International 22q11.2 Foundation

# Newsletter – December 2025


Dear Friends,

As we come to the close of another year, we pause with gratitude and reflection—grateful for the strength of our community and hopeful for what lies ahead.

For 22 years, the International 22q11.2 Foundation has stood alongside individuals and families affected by chromosome 22q differences, ensuring that no one faces this journey alone. What began in 2003 has grown into a global network of support, education, and connection—made possible by people like you.

Throughout this year, your support has helped us provide trusted resources through 22q.org, expand educational and scientific programming, increase awareness worldwide, and foster meaningful connections among families, clinicians, and researchers. Together, we continue working toward a future where individuals affected by 22q differences can thrive.

As 2025 comes to an end, we respectfully ask you to consider making a year-end, tax-deductible gift to the International 22q11.2 Foundation. Your generosity directly supports families navigating complex medical, educational, and emotional challenges—and helps us remain the “go-to” resource for everything 22q.

Every contribution, no matter the size, makes a meaningful difference. With your help, we can continue advancing awareness, supporting families, and improving the quality of life for those impacted by 22q differences—today and for generations to come.

Thank you for your compassion, trust, and continued partnership. We wish you and your loved ones a joyful holiday season and a happy, healthy new year.

With heartfelt gratitude,

**The International 22q11.2 Foundation, Inc.**

*Together, we are making a difference.*

Marc Weinberg, Chair

On behalf of the
Board of Directors,
The International 22q11.2 Foundation

# Faces of Sunshine Calendar 2026

## Announcing the featured individuals of our 20th calendar


On behalf of the International 22q11.2 Foundation, Inc., it is our pleasure to announce the featured individuals for each month of our 2026 "Faces of Sunshine" Calendar. Each year is particularly challenging as so many wonderful photos are submitted, we have also 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 support for 22q. Show your pride and help spread awareness with the 2026 Faces of Sunshine Calendar. Calendars may be ordered via our website. All proceeds benefit the work of the International 22q11.2 Foundation, Inc.

**January** – **Ruby C**, from Somerset, Kentucky
**February** – **Conor M**, from Fulton, New York
**March** – **Zion D**, from Philadelphia, Pennsylvania
**April** – **Brycen B**, from Mogadore, Ohio
**May** – **Jayla M**, from Lordsburg, New Mexico
**June** – **Dougie B**, from Coal City, Illinois
**July** – **Lily A**, from Greer, South Carolina
**August** – **Collins H**, from Brecksville, Ohio
**September** – **Leo B**, from Grove City, Ohio
**October** – **Michael C**, from Wausau, Wisconsin
**November** – **Jaxson P**, from Hudson, North Carolina
**December** – **Evie D**, from Princeton, North Carolina

…and the **2026 Cover** Goes to:

**Sadie W**, from Binghamton, New York

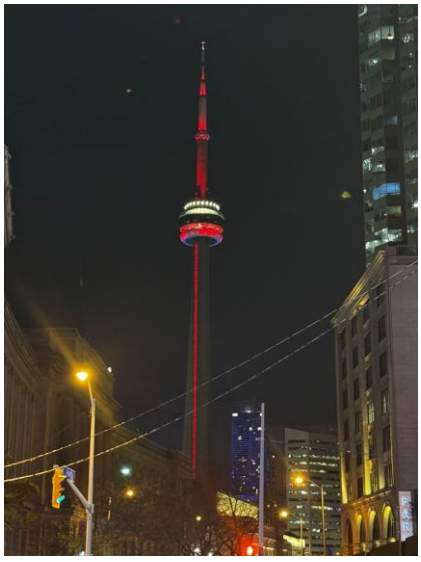
# We Lit Up the Night for 22q!

To increase public awareness of 22q differences, 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 in North America) as a play on the name of the chromosome difference.

Here are the photos of some of the landmarks that were lit up on November 22nd, 2025. For more photos, please check out our **Facebook** and **Instagram**.

Ben Franklin Bridge – Philadelphia, Pennsylvania and Camden, New Jersey, USA
(Instagram – drpabridges (Delaware River Port Authority))

Niagara Falls – Ontario, Canada and New York, USA
(Earthcam)



CN Tower –
Toronto, Ontario, Canada

# We Lit Up the Night for 22q! (Continued)

Town Hall – Oviedo, Spain

City Hall, Santander, Spain

San Mamés Stadium – Bilbao, Spain

Torre de Mangana – Cuenca, Spain

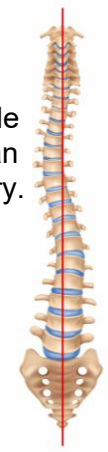
## Activities That Also Took Place During 22q Awareness Month


22q11 Europe's conference brought families, clinicians, scientists, and researchers together to share knowledge and expertise about 22q11 syndrome and foster meaningful connections.


Our Foundation actively posted on social media to spread awareness. We also posted resources for our families.

Recent research findings on…

# Scoliosis in Young People with 22q11.2DS



The spine is our backbone. It has a natural front-to-back curve.

**Scoliosis** is the **abnormal side-to-side curve of the spine**. It happens in about half of the people with 22q11.2 deletions. Mild curving does not usually lead to big problems, but severe curving can cause back pain and bad posture. Treatment for scoliosis includes exercises, braces, and surgery.

## Measuring…

### How bad the curve is

- The side-to-side curve of the spine can be seen on x-ray images
- Doctors determine where the bending starts and ends. They draw lines from those spots and determine the **Cobb angle**.
- If the Cobb angle is at least 10°, the person has scoliosis.
- **The higher the Cobb angle, the worse the scoliosis curve.**

### How mature the bones are

- Bones form and harden layer by layer at growth plates (ends of the bone).
- On x-ray images, growth plates look like dark lines.
- The upper-most growth plate on the pelvis is the site that is x-rayed to check for bone maturity. The bone formation progress here gets a score called the **Risser Grade** (0-5).
- There are variations of the Risser Grades (Europe, North America, Risser+)
- **Lower Risser Grades mean ongoing bone growth. There is a higher risk of scoliosis getting worse.**
- Higher Risser Grades mean bone growth is almost complete. There is a lower risk of scoliosis progression.

Graphics from Freepik.com

Researchers in the Netherlands and Italy studied scoliosis in young people who have 22q11.2DS. They wanted to find out:

- How scoliosis starts and continues;
- What makes scoliosis get worse or better;
- When the curve gets worse most quickly;
- What predicts the final curve extent.

In one study (1), the researchers checked the medical records of 161 patients from the time when their bones were still immature (Risser Grade 0 (see box on left)) onwards for at least 2 years. They found that:

- In 19/161 (12%) patients, the Cobb angle (see box on left) got worse to >30°. For others, it did not change much or never exceeded 29°.
- Scoliosis was more likely to get worse in girls than boys.
- The **scoliosis curve got worse very quickly** when the **triradiate cartilage at the pelvis closes** (as part of normal bone formation). This closure marks the **beginning of the puberty growth spurt**.
- No factors could predict how the scoliosis curve would progress.

In another study (2), doctors examined 292 patients at least once every other year and reviewed their 722 x-rays. They found that:

- At the most recent follow up, 103/292 (35%) had a Cobb angle between 10° and 30°, and 13/292 (6.5%) had one that was >30°. Some patients in the latter group were treated with braces (3/13) and surgeries (7/13).
- 21/292 (7.2%) children had scoliosis that went away without treatment.
- Of the 175 children who had x-rays before 10 years of age, 85 (49%) already had scoliosis at that point.
- Scoliosis worsens most quickly in immature bones (Risser Grades 0-1).
- **Children under 10 years old whose spine curvature changed back and forth a lot were more likely to develop scoliosis later.**

It is recommended that children with 22q11.2 receive **routine scoliosis screening from age 6 years onwards**, with **x-rays every 2 years** until their bones reach maturity. This allows for early detection and treatment.

### Recent Research Reports on Scoliosis and 22q11.2DS


1. What can we learn from scoliosis in children with the 22q11.2 deletion syndrome? Prognostic factors at pre-adolescent age for fast progressive, mild and self-resolving forms during adolescence.
   Donzelli S, Lafranca P, Van Smeden M, Castelein R, Schlösser T.
   Spine Deform. 2025 Jul;13(4):1179-1187. doi: 10.1007/s43390-025-01073-4. Epub 2025 Mar 20.
   Erratum in: Spine Deform. 2025 Aug 27. doi: 10.1007/s43390-025-01155-3.
2. Natural history study of scoliosis in patients with 22q11.2 deletion syndrome, starting before disease onset.
   Lafranca PPG, de Reuver S, Abdi A, Houben ML, Kruyt MC, Ito K, Castelein RM, Schlösser TPC.
   Spine Deform. 2025 Oct 5. doi: 10.1007/s43390-025-01193-x.

# Facts about 22q11.2DS and 22q11.2DupS

## 22q11.2 deletion syndrome (22q11.2DS)

1. A deletion in chromosome 22q11.2 results in the **loss of 1 of 2 copies** of ~50 genes that direct how the body is formed and may function.
2. It happens in about 1 in 2148 babies, but not many people know about it.
3. It used to be known by other names such as DiGeorge syndrome & velocardiofacial syndrome.
4. Most children with a 22q11.2 deletion have differences in the heart and palate. They may also have difficulty fighting infection, feeding and swallowing, and maintaining normal calcium and thyroid levels.
5. Adults with a 22q11.2 deletion often have congenital heart conditions that require monitoring, low calcium, obesity, diabetes, cognitive deficits, and psychiatric illness.
6. Each person (even members within the same family) is affected differently in terms of symptoms and severities.
7. Usually arises as a new event in a family. Only about 10% of patients have a parent who is also affected.
8. Once a person has the deletion, he/she has a 50% chance of passing it onto the next generation at each pregnancy.

## 22q11.2 duplication syndrome (22q11.2DupS)

1. A duplication at chromosome 22q11.2 results in 3 copies (rather than 2) of about 50 genes that direct how the body is formed and may function.
2. It happens in about 1 in 850 pregnancies, but not many people know about it.
3. Much less is known about the duplication syndrome compared the deletion syndrome.
4. Features of 22q11.2 duplication syndrome can include medical differences of the heart and the palate, learning and developmental delay, autism spectrum disorder, vision and ear problems, and other medical differences.
5. 22q11.2DupS is frequently identified in children with developmental delay or behavioral differences.
6. Each person (even members within the same family) is affected differently in terms of symptoms and severities.
7. 22q11.2DupS usually runs in the family. About 60%-70% of patients has a parent with the duplication.
8. Once a person has the duplication, he/she has a 50% chance of passing it onto the next generation at each pregnancy.

**To find out more, please visit www.22q.org.**

# Organs and Systems Involved in 22q Differences

22q11.2 deletion syndrome (22q11.2DS) tends to involve the organs and body systems shown in the following diagrams. 22q11.2 duplication syndrome (22q11.2DupS) involve similar ones but at lower frequencies.

For more information, see pages 6 and 8.

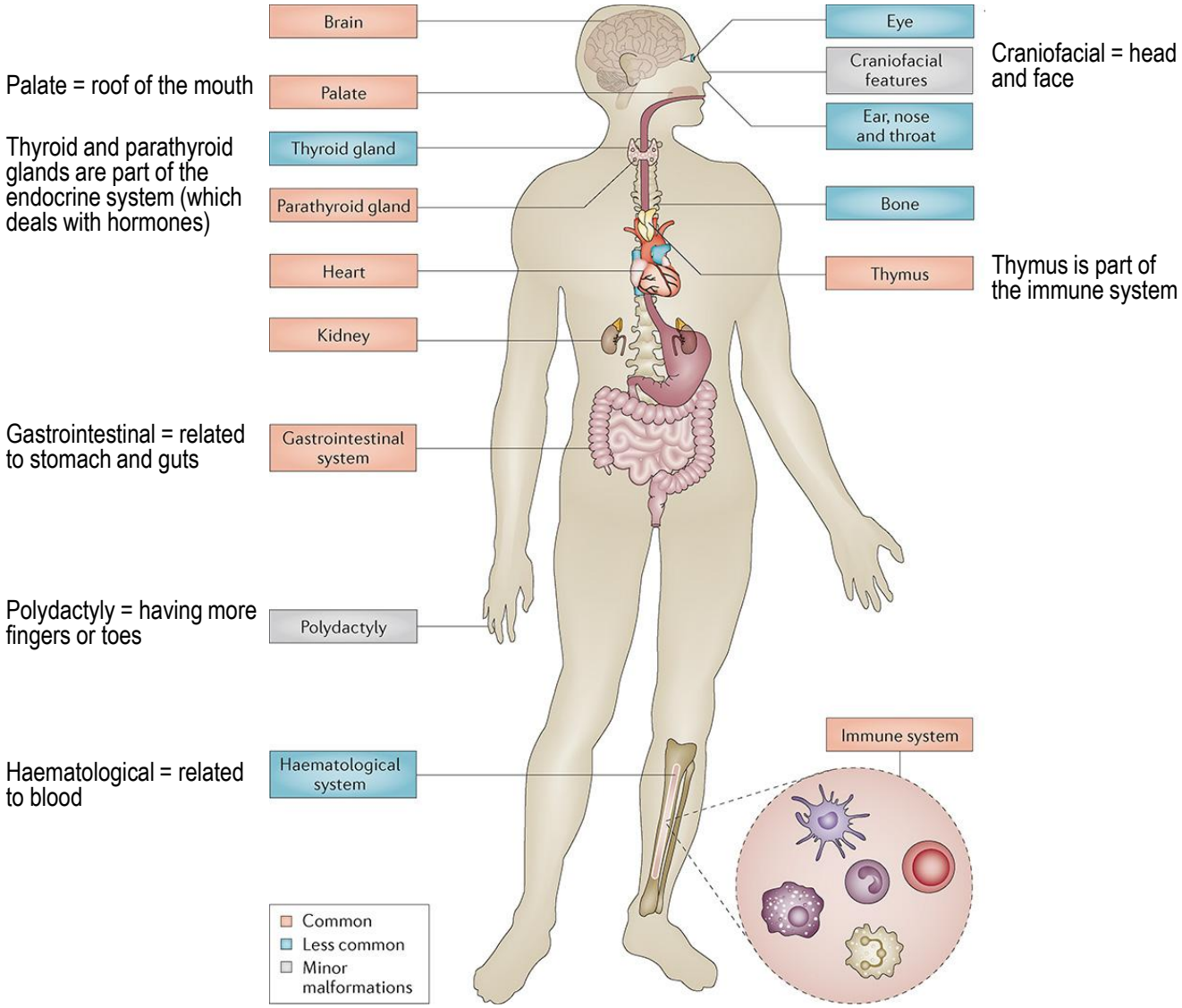



Image source: McDonald-McGinn DM, Sullivan KE, Marino B, Philip N, Swillen A, Vorstman JA, Zackai EH, Emanuel BS, Vermeesch JR, Morrow BE, Scambler PJ, Bassett AS. 22q11.2 deletion syndrome. Nat Rev Dis Primers. 2015 Nov 19;1:15071. doi: 10.1038/nrdp.2015.71. PMID: 27189754; PMCID: PMC4900471.

The article is available for free at: https://pmc.ncbi.nlm.nih.gov/articles/PMC4900471/

# Clinical Recommendations for 22q11.2 Deletion Syndrome

The recommendations for the care of individuals with 22q11.2 deletion syndrome (22q11.2DS or 22q) can be found in these documents:

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

- English (Original)
- French – Français
- Spanish – Español
- Simplified Chinese – 简体中文

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

- English (Original)
- French – Français
- Spanish – Español
- Traditional Chinese 繁體中文

**Speech-Language Disorders in 22q11.2 Deletion Syndrome: Best Practices for Diagnosis and Management**

- English (Original)

**Prenatal Screening and Diagnostic Considerations for 22q11.2 Microdeletions**

- English (Original)

# Information on 22q11.2 Duplication Syndrome

Expert healthcare providers are currently preparing a document that will provide detailed information on features associated with the chromosome 22q11.2 duplication syndrome (22q11.2DupS).

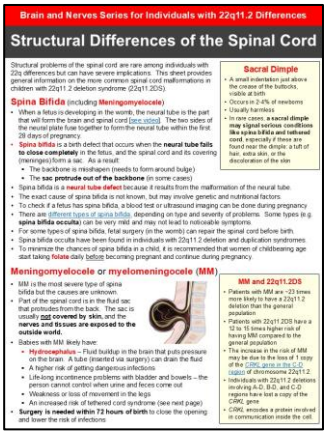
In the interim, these experts are using the same healthcare guidelines prepared for the 22q11.2 deletion syndrome, as the associated features are quite similar, just with lower frequency.

Our Foundation will be in touch as soon as the new paper for the 22q11.2 duplication syndrome is published. Thank you for your patience and understanding.

# Brain and Nerves Series – 9 Topics

We have now published all the topics for the **Brain and Nerves Series** on the "**Health Conditions Explained**" page of our website.

The nervous system is the command center of the body. It controls how our body responds to what's happening both inside and outside of the body. Some individuals with 22q differences (especially 22q11.2DS) experience complex health conditions that affect the brain and the nerves. Our Brain and Nerves Series provides background information on the common ones.


Brain and Nerves Series for Individuals with 22q11.2 Differences

Structural Differences of the Spinal Cord

Structural problems of the spinal cord are rare among individuals with 22q differences but can have severe implications. This sheet provides general information on the more common spinal cord malformations in children with 22q11.2 deletion syndrome (22q11.2DS).

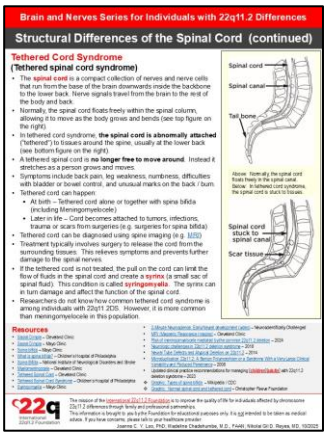
Spina Bifida (including Meningomyelocele)

Sacral Dimple

Meningomyelocele or myelomeningocele (MM)

MM and 22q11.2DS

Special thanks to Joanne Loo, Programmatic and Educational Tool Developer; Dr. Madeline Chadehumbe, Pediatric Neurologist; and Dr. Nikolai Gil D. Reyes, Neurologist.


Brain and Nerves Series for Individuals with 22q11.2 Differences

Structural Differences of the Spinal Cord (continued)

Tethered Cord Syndrome
(Tethered spinal cord syndrome)

Resources

- **Introduction to the Brain and the Nerves**
- **Seizures and Epilepsy**
- **Hypotonia**
- **Parkinsonism and Parkinson's Disease**
- **Other Movement Disorders** (info added 11/2025)
- **Structural Differences of the Brain** (new)
- **Structural Differences of the Spinal Cord** (new)
- **Nervous System Issues in 22q11.2 Deletion Syndrome** (new)
- **Nervous System Issues in 22q11.2 Duplication Syndrome** (new)

# 22q Glossary – Fourth edition now available!

To help families understand terms that they may encounter, we have been developing a glossary for 22q differences. We have just published the fourth edition, which contains **294** terms with explanations plus **32** additional terms with redirections ("See ___"). The latest version had a big increase in the number of terms related to the immune system, sleep, and the brain and the nerves, and is now available on the **Health Conditions Explained** section of our website. We will add more terms in future editions.

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

Click this link below to access the glossary

**22q GLOSSARY (4th Edition)**

Disclaimer: This information is brought to you by the International 22q11.2 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.

# Health Conditions Explained

Complete List of Topics Published as of December 2025

## HEART SERIES

- The Heart and Normal Blood Flow
- Ventricular Septal Defect (VSD)
- Tetralogy of Fallot (ToF)
- Truncus Arteriosus (TA)
- Interrupted Aortic Arch (IAA)

## DENTAL SERIES

- Let's Talk 22q Teeth – Info for Families
- Dental Health in Children with 22q – Info for Dentists

## PALATE SERIES

- The Velopharynx
- Cleft Palate & Submucous Cleft Palate
- Velopharyngeal Dysfunction: Introduction and Causes
- Velopharyngeal Dysfunction: Diagnosis
- Velopharyngeal Dysfunction: Surgery
- Velopharyngeal Dysfunction: Speech Issues

## GI SERIES

- GI Issues in 22q11.2 Deletion Syndrome
- The Digestive System
- Feeding Difficulties
- Swallowing and Dysphagia
- Constipation
- Gastroesophageal Reflux Disease (GERD)
- Esophageal Dysmotility
- Gastroparesis
- Cholelithiasis (Gallstones)
- Nausea and Vomiting
- Autoimmune Issues in the GI System
- Inguinal Hernias
- Nonalcohol Fatty Liver Disease (NAFLD)

## IMMUNE SYSTEM SERIES

- The Immune System
- Vaccination
- Immune Deficiencies Related to T Cells
- Immune Deficiencies Related to B Cells
- Autoimmune Disorders
- Allergies and Asthma
- Immune System Issues in 22q11.2DS
- Immune System Issues in 22q11.2DupS

## MENTAL HEALTH SERIES

- Mental Health and 22q11.2 Deletion Syndrome
- Mental Health and 22q11.2 Duplication Syndrome
- What Parents Can Do for Their Children
- Attention Deficit Hyperactive Disorder
- Autism Spectrum Disorder
- Anxiety Disorders
- Psychotic Disorders [Including schizophrenia]
- Mood Disorders
- Mental Health Q&A (Spring 2023)

## SPEECH SERIES

- 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

## SLEEP SERIES

- Introduction to Sleep
- Obstructive Sleep Apnea (OSA)
- Insomnia & Delayed Sleep Phase Disorder (DSPD)
- Restless Legs Syndrome (RLS) & Periodic Limb Movement Disorder (PLMD)
- Parasomnias

## BRAIN AND NERVES SERIES

- Introduction to the Brain and the Nerves
- Seizures and Epilepsy
- Hypotonia
- Parkinsonism and Parkinson's Disease
- Other Movement Disorders
- Structural Differences of the Brain
- Structural Differences of the Spinal Cord
- Nervous System Issues in 22q11.2 Deletion Syndrome
- Nervous System Issues in 22q11.2 Duplication Syndrome

## 22q GLOSSARY

- Fourth Edition

# The ACPA Randall/LaRossa College Scholarships

The Randall/LaRossa College Scholarship Fund was established in 2009 to honor Peter Randall, MD and Don LaRossa, MD from the Children's Hospital of Philadelphia (CHOP). Both of them were pioneers in palate repairs in children with 22q11.2 deletion syndrome.

The American Cleft Palate-Craniofacial Association (ACPA) awards four college scholarships annually to outstanding students born with cleft or craniofacial conditions. Please visit the scholarship website for more information and to submit an application. Spread the word to any eligible patients or students who would be interested in applying. The **deadline to apply is January 9th, 2026**.

Please click on the image on the right for a pdf version of the flyer.



# 22q at the Zoo Worldwide Awareness Day

## Sunday May 17th, 2026

Save the date for the 15th Annual 22q at the Zoo – Worldwide Awareness Day, which will take place on **Sunday, May 17th, 2026**. It is an opportunity for families, friends and professionals to socialize, network and raise public awareness of 22q11.2 syndromes.

See the 22q at the Zoo photos from:

**2025 | 2024 | 2023**

If there were no 22q at the Zoo events in your area in 2025, you can host one in 2025! Please visit the Host a 22q at the Zoo Event section on our website for more information. We even have an event planner letter ready for you! No zoo? No problem. Consider gathering at a park, a farm, or even an aquarium.

If you have any questions, please email us: info@22q.org. Thank you.

# 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. **Thank you**!

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Please visit the **leadership** page on our website for more information about the directors

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