International
22q11.2 Foundation

Newsletter September 2025

In 2025, the International 22q11.2 Foundation celebrates its 22nd anniversary!

To mark the occasion, we hosted a professional conference and a family meeting at the Children's Hospital of Philadelphia (CHOP) from September 4 to 6. We invited experts from multiple countries to share their expertise on 22q11.2 deletion and duplication syndromes. The topics ranged from basic science to conditions that affect different body systems. We also celebrated together at the "Around the World with 22q" gala at the Penn Museum on September 6. It was a wonderful event to celebrate our 22 years of commitment to the global 22q community. You will find photos from these events on pages 2-4 of this newsletter.

As September is back-to-school season, this newsletter includes tips and resources to help your child blossom at school (pages 5-6). At the same time, we are delighted to bring you the first five topics of the Brain and Nerves Series (page 7) and the latest research findings on movement disorders in adults with 22q11.2DS (page 8). You will also find information on upcoming events (pages 9-10) and the annual calendar campaign (page 11).

We hope you find the information in this newsletter useful. Together, we will continue our mission – to improve the quality of life for individuals affected by 22q differences through family and professional partnerships.

Thank you very much for your support. Here's to many more years of making a difference!

In this newsletter…

Celebrating our 22nd Anniversary – Recent events

Pathways to Understanding 22q11.2 Copy Number Variants

Scientific Summit – September 4, 2025

The World View on 22q

A Meeting for Families and Caregivers – September 5 to 6, 2025

Around the World with 22q!

Gala – September 6, 2025

Photos: Professional Conference and Family Meeting

Donna McDonald-McGinn, Anne Bassett, and Natalie Blagowidow

Donna McDonald-McGinn and Carol Cavana

Kathleen Sullivan, Donna McDonald-McGinn, Natalie Blagowidow, Maria Mascarenhas, and Anne Bassett

Parent participants

Donna McDonald-McGinn

Photos: Around the World with 22q Gala

Group photo at the gala

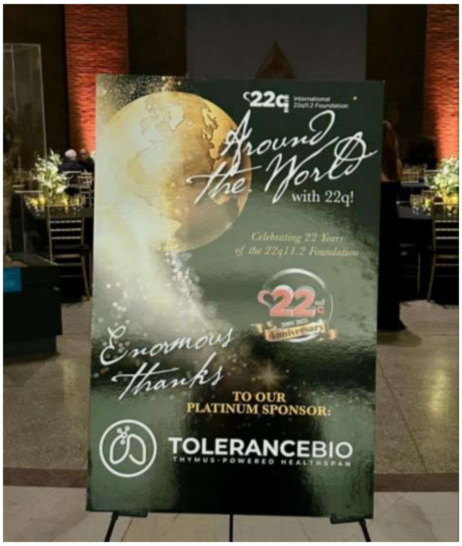
Foundation banner

Kate Baum, Madeline Chadehumbe, Raquel Gur, and Donna McDonald-McGinn

Madeline Chadehumbe and Kayla Disibio

Louis Cavana, Carol Cavana, and Michael Weinberg

Addison Milner and Madeline Chadehumbe



Gala banner

Photos: Around the World with 22q Gala (continued)

Madeline Chadehumbe, Debbie DeLoach,
Sam Mackenzie, and Deena Chisholm

Team Members of the 22q Clinic at the Children's Hospital of Philadelphia (CHOP)

Raul Alejandro Escobar and Hanadys Ale

Addison Milner and Rhonda Milner

Carmella Benigno, Carol Cavana,
Julie Wootton, and Laura Perez

Debbie DeLoach, Carol Cavana,
and Carmella Benigno

Francisco Leon and Louis Cavana

Items for auction
Upper Baugh Penn Museum , Philadelphia, PA

Thank you very much to everyone
who organized the meetings and the gala, who sponsored the events, who shared their expertise and experiences, and who celebrated with us.
Thank you for your continued support.

Helping Your Child Blossom at School

Advocating for the Services They Need

Most children (94%) with 22q11.2 deletion or duplication syndrome require special education support in the form of an Individualized Education Program (IEP). Here, we present some general information and share tips from other parents. Nevertheless, the situation in each school district is different, and we strongly recommend that parents connect with the school to discuss their children's needs, as early as possible.

Getting an Individualized Education Plan (IEP)

In the USA, an IEP is a written plan that includes **realistic goals** designed to allow children with disabilities to access a Free and Appropriate Public Education (FAPE). Crucial to this process, an IEP must include:

1. **Specific support services** that will enable the student with special needs to learn at a suitable pace, and
2. Clearly defined "**progress monitoring**" to periodically assess whether the student is in fact making adequate progress.

All children with 22q differences, learning difficulties, and/or intellectual disabilities benefit from an IEP if they are struggling in school. For more info about the referral and assessment processes, as well as the development of an IEP, check with your child's school district or with the Department of Education for your state.

Attending IEP Meetings

An IEP team usually includes the school nurse, a speech therapist, the classroom teacher, and others. The IEP process and meetings can be complex and intimidating. Nevertheless, remember that **you, as the parent, are an important member of the IEP team! You know your child best. Use your knowledge about your child to advocate for the services he/she needs to blossom.**

- Prepare **printed handouts** for each person in the IEP team. List your questions and concerns. This list will help the team **stay on task** and address the main points.
- If your child has a problem that involves only one subject, **meet that teacher individually** ahead of time. This strategy provides more time to solve that specific problem and saves other people's time during the actual IEP meeting.
- At the beginning of follow-up meetings, ask the IEP team how the support services and modifications are helping the child.
- Use a compliment sandwich (compliment-complaint-compliment) when dealing with sticking points. This strategy increases the chances that progress is made without someone being offended.
- If you feel that progress is not being made, reach out (or copy your email) to the school district's special education coordinator, who may be able to add more resources.

Your Role as the Parent

Parents of children with 22q differences often take on these roles:

- **Coordinator** – Plan the many medical appointments and any catch-up sessions for missed school work.
- **Accountant** – Keep a tab on medical expenses and seek out financial support (e.g. disability benefits)
- **Historian** – Record all medical info, e.g. weight, height, feeding habits, developmental milestones, changes in behavior, medications (and reactions), vaccinations, etc.
- **Researcher** – Use trusted sources to learn about the child's health conditions and how best to help him/her.
- **Advocate** – Ask for the services your child needs to blossom.

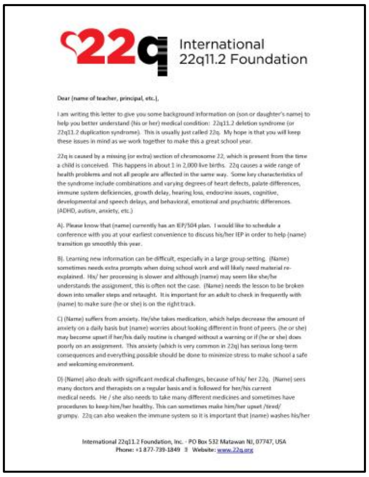
Parents are very important members of the child's medical and education teams. **You know your child best!**

Aspects of 22q differences that may affect a child's experience at school

- Intellectual disabilities
- Learning disabilities
- Memory problems
- Language and communication difficulties – Many children with 22q11.2DS are good at decoding words, but this ability masks their inability to understand the overall message.
- Attention deficit hyperactive disorder (ADHD)
- Autism spectrum disorder (ASD)
- Behavior differences
- Anxiety disorders / phobias
- Mood changes
- Lack of impulse control
- Poor social judgement leading to increased vulnerability (risk of bullying)
- Vision and hearing impairments
- Sleep disturbances
- Fatigue (physical and/or cognitive)
- Other medical aspects of 22q difference that may cause pain or distract the child
- Frequent absences from school due to medical appointments and hospital stays

Education-Related Resources

Sample Letter to Teacher (Editable Word Document)



International
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Dear (name of teacher, principal, etc.),

I am writing this letter to give you some background information on (son or daughter's name) to help you better understand (his or her) medical condition: 22q11.2 deletion syndrome (or 22q11.2 duplication syndrome). This is usually just called 22q. My hope is that you will keep these issues in mind as we work together to make this a great school year.

22q is caused by a missing (or extra) section of chromosome 22, which is present from the time a child is conceived. This happens in about 1 in 2,000 live births. 22q causes a wide range of health problems and not all people are affected in the same way. Some key characteristics of the syndrome include combinations and varying degrees of heart defects, palate differences, immune system deficiencies, growth delay, hearing loss, endocrine issues, cognitive, developmental and speech delays, and behavioral, emotional and psychiatric differences. (ADHD, autism, anxiety, etc.)

A). Please know that (name) currently has an IEP/504 plan. I would like to schedule a conference with you at your earliest convenience to discuss his/her IEP in order to help (name) transition go smoothly this year.

B). Learning new information can be difficult, especially in a large group setting. (Name) sometimes needs extra prompts when doing school work and will likely need material re-explained. His/ her processing is slower and although (name) may seem like she/he understands the assignment, this is often not the case. (Name) needs the lesson to be broken down into smaller steps and retaught. It is important for an adult to check in frequently with (name) to make sure (he or she) is on the right track.

C) (Name) suffers from anxiety. He/she takes medication, which helps decrease the amount of anxiety on a daily basis but (name) worries about looking different in front of peers. (he or she) may become upset if her/his daily routine is changed without a warning or if (he or she) does poorly on an assignment. This anxiety (which is very common in 22q) has serious long-term consequences and everything possible should be done to minimize stress to make school a safe and welcoming environment.

D) (Name) also deals with significant medical challenges, because of his/ her 22q. (Name) sees many doctors and therapists on a regular basis and is followed for her/his current medical needs. He / she also needs to take many different medicines and sometimes have procedures to keep him/her healthy. This can sometimes make him/her upset /tired/ grumpy. 22q can also weaken the immune system so it is important that (name) washes his/her

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hands often to help prevent exposure to germs.

E) (Name) also has difficulty with social skills and has trouble making and keeping friends. He/she may need some help with peer relationships or finding a partner for group activities. If there are any social groups or 'lunch bunch' groups at school, please make those resources available to (him or her).

Thank you, in advance, for taking time to read this short summary of some of (name) challenges. Much more detailed information is included in (his or her) file. However, I wanted to give you a brief introduction so the year can be off to a great start. I am committed to doing everything we can to make this school year a success, so please communicate with me frequently so that I can support you and my child at home. Thank you so much for all you do.

With Respect,

(Name)

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From the International 22q11.2 Foundation

- School and Learning
- Developmental Skills
- Behavioral and Mental Health Resources
- Health Conditions Explained and 22q Glossary

From the Dalglish Family 22q Clinic (Toronto, Canada)

- Information for Educators

From the 22q Family Foundation

- Student Success-A Handbook for Parents & Educators of Students with 22q11.2 Deletion Syndrome

Clinical Practice Recommendations for 22q11.2DS

These two documents are each available in 4 languages. The checklist for each one is available in English.

- Updated clinical practice recommendations for managing **children** with 22q11.2 deletion syndrome (other languages / checklist)
- Updated clinical practice recommendations for managing **adults** with 22q11.2 deletion syndrome (2023) (other languages / checklist)

The following document is available in English.

- **Speech-Language Disorders** in 22q11.2 Deletion Syndrome: Best Practices for Diagnosis and Management (2019)

Examples of Policies on Special Education

- USA: Disability Discrimination: Providing a Free Appropriate Public Education (FAPE)
- Ontario, Canada: Individual Education Plans

Brain and Nerves Series

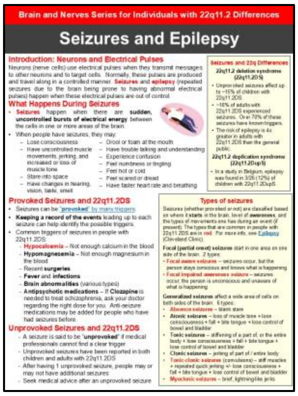
Five topics now available on our website

We have now published the first five topics of the **Brain and Nerves Series** in the "**Health Conditions Explained**" section 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.

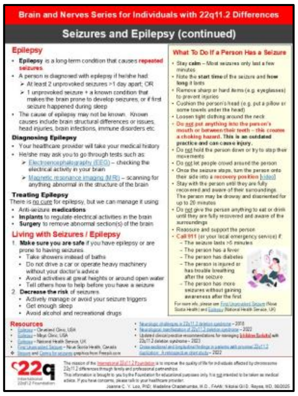
Special thanks to Joanne Loo, Programmatic and Educational Tool Developer; Dr. Madeline Chadehumbe, Pediatric Neurologist; and Dr. Nikolai Gil D. Reyes, Neurologist.

Brain and Nerves Series



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

Seizures and Epilepsy



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

Seizures and Epilepsy (continued)

- Introduction to the Brain and the Nerves
- Seizures and Epilepsy
- Hypotonia
- Parkinsonism and Parkinson's Disease
- Other Movement Disorders

More topics will be available later this year.

Recently Published: 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 (focusing on Sleep Terrors, Confusional Arousals, and Sleep Walking)

Be sure to check out all the topics in the "**Health Conditions Explained**" section of our website!

- **Heart Series**
- **Dental Series**
- **Palate Series**
- **Speech Series**
- **Mental Health Series**
- **Sleep Series**
- **22q Glossary (3rd ed.)**
- **Gastrointestinal (GI) Series**
- **Immune System Series**
- **Brain and Nerves Series**

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 any concerns, please talk to your healthcare provider.

Movement Disorders in Adults with 22q11.2DS

Dissecting the phenotypic spectrum and complexity of movement disorders in 22q11.2 deletion syndrome.

Reyes NGD, Grippe T, Callister M, Abkur T, Villanueva EQ 3rd, Heung T, Chen R, Boot E, Bassett AS, Lang AE.

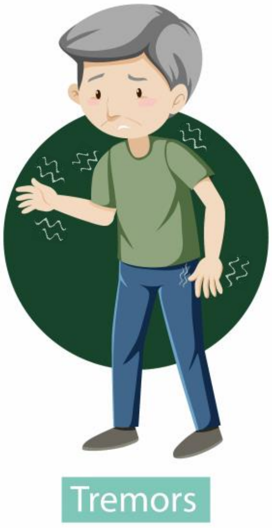
Eur J Neurol. 2025 Jun;32(6):e70256. doi: 10.1111/ene.70256. PMID: 40530538; PMCID: PMC12174967.

Since 2013, research has shown that Parkinson's disease and other movement disorders are part of the multi-system complexity of 22q11.2DS. In this study, Dr. Anne Bassett (Medical Director of our Foundation) and her team aimed to learn more about these movement disorders.

The team reviewed the medical records of 31 adults with both 22q11.2DS and movement disorders who underwent detailed neurologic assessments (checks for the brain and the nerves), including videotaped exams, at the Toronto Western Hospital Movement Disorders Centre between 1996 and 2023. The most common movement disorders were **non-parkinsonian tremors**, **parkinsonism**, and **dystonia**. With the help of modern techniques, they also identified rarer movement disorders that were previously not reported in 22q11.2DS. Importantly, there were no major differences between those who had taken antipsychotic medications (used to treat certain mental health conditions) and those who had not.

The main new finding is that **77% of adults with 22q11.2DS have more than one movement disorder** when followed on average for 1.5 years. In fact, each individual had between 2 and 7 (median of 3) movement disorders, with a median age of onset of 35.5 years. Fortunately, when accurately diagnosed, **these movement disorders are effectively managed with standard therapies**, similar to how they are treated in the general population.

This study reveals that movement disorders in adults with 22q11.2DS are more complex than previously thought but are treatable when accurately diagnosed. This strengthens the recommendation for periodic neurologic assessments and testing, when clinically indicated, in order to provide personalized care.



Would you like to find out more about tremors, parkinsonism, and dystonia? Check out these info sheets in the new **Brain and Nerves Series**:

- Parkinsonism and Parkinson's Disease
- Other Movement Disorders

The recommendations for periodic assessments (for neurologic and other aspects of 22q11.2DS) can be found in these documents:

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

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

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

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

Still Looking for Identical Twins for Research

Dr. Beata Nowakowska, a geneticist and 22q11 specialist, and her research team are conducting a study to better understand how this syndrome works at the genetic level. They are currently looking for **identical (monozygotic) twins** (of any age) diagnosed with a **22q11 deletion or duplication** to take part in this innovative study.

Why identical twins?
Because they share the same DNA, yet often show different symptoms. This fascinating difference offers a unique opportunity to explore how genetic and epigenetic factors influence the syndrome.

For more information, contact: **Dr. Beata Nowakowska** – beata.nowakowska@imid.med.pl
You can see her profile on the website of the 22q11.2 Society.

Please feel free to **share this message** with your networks, support groups, friends, and any families affected by 22q11. **The deadline to participate is December 2025.**

Together, we can make a impact on the future of 22q11 research! Thank you for your support.

2025 Lighting Request Letter Now Available

Let's raise awareness for 22q differences!

We invite you and your town to join an international movement of lighting up buildings and monuments in red on the evening of **November 22nd** (**22/11**; or 11/22 in North America) as a play on the name of the 22q11.2 deletion and duplication syndromes.

Some buildings may already have an online system for submitting lighting requests. If you do not find an online form, you can use a letter from our Foundation to **request red lighting on Saturday, November 22nd, 2025**. Click on image of the letter on the right to access the pdf file of the letter. **Download** the file to your own drive or cloud. Fill in the name of the building and the city or town, then email or mail the letter to the government or organization that manages the building.

On the night of **November 22nd, 2025**, take pictures of yourself with the illuminated building and share them on social media.

Together, we will light up the night for 22q!

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 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, 2025** 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/

Lighting Request Letter

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. 26**!

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, October 26th
12 p.m. - 4 p.m. (EDT)

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

To purchase tickets, please visit:
https://www.eventbrite.com/e/22q-boo-tickets-1708913409009

Questions? Please email 22q@chop.edu.

Faces of Sunshine Calendar 2026

The **2026 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!! 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, 2025**. **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 Box

You can also celebrate your loved one's special day by **buying a birthday box** by **November 1st**.

When you buy a birthday box, the Foundation will include an acknowledgment (first name only) and a photo of your child or adult on their birthdate on the 2026 **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.

22q
International
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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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