Support Research
- Support family conferences
- Support awareness
- Support Newborn Screening
- Support and raise awareness for 22q!

Your support makes a difference!

There are so many options for **Team 22q Fundraising**! Take part in one of the Foundation's event or create your own.

Please visit the **Donate** page on our website for information on donating online, by mail, or via other methods.

Please also **Shop** on our webstore.

For more information, please visit our website at **www.22q.org** or email us at **info@22q.org**.

Team 22q Fundraising

- **Tell 22 Friends** about 22q
- Become a **monthly donor**
- Be a 22q Citizen Journalist and share your event on social media!
- Plan a **22q at the Zoo** event
- Plan a **22K for 22q** event
- (It can also be 2.2K!)
- Create Your Own Event or Fundraiser Contact us at **info@22q.org**

Thank you!

Connect With Us

The mission of the **International 22q11.2 Foundation** is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.
This information is brought to you by the Foundation for educational purposes only. It is not intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.

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International 22q11.2 Foundation, Inc.
PO Box 532, Matawan, NJ 07747
www.22q.org (877) 739-1849 **info@22q.org**