International 22q11.2 Foundation

Newsletter September 2023

Greetings from the International 22q11.2 Foundation. We hope you are doing well.

November is **22q Awareness Month**, so let's spread the word about 22q differences! We invite you to apply to **light up buildings in red for 22q on November 22nd, 2023**. See the bottom of this page and the letter on the next page for more information. In addition, please **tell people** about 22q11.2 deletion and duplication syndromes. The quick notes on page 3 can help you inform relatives and friends, while the clinical recommendations (links on page 4) can help your healthcare providers better manage 22q11.2DS.

Caregivers play very important roles in the lives of individuals with 22q differences. A new research article reports on the **needs of the caregivers** themselves (see page 4).

The second edition of the **22q Glossary** (see page 5) is now available! Be sure to have a look at this document that aims to help families with medical terminology that they may encounter.

Come and join **22q & Boo!** − an educational symposium and Halloween party for children at CHOP on Sunday October 29th. You can find more information and the registration link on page 6. Of course, don't forget to grab your cameras and submit your best photos for the **2024 Faces of Sunshine Calendar** (page 7). You can even pre-order the calendar now!

Keep smiling.

International 22q11.2 Foundation

Lighting up the night for 22q!

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 Wednesday, November 22nd, 2023**. Click on the "LETTER" link on the right or print out the next page of this newsletter. 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, 2023**, take pictures of yourself with the illuminated building. Post your photos on social media and tag them with **#Lucesporel22q** or **#Light_the_night_for_22q**.

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

Click this link for the pdf-fillable version of the "Lighting up the night for 22q!" letter.

LETTER

You can also print the same letter from the next page of this newsletter.

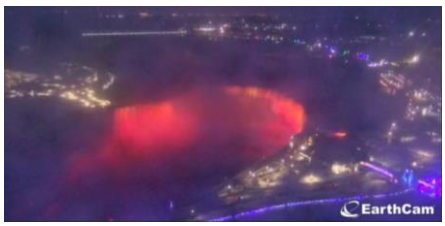
Light up the Night for 22q11.2!

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, 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, 2023** 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.
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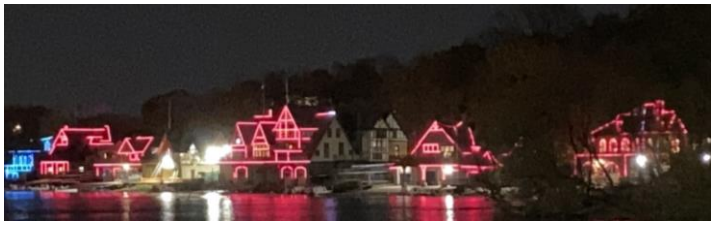
Matawan, NJ 07747, USA
www.22q.org

22q
International
22q11.2 Foundation



Telling people about 22q

Not sure what to tell people about 22q11.2DS & 22q11.2Dup? Here are some points.

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

1. Caused by a missing piece on chromosome 22q. One copy (out of 2) of some of the genes that usually tell the body how to form and function go missing
2. Happens in about 1 in 2148 babies, but not many people know about it
3. Used to be known by other names such as DiGeorge syndrome & velocardiofacial syndrome
4. Common features include:
    - Birth defects of the heart
    - Birth defects of the palate
    - Developmental delays
    - Intellectual disabilities
    - Anxiety disorders
    - Schizophrenia
    - Calcium level issues
    - Epilepsy
    - Immune deficiencies
    - Plus many others
5. Each person (even members within the same family) is affected differently in terms of symptoms and severities.
6. Usually arises as a new event in a family. Only about 10% of patients have a parent who is also affected.
7. 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. Caused by an extra piece on chromosome 22. There are 3 copies (instead of 2) of some of the genes that usually tell the body how to form and function
2. Happens in about 1 in 4000 babies, but not many people know about it
3. Much less is known about the duplication syndrome compared the deletion syndrome.
4. Common features include:
    - Birth defects of the heart
    - Birth defects of the palate
    - Developmental delays
    - Intellectual disabilities
    - Autism spectrum disorder
    - ADHD
    - Vision problems
    - Ear anomalies
    - Immune deficiencies
    - Plus many others
5. Each person (even members within the same family) is affected differently in terms of symptoms and severities.
6. Often runs in the family. About 60%-70% of patients has a parent with the duplication.
7. 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.

Telling healthcare professionals about 22q

22q11.2 deletion syndrome (22q11.2DS) is not a well-known condition, even among medical professionals. To help them care for individuals with 22q11.2DS, experts have updated the clinical practice recommendations and published them in early 2023. **We encourage you to send a copy (or a link) of the relevant guidelines to your healthcare providers.**

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

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

Prenatal screening and diagnostic considerations for 22q11.2 microdeletions

Note: The situation of each individual is different. If you have any concerns about your health, please talk to your healthcare provider.

Research: Finding out what caregivers need

An online survey to understand the needs of caregivers of family members with 22q11 deletion syndrome

Cosman T, Finless A, Rideout AL, Lingley-Pottie P, Palmer LD, Shugar A, McDonald-McGinn DM, Swillen A, McGrath PJ, Bassett AS, Cytrynbaum C, Orr M, Meier S. J Intellect Disabil Res. 2023 Jul 14. doi: 10.1111/jir.13061. Epub ahead of print. PMID: 37449408.

Children and many adults with 22q11.2DS need extra support from family members to cope with the numerous lifelong medical issues. The caregivers are the unsung heroes, but they themselves often feel lost, guilty, exhausted, frustrated, isolated, and unsupported.

To help caregivers with their wellbeing, researchers surveyed them online about their needs and the type of support they would value. On the right is a summary of the study.

Overall, the caregivers would like to have a brief online intervention that connects them with their peers and allow them to learn new skills. Medical professionals will use this information to plan online interventions for the caregivers' community in the future.

Participants

- 103 caregivers participated in online survey
  - From North America and Europe
  - Mostly mothers, also guardians and other relatives
  - One-third reported mental health problems
  - Care for individuals 0 to 42 years

Results

- What makes it hardest for the caregivers to get help?
  - Family's needs come first
  - Lack of time
  - Cost
- How would the caregivers like to get help?
  - Online intervention (with and without coach)
  - Mental health apps
  - Evidence-based and easy-to-read letters
- What motivates the caregivers to get help online?
  - A chance to discuss with others and share tips
  - An opportunity to learn new skills (with and without coach)
- What are the topics that are most important for the caregivers?
  - Practical caregiving skills
  - Problem-solving strategies with individuals with 22q11.2DS
  - Helping the child deal with learning problems and anxiety, fostering independence

22q Glossary

Second edition now available!

To help families understand terms that they may encounter, we have been developing a glossary for 22q differences. The first edition (87 entries) was published in August 2022, and we have just published the second edition, providing explanations for 217 terms. The latest version is now available on the **Health Conditions Explained** section of our website. We will add more terms in future editions. Here are 3 of the newest entries.

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

Click this link to access the glossary

22q GLOSSARY (2nd Edition)

Amblyopia ("Lazy eye") – Amblyopia refers to vision which is reduced in one eye (or both eyes in rare cases) as a result of the visual pathway between the brain and the eye not being properly stimulated due to unequal need for glasses in either eye or misaligned eyes. This condition occurs in early childhood but can be corrected using eyeglasses or penalization (patching or atropine drops). If untreated, amblyopia can lead to permanent vision loss. Since parents may not realize that the symptoms indicate amblyopia, it is recommended that children have a vision screening in their early years.

Intellectual Disabilities (ID) – Individuals with ID have significant limitations in (1) intellectual functioning (issues in learning, reasoning, and problem solving), and (2) adaptive behavior (language and math concepts, social skills, and daily living skills), originating before age 22. Many individuals with 22q11.2DS have mild ID. Severe ID in 22q11.2DS is rare. It is recommended that individuals with 22q11.2DS be assessed for both intellectual functioning and adaptive behavior every few years so that support can be provided if necessary.

Bruise – When an area of our body gets bumped, one or more blood vessels breaks. A small amount of blood leaks out into the skin and appears as the dark mark – which is the bruise. Usually, the body eventually reabsorbs the blood, and the bruise gradually disappears. However, frequent large bruising, or bruises that are raised and can be felt, may indicate a more severe problem (such as an issue with blood clotting).

Looking for more information?

For information developed for individuals and families affected by 22q differences, please visit the "**Health Conditions Explained**" section of our website. The topics published so far are:

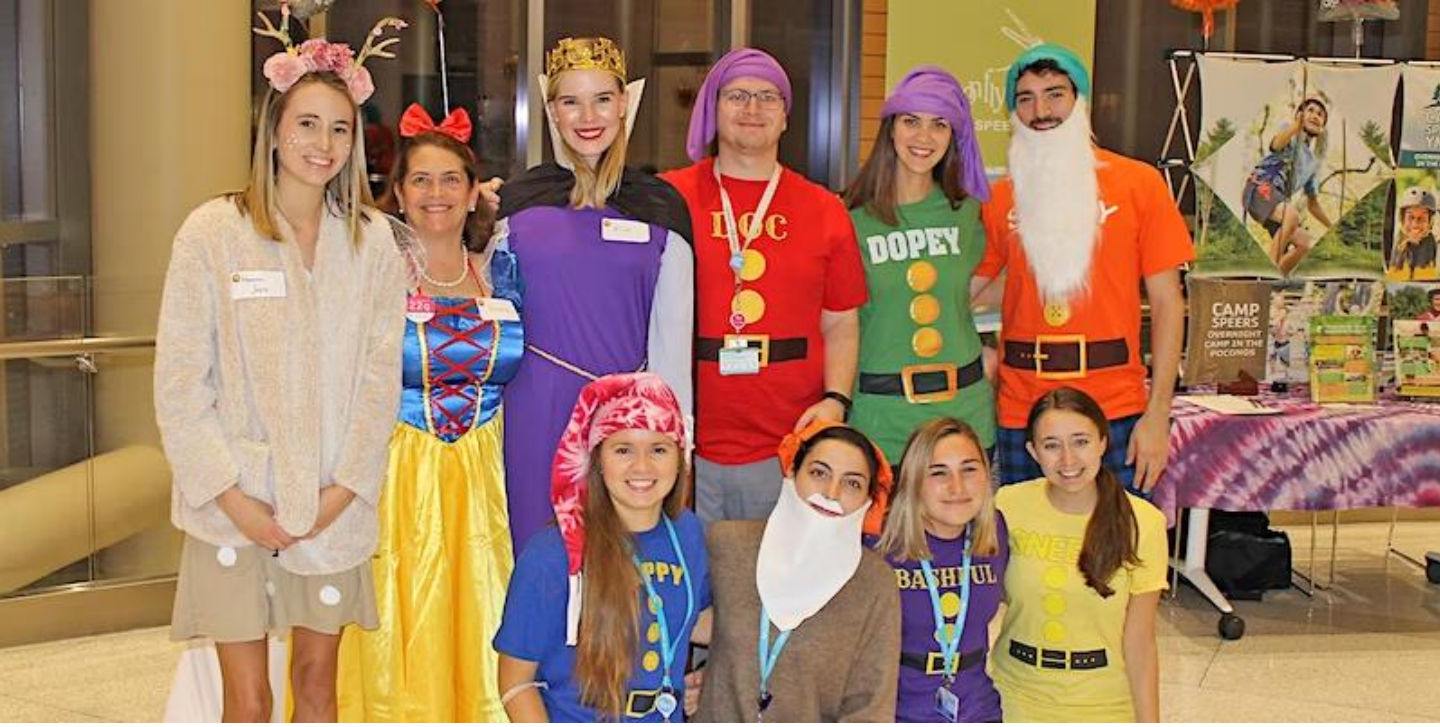
- Heart Series (5 sheets)
- Dental Series (2 sheets)
- Palate Series (6 sheets)
- Mental Health Series (9 sheets, see right panel)
- 22q Glossary

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.

Published in March 2023:

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)



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. 29!

Activities for this year's in-person event will include arts & crafts, games, and refreshments. Costumes are strongly encouraged!

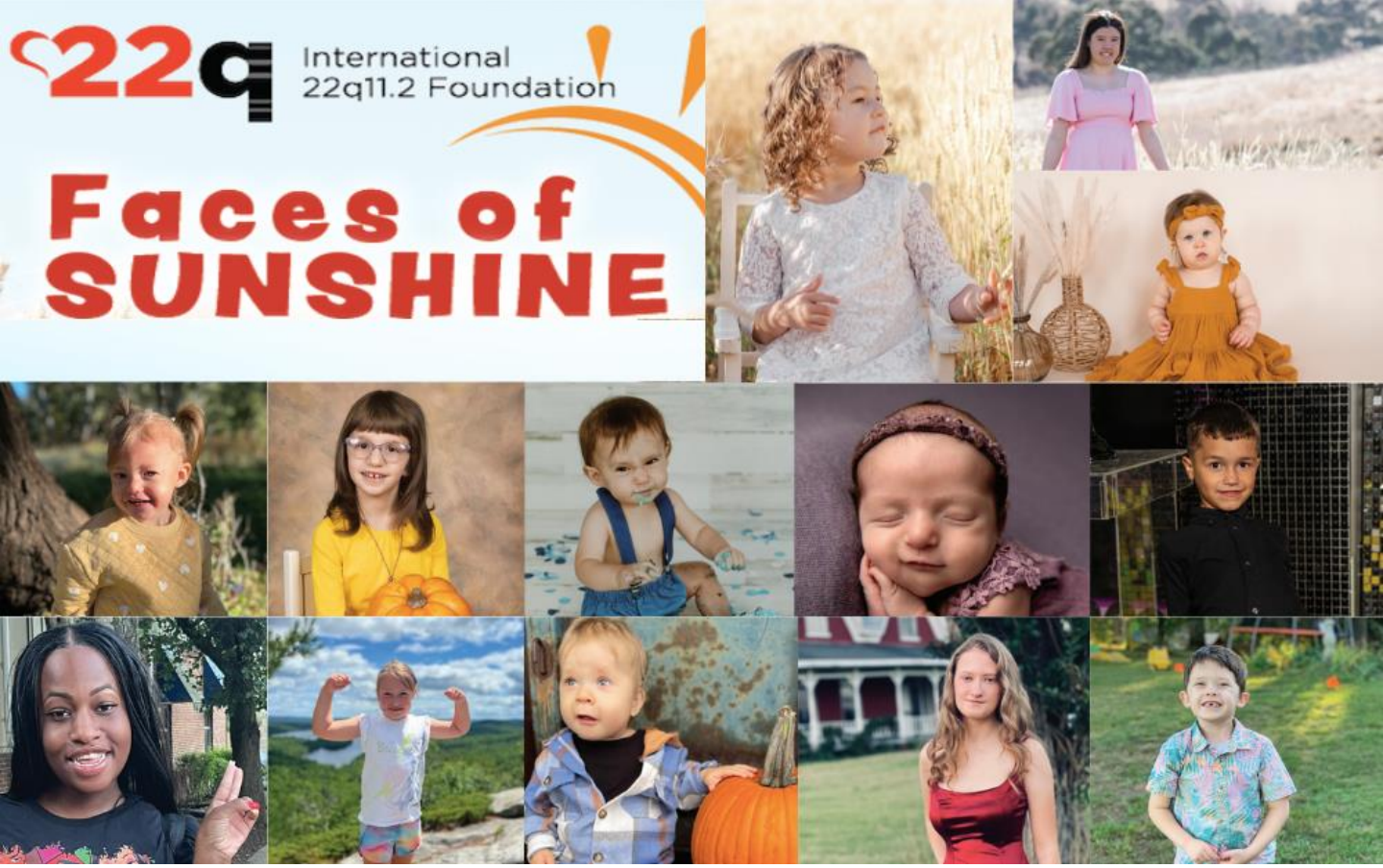
There will also be an educational component. Please visit the event website in October for more information:

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

Oct 29, 2023
12 p.m. - 3 p.m. (ET)

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

Questions?
Please call 215-590-2920 or email **22q@chop.edu**.



2024 Faces of Sunshine Calendar

The **2024 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** photo(s) to **info@22q.org by November 1st, 2023**. **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 2024 **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 Goal: 2.2 Million Dollars

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

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!

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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