International 22q11.2 Foundation

Newsletter September 2022

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

The 22q community was delighted to get together in person once again for our Professional and Family Meetings at the **12th Biennial International 22q11.2 Conference** in Croatia. See the photos on pages 6-7.

We are sharing some important information about monkeypox (pages 8-9), especially for individuals with immune deficiencies.

The first edition of the **22q Glossary** (page 2) 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 23rd (page 9).

Two important campaigns are coming up: the **Faces of Sunshine Calendar** (page 10) and **22q Awareness Month** in November (pages 1-3). In particular, we invite you to **apply to light up buildings in your city in red** to spread the word about 22q differences (pages 2-3).

Not sure how to **tell people about 22q deletion or duplication**? Check out page 5 for a quick guide. You can also see how a high school student is raising awareness about 22q11.2 duplication syndrome through the **22q Crew Awareness Project** (page 4).

Together, we can tell the world about 22q differences!

International 22q11.2 Foundation

November is 22q Awareness Month

Let's spread the word about 22q differences!

22q11.2 deletion syndrome (22q11.2DS) and 22q11.2 duplication syndrome (22q11.2DupS) are not well-known conditions, even among medical professionals. It is difficult for individuals with these syndromes to get help and be understood. Fortunately, many individuals have stepped up to raise awareness for 22q differences.

22q Awareness Month began in 2014, when proud Mom, Raj Hemus, created a Facebook Page, called **22qAwarenessDays**, that has now grown to over 9,000 followers. The International 22q11.2 Foundation agreed to share her site on our website and social media outlets to help further the mission to raise awareness, and help people learn about 22q11.2 deletion and duplication.

We encourage everyone to choose an activity and raise awareness for 22q differences. Have a look at the box on the right and start planning!

Plan now for 22q AWARENESS MONTH

Light up the night for 22q!
- See pages 2 and 3

22q Crew Awareness Project
- See page 4

Tell others about 22q differences
- See page 5

Share our posts about 22q differences

Lighting up the night for 22q!

Let's spread the word about 22q11.2 differences in your city!

In 2018, La Asociación Síndrome 22q11 in Spain launched "**Luces por el 22q**" to raise awareness for 22q11.2 deletion syndrome. Buildings and monuments were illuminated in red on the evening of **November 22nd** (**22/11**; or 11/22 in North America) as a play on the name of the syndrome. Other cities in Europe soon followed, and Philadelphia and Toronto also took part in 2021. Increasingly, the international 22q community is raising awareness for both **22q11.2DS** and **22q11.2DupS**.

We invite you to tell your local government and organizations about 22q11.2DS and 22q11.2DupS and get them involved as well! 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 November 22nd, 2022**. Click on the "LETTER" link on the right or print out page 3 of this newsletter. Simply fill in the name of the building, the city or town, and the contact information, then email or mail the letter to the government or organization that manages the building.

On the night of **November 22nd, 2022**, 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

22q Glossary

First edition now available!

To help families understand terms that they may encounter, we have been developing a glossary for 22q differences. The first edition, which contains 87 entries, is now available in the **Health Conditions Explained** section of our website. The glossary is an ongoing project, and we will add more terms in future editions. On the right are two of the entries in the first edition.

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

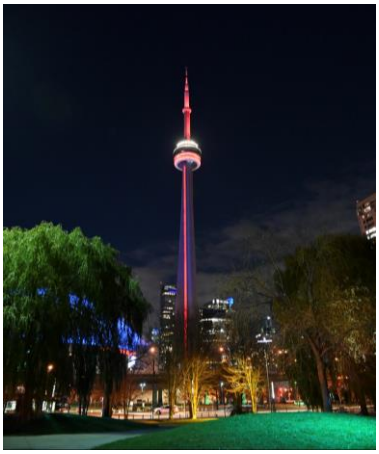
Click this link to access the glossary

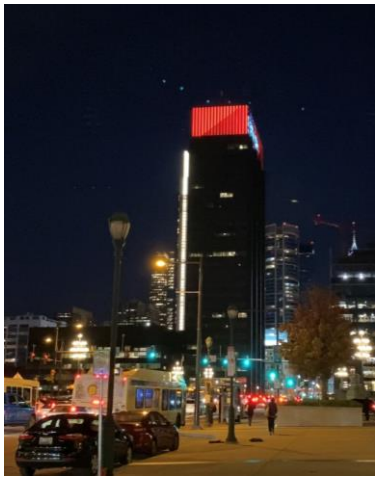
22q GLOSSARY

Phonological/Developmental Speech Sound Disorder – A phonological/developmental speech sound disorder is one in which a child is following a typical pattern of speech development but demonstrating errors that should have disappeared earlier. Phonological disorders are predictable, rule-based errors that may affect more than one sound. Phonological/developmental speech sound disorders are present in ~50% of children with 22q11.2DS.

Aspiration – Aspiration happens when someone breathes in a liquid or solid, which then goes down the airways or even to the lungs. Patients can present with coughing, choking, difficulty breathing and recurrent pneumonias.

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 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. On the left of this page, you will find example photos from November 22, 2021, where buildings in Toronto and Philadelphia 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, 2022** 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
www.22q.org

22q
International
22q11.2 Foundation

22q Crew Awareness Project

My name is Laci Wright, and I am a sophomore at Justin Garza High School, a brand-new high school located in Fresno, California. 22q11.2 duplication syndrome (22q11.2DupS) is a condition that is not known and recognized by many people, which is why I am writing this story – to promote a project that I am beginning this year, 2022.

First, you should know some background about why I want to spread awareness for 22q11.2DupS in the first place. My older brother Elijah suffers from 22q11.2 duplication syndrome, which is common but not widely known. The lack of knowledge about this genetic condition makes it difficult for those who suffer from it to get the help and support they need every day. According to the National Organization for Rare Disorders (NORD), "*22q11.2 duplication syndrome is a condition caused by an extra copy of a small piece of chromosome 22.*" 22q11.2DupS affects my brother in so many ways every day, and I want to do something to help. Since it is 2022, it's the perfect year for me to do my part to help those with 22q11.2DupS. The project that I am beginning is called the 22qCrew Awareness Project, and it will take place later this year.

First, I am asking various athletes from various sports for the month of November 2022 (22q Awareness Month), display the International 22q11.2 Foundation logo on their cleats, jersey, etc. for each game they have that month.

Secondly, I am asking the athletes to use their social media to share with everyone why they are displaying the logo, some basic information about what 22q11.2DupS is, and how everyone can help support the Foundation.

My overall goal is to select athletes who are the #22 to help with this important project. Those who join will be a part of the 22qCrew, raising awareness for the condition. This project will mainly focus on 22q Duplication, because that is what my brother has, and it is less known.

I am really hoping that this project will be a success, because it is something I have put a lot of time and effort into, but more importantly, I believe it is essential to spread awareness for 22q11.2DupS so that my brother and all who suffer from the condition can get the help & support they need. I have already contacted various athletes in numerous ways, but I have struggled to get results that I need. While working on this project, I have learned a lot – especially patience. I am grateful for those who took me seriously in my emails, and I truly believe that because of those people who trust what I am capable of, this project will be a success. I am currently trying to find new and unique ways to get the word out to the athletes I need to hear from, but I have learned that it is not easy. I am so excited to see what I can do for those with 22q11.2DupS with this awareness campaign!

Thank you for taking the time to read this story. I truly appreciate it.

Laci can be reached at

aewright2025@gmail.com

– **Laci Wright**

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.

The 12th Biennial International 22q11.2 Conference

Photos from our meetings in Split, Croatia, June 28 - July 1, 2022

Top left – Junior Investigator Award 2022 recipients with previous recipient and current 22q11.2 Society Trustees: (L-R) Donna McDonald-McGinn, Anne Bassett, Bernice Morrow, Daniel McGinn (2022), Steven de Reuver (2022), Daniella Miller (2022), Lisanne Vervoort (2018), Ann Swillen, and Peter Scambler

Top right – (L-R) Anne Bassett with Unsung Hero Award 2022 recipient Ann Lawlor

Bottom left – Angelo DiGeorge Medal – Previous and new recipients: (L-R) Anne Bassett (2014), Nicole Sarles-Philip (2022), Bernice Morrow (2018), Bruno Marino (2022), Donna McDonald-McGinn (2012), Peter Scambler (2010), and Ann Swillen (2016)

Bottom right – (L-R) Donna McDonald-McGinn with Unsung Hero Award 2022 recipient Candice Hamilton-Utgyensu

Congratulations to all the award recipients!

Family Meeting
June 26 – 28
Group Photo

Professional Meeting
June 29 – July 1
Group Photo 1

Professional Meeting
June 29 – July 1
Group Photo 2

See you in Portugal in 2024!

Monkeypox Q&A

Monkeypox is a rare disease that used to be found only in Central Africa in the past, but has now spread to many more countries. Here is some basic information about this disease.

What is monkeypox?

Monkeypox is a rare disease caused by the monkeypox virus, which is similar to the smallpox virus. Symptoms usually start within 3 weeks of exposure to the virus. People who get infected develop a rash on the face, inside the mouth, and on other parts of the body, including their genitals. The rash goes through multiple stages (2 to 4 weeks) before healing completely. Other symptoms include fever, chills, swollen lymph nodes, exhaustion, muscle aches and backache, headache, sore throat, nasal congestion, and cough.

How does monkeypox spread?

- Touching monkeypox rash, sores, or scabs from a person with monkeypox.
- Touching objects, fabrics (clothing, bedding, or towels), and surfaces that have been used by someone with monkeypox.
- Touching respiratory secretions, through kissing and other face-to-face contact.

Who are at risk of catching monkeypox?

So far in USA, men who have sexual relationships with men are at the highest risk. However, **anyone can catch monkeypox** through physical contact with someone who has it or contaminated materials such as bedsheets and towels. At least 2 children have been infected, likely through their families. [see CNBC news article]

Source: CDC's website on monkeypox

See the CDC website for the specific type of rash associated with monkeypox

Can people die from monkeypox?

The monkeypox virus that has been spreading widely in 2022 is of the West African type. Over 99% of people who get infected with this type of virus survive. However, people with weakened immune systems, children under 8 years of age, people with a history of eczema, and people who are pregnant or breastfeeding may be more likely to get seriously ill or die.

What if I think I have monkeypox?

- Avoid touching or being very near other people.
- Get checked out by a healthcare provider (wear a mask when you see them).

How can I avoid catching monkeypox?

- Avoid skin-to-skin contact with people who have a rash that looks like monkeypox.
- Avoid touching things that a person with monkeypox 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.

…Continued on the next page

Monkeypox Q&A (continued)

Can monkeypox be treated?

There are no treatments that are designed specifically for monkeypox. Nevertheless, monkeypox and smallpox are very similar, so some antiviral drugs and vaccines that target smallpox can be used to treat monkeypox. For individuals with a weakened immune system and may get seriously ill, antiviral medications may be recommended.

Can vaccines help prevent sickness?

In USA, two vaccines are available: ACAM2000 and JYNNEOS. Either one can be given before a person is exposed to monkeypox or within 4 days of exposure to prevent the onset of the monkeypox disease. Please see the CDC's Considerations for Monkeypox Vaccination for more information.

Important

Individuals who have immune deficiency disorders cannot receive the ACAM2000 vaccine because it contains live virus that can replicate. This group can receive the **JYNNEOS** vaccine, which contain live virus that is not able to replicate. However, the **JYNNEOS** vaccine is in **very short supply**.

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. 23!

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

In addition, there will also be an educational component to this year's event, focusing on "Brain and Behavior" in individuals with 22q11.2 deletion/duplication syndrome. Speakers include: Donna McDonald McGinn, MS, LCGC, Madeline Chadehumbe, MD, Raquel Gur, MD, PhD, Sarah Hopkins, MD, MSPH, Maria Mascarenhas, MBBS, Ed Moss, PhD and Cindy Solot, MA, CCC- SLP.

REGISTER

Event information

Oct 23, 2022
12 p.m. - 3 p.m. (ET)

Children's Hospital of Philadelphia
Abramson Pediatric Research Center
Room 123 A/B/C
3615 Civic Center Blvd.
Philadelphia, PA 19104

Parking for this event is available across the street in the Buerger Center for Advanced Pediatric Care, 3500 Civic Center Blvd., Philadelphia, PA, 19104. Parking vouchers will be available at the event.

Questions?

Please contact Kimberly Gaiser at 215-590-2920 or **22q@chop.edu** .

Faces of Sunshine Calendar

The **2023 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 children 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.

Quick Links

Calendar Campaign

Buy a Birthday

Pre-order your calendar

Photo Submission

To enter an individual in the calendar, please email your photo(s) (maximum 2 photos per featured person) to **info@22q.org by November 1st**. Please also sign the consent form. 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.

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 2023 **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.

We appreciate your financial support!

There are many things you can do to raise awareness and funding for 22q …from sending an email from your home computer to meeting with your member of congress in Washington DC!

Here are some of our campaigns you can choose from — or you can bring your own creative ideas and we'll help you. Just contact us at **info@22q.org**

Team 22q Fundraising Toolkit

Tell 22 Friends about 22q

Become a monthly donor

Select 22q as your Charity of Choice on AmazonSmile

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

Your support makes a difference!

Please visit the **Donate** page on our website to donate 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**.

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.

International 22q11.2 Foundation, Inc.
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