



International  
22q11.2 Foundation

# Newsletter December 2022

Greetings! As we come to the close of 2022, we want to thank you – our 22q families, friends, supporters and professionals – from the bottom of our collective hearts for your continuing support of the International 22q11.2 Foundation.



## Announcing: The International 22q11.2 Foundation's 17<sup>th</sup> Annual Faces of Sunshine Calendar

On behalf of the International 22q11.2 Foundation, Inc., it is our pleasure to announce the featured children for each month of our **2023 Calendar**. This year was particularly challenging as so many wonderful photos were submitted, but we have managed to include all eligible entries throughout the calendar.

Many thanks to all who submitted photos; we very much hope you enjoy the calendar as well as all of the beautiful faces of sunshine throughout the New Year!

This calendar is a great way to show your 22q pride. Additionally, this item makes a lovely holiday gift; so order now to ensure all orders can be filled.

Calendars may be [ordered via our website](#). All proceeds benefit the work of the International 22q11.2 Foundation, Inc. **Calendars will ship beginning December 15<sup>th</sup>.**

Regards,  
International 22q11.2 Foundation, Inc



- January** – Jasmine J from Philadelphia, Pennsylvania
  - February** – Weston S from Greenfield, Indiana
  - March** – Davis G from Calhoun, Georgia
  - April** – Madelyn G from Chelmsford, Massachusetts
  - May** – Julie K from Alamogordo, New Mexico
  - June** – Mateo V from Highspire, Pennsylvania
  - July** – Charlotte K from Charlton, Australia
  - August** – Peyton E from Downingtown, Pennsylvania
  - September** – Saralyn W from Lake Lynn, Pennsylvania
  - October** – Emerson E from Oconomowoc, Wisconsin
  - November** – Olivia W from Lithopolis, Ohio
  - December** – Hannah C from Sparta, Kentucky
- ...and the **2023 Cover** Goes to  
Myla K from Perryopolis, Pennsylvania

# Coming Soon:

## Updated Clinical Guidelines for 22q

To help professionals care for individuals affected by 22q and their families, 22q experts, led by [The 22q11.2 Society](#), have worked tirelessly in the past few years to update the clinical guidelines for both children and adults.

We are excited to let you know that the two sets of updated guidelines have been **accepted for publication** by the journal, *Genetics in Medicine*!

### Updated clinical practice recommendations for managing children with 22q11.2 deletion syndrome

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

### Updated clinical practice recommendations for managing adults with 22q11.2 deletion syndrome

Erik Boot, Sólveig Óskarsdóttir, Joanne C.Y. Loo, T. Blaine Crowley, Ani Orchanian-Cheff, Danielle M. Andrade, Jill M. Arganbright, René M. Castelein, Christine Cserti-Gazdewich, Steven de Reuver, Ania M. Fiksinski, Gunilla Klingberg, Anthony E. Lang, Maria R. Mascarenhas, Edward M. Moss, Beata Anna Nowakowska, Erwin Oechslin, Lisa Palmer, Gabriela M. Repetto, Nikolai Gil D. Reyes, Maude Schneider, Candice Silversides<sup>22</sup>, Kathleen E. Sullivan, Ann Swillen, Therese A.M.J. van Amelsvoort, Jason P. Van Batavia, Claudia Vingerhoets, Donna M. McDonald-McGinn, and Anne S. Bassett

## Topics Covered

- Genetics
- Prenatal
- Cardiology (Heart) and Respiriology (Breathing)
- Ear-Nose-Throat / Palate / Speech
- Ophthalmology (Eye)
- Odontology (Teeth)
- Endocrinology (Hormones)
- Metabolism
- Growth
- Sensory Deficits
- Gastroenterology (Digestive System)
- Nutrition
- Genitourinary
- Gynecology (Women's Health)
- Sexual and Reproductive Health
- Obstetrics (Giving Birth)
- Immunology
- Hematology (Blood) / Oncology (Cancer)
- Skeletal (Bones and Muscles)
- Neurology (Nerves and the Brain)
- General Surgery
- Sleep
- Cognitive Functioning and Development
- Adaptive Functioning
- Psychiatry (Mental Health)
- Aging

# A Big Thank You!

The intensive process of updating the guidelines involved expert 22q healthcare providers from 10 countries, covering over 20 subspecialties. The leaders from 8 patient advocacy organizations, representing over 7000 families, also provided their input through a survey.

The International 22q11.2 Foundation would like to thank everyone who contributed to the project.

## **The 22q11.2 Society and all authors**

### **Key Leaders**

Sólveig Óskarsdóttir, Erik Boot, Anne S. Bassett, Donna M. McDonald-McGinn

### **External Reviewers**

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## **Patient Advocacy Groups**

[International 22q11.2 Foundation](#)

[22q Foundation Australia & New Zealand](#)

[22q11 Ireland](#)

[Max Appeal, The United Kingdom](#)

[Aidel 22, Italy](#)

[Stichting Steun 22q11, The Netherlands](#)

[22q11 Denmark](#)

[22q11 Europe](#)

## **Resource Curators**

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Once the two sets of updated guidelines are published,  
we will provide you with more information.

# Donna M. McDonald-McGinn receives the 2022 NSGCares Community Engagement Award



Donna and her team plan many events for the 22q Community. Here are photos from **22q and Boo** at CHOP.



The International 22q11.2 Foundation is proud to announce that our very own Founding Board Member, **Donna McDonald-McGinn**, MS, LCGC, received the National Society of Genetic Counselors 2022 NSGCares Community Engagement Award. NSGC released this video:

[https://youtu.be/50im\\_YSQCzE](https://youtu.be/50im_YSQCzE)

Donna was nominated for the award by fellow genetic counsellors Kathleen Valverde and Margaret Harr. The award ceremony took place in Nashville TN, during the 41<sup>st</sup> Annual Conference of the [National Society of Genetic Counselors](#), from November 16 to 20, 2022. Donna received the award from her son, Daniel McGinn, a Genetic Counseling Assistant at Children's Hospital of Philadelphia (CHOP).

Our Foundation would like to acknowledge Prof. McDonald-McGinn's longstanding and exemplary commitment to support patients and families affected by chromosome 22q11.2 differences as well as the larger 22q scientific and clinical community.

Sending huge congratulations and best wishes to Donna now and in the future!

# Palate Series

Information sheets now available on our website

<https://22q.org/symptoms-care/health-conditions-explained/>

Medical terms can be overwhelming, especially for families who are affected by multiple health conditions. To help our patients and families navigate through their medical journeys, we started a page called “[Health Conditions Explained](#)” on 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.

Since we began this series, we have published the Heart Series (02/2022), the Dental Series (05/2022), and the First Edition of the 22q Glossary (08/2022). We are proud to present the latest addition: the **Palate Series**! Published in 12/2022, this series of 6 information sheet introduces the velopharynx (VP; soft palate + throat) and the management of VP problems common among children with 22q11.2 deletion syndrome.

## PALATE SERIES

[The Velopharynx](#)

[Cleft Palate & Submucous Cleft Palate](#)

[Velopharyngeal Dysfunction: Introduction and Causes](#)

[Velopharyngeal Dysfunction: Diagnosis](#)

[Velopharyngeal Dysfunction: Surgery](#)

[Velopharyngeal Dysfunction: Speech Issues](#)

## DENTAL SERIES

[Let's Talk 22q Teeth – Info for Families](#)

[Dental Health in Children with 22q – Info for Dentists](#)

## HEART SERIES

[The Heart and Normal Blood Flow](#)

[Ventricular Septal Defect \(VSD\)](#)

[Tetralogy of Fallot \(ToF\)](#)

[Truncus Arteriosus \(TA\)](#)

[Interrupted Aortic Arch \(IAA\)](#)

Palate Series for Individuals with 22q11.2 Differences

### Velopharyngeal Dysfunction: Diagnosis (Continued)


**Video Fluoroscopy**

- The radiologist will place barium into the child's nose to highlight the velopharyngeal mechanism during X-ray.
- The child must imitate a short standard speech sample while the radiologist takes a video x-ray of their palate.
- The child must be able to talk, and the words must include oral pressure consonants (P, B, T, etc.)
- Crying or only making M and N sound will not provide a proper diagnosis.

**Nasopharyngoscopy**

- In some medical centers, the child may get a spray of medicine to numb the nose.
- The surgeon or speech pathologist puts a thin, flexible fiberoptic camera through the nose to the back of the throat.
- Once the camera is inserted to view the VP valve, the speech pathologist asks the child to produce a speech sample (which must include oral consonants (such as P, B, T, F, S, K)).

**Analysis of the VP Imaging Results (During Speech)**



**Normal closure**  
There is an airtight seal for pressure sounds. The soft palate, the back & sides of the throat, come together to squeeze down and close off the nose from the mouth.

**Velopharyngeal dysfunction**  
When the child talks, complete closure is not achieved. Also, in at least 20% of children with 22q11.2DS, pulsations may be present at the back of the throat, due to a displaced carotid artery.

**VP imaging results are necessary for VPD diagnosis and surgical planning.**

For more info, see the video [22q11.2 VPD AND HYPERNASAL SPEECH](#) from the 22q Family Foundation.

**22q**  
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. The 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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www.22q.org

Click the link below to access the first edition of the glossary (87 entries)

**22q GLOSSARY**

We will add more contents on various topics as they become available.

# Facts about 22q11.2 Differences

## 22q11.2 Deletion Syndrome

- The person is missing piece of chromosome 22. He/she has 1 copy (rather than 2) of some of the genes that direct how the body is formed and works.
- Happens in about 1 in 2148 babies
- There is a wide variability of clinical features. Each person (even members of the same family) is affected differently.
- Usually happens as a new event in the family
- There is a 50% chance of passing the deletion on in each pregnancy.
- People may be undiagnosed for many years, even into adulthood.

### Common Features

- Birth defects (e.g. heart, palate, & kidney problems)
- Low calcium
- Difficulty fighting infections
- Feeding and swallowing differences
- Seizures
- Developmental delay - particularly in speech
- Learning differences
- Behavioral problems
- Other differences

## 22q11.2 Duplication Syndrome

- The person has an extra piece of chromosome 22. He/she has 3 copies (rather than 2) of some of the genes that direct how the body is formed and works.
- Happens in about 1 in 850 pregnancies
- There is a wide variability of clinical features. Each person (even members of the same family) is affected differently.
- Often familial and very mild
- There is a 50% chance of passing the duplication on in each pregnancy.
- People may be undiagnosed for many years, even into adulthood

### Common Features

- Birth defects (e.g. heart, palate)
- Vision and ear problems
- Difficulty fighting infection
- Developmental delay
- Learning problems
- Autism
- ADHD
- Other differences

## Let's meet at the zoo!

The 13<sup>th</sup> Annual 22q at the Zoo Worldwide Awareness Day® will be held on **May 21<sup>st</sup>, 2023 (Sunday)**. This event aims to raise awareness about 22q chromosomal differences. It also gives families, friends, and professionals a chance to socialize and network. More information will be available in 2023.



# We Lit up the night for 22q!

**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 or 11/22) as a play on the name of the chromosome difference. Here are some photos from November 22<sup>nd</sup>, 2022 – Even the Niagara Falls was lit in red!



A private residence, NJ, USA



Spazio Cultura, Italy



Confederation Building, St. John's, NL, Canada  
(Photo: Laura Pottle-Hayley)



The Niagara Falls, ON, Canada  
(Photo: Earthcam)



CN Tower, Toronto, ON, Canada



Juelerye, NC, USA



Children's Hospital of Philadelphia  
Philadelphia, PA, USA

# Finding My Motivation with 22q

## The inspirational story of Tom Chase

"I choose not to let my issues define me," says Tom who just turned 40 years old and was not diagnosed with 22q until the age of 28. Life for Tom has not always been easy, but read on to find out how the will to succeed can help you to accomplish many things!

Tom Chase was born in Boscawen, NH, and talks about his childhood being different from others, "I had delays in most everything; walking, speaking, etc." His first 5 years of life were spent in and out of hospitals. With several learning differences, anxiety, difficulty reading and taking tests he required constant help with school work. Since at the time he had no diagnosis, the administration at school told his family that he was "coddled" too much and "lazy". Thankful for the frequent trips his parents were required to make to school for IEP and other meetings, Tom says, "I owe so much to my family, particularly my mom staying up late with homework keeping me busy with activities and making sure the schools did the right thing by me! And my dad was a huge help too in working and supporting the family."

Growing up without any type of diagnoses Tom found it hard to make friends; they couldn't understand his issues and of course, he was not able to explain. He did find a few friends he could trust and participated in activities best he could but most sports were difficult because of bad ankles. During middle school, he was bullied daily, physically and emotionally. Thus he disappeared in reading. This was until he discovered riding a bike. "I fell in love with biking I was on the road by myself miles on end, long trips short ones, didn't matter." High school wasn't any better academically or socially. Tom's mom signed him up for a swim team and shortly after he also started lifting weights both of which he loved. In his last year of high school, he got really sick and missed a lot of school but when told he wasn't going to graduate, what did Tom do? He buckled down, missed all senior-related activities and graduated on schedule. After high school, he entered the workforce, "I chose not to let my issues define me if I can't do the job it's not for me."

In 2000, Tom began martial arts training giving him self-discipline and confidence; this eventually led to him teaching Tae Bo and spin classes! But at age 28 Tom suffered an injury to his shoulder at work which required surgery. The injury activated many other issues including back and nerve pain leading his doctor to further review his medical history, he ran more tests and finally came the diagnosis, 22q. "This was a curse and blessing," he says, "I can now explain things but it's too late to receive help; funding, counseling, etc." After the accident, Tom had many health-related difficulties and gained a lot of weight, in fact at one time he weighed 340 pounds. Just as doctors considered putting him in a wheelchair, his self-discipline kicked in and he took the matter back into his own hands by working out and eating healthy. Today he is just over 200 pounds, works out 6 days a week and posts videos of his beach body workouts for inspiration to others. Tom is a hard worker, working 40 hours a week as a custodian, "A good union paying job with benefits!" Happily married, for 15 years he and his wife recently purchased their first home and continue making plans for the future. Tom and his wife both have online businesses since his wife also suffers from health issues, Neurofibromatosis, they realize that one-day full-time employment outside of the home may not be an option. Tom's self-discipline, work ethic and motivation is an inspiration to all!

### **What is Tom's advice for others living with 22q?**

For parents of children with 22q: "I would highly encourage parents to research and get their kids early help; grants, physical therapy and speech therapy."

For children with 22q: "Functional fitness is huge, don't go into hiding as I did. Be courageous, make friends lots of them. Enjoy growing up."

For adults with 22q: "Don't let 22q define you, you are strong and can do anything with the right support and training."

**Thank you, Tom, for sharing your story and encouraging so many others!**



# We appreciate your financial support!

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

At the International 22q11.2 Foundation, we help families that need important resources and information to meet the needs of loved ones with 22q. We work with top medical experts from around the world to build our understanding of 22q, improve treatment options for families, and conduct the necessary research for longer, healthier lives for patients.

## Your support makes a difference!

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](http://www.22q.org) or email us at [info@22q.org](mailto:info@22q.org). **Thank you!**



### 22q Fundraising Toolkit

Become a [monthly donor](#)

Plan a [22q at the Zoo](#) event

[Tell 22 Friends](#) about 22q

Plan a [22K for 22q](#) event.  
It can also be 2.2K!

Select 22q as your  
Charity of Choice on [AmazonSmile](#)

Create Your Own Event or Fundraiser  
Contact us at [info@22q.org](mailto:info@22q.org)

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Please visit the [leadership](#) page on our website for more information about the directors

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