International 22q11.2 Foundation

Newsletter June 2022

Fun time at the Zoo!

What a fabulous day at the **22q for Zoo Worldwide Awareness Day**! We are still celebrating all of the participation worldwide to bring awareness to 22q11.2 differences. Our strength in unity included families, friends, organizations, and medical professional; we could never do "22q at the Zoo" without YOU! Please see pages 2 and 5 for photos.

A special thank you to the event coordinators, foundations, event locations, and 22q clinics around the world that continue to collaborate and work tirelessly to bring 22q to the forefront every day. This past celebration is because of YOU!

THANK YOU!

International 22q Family Meeting in Split, Croatia

Last call! Registration now at

https://chop.cloudcme.com/course/courseoverview?P=5&EID=2735

The 2022 Biennial International 22q11.2 Family Conference will be held in beautiful **Split, Croatia** from Sunday, June 26th through Tuesday, June 28th. Invited speakers, including family voices, are excited to share their respective expertise while learning from one another.

If you would like to attend the Family Conference, please **register by Friday June 10th**. In addition, the availability of rooms at the hotel may be limited, so please make your reservations as soon as possible. You can do so by calling Le Méridien directly at (+385 21 500 264) or emailing them at **reservations@lemeridiensplit.com**. Please let reservation staff know that you are taking part in the **12th Biennial International 22q11.2 Family Conference**.

All are invited and encouraged to attend. If you have any questions, please contact the conference planning officers at **22qmeeting@chop.edu**. We look forward to seeing you there!

22q at the Zoo - Worldwide Awareness Day

Photos from May 22nd, 2022 (Sunday)


22q11
IRELAND


phoenixzoo
May 22, 2022


Welcome to 22q at the Zoo Worldwide Awareness Day
22Q AT THE ZOO WORLDWIDE AWARENESS DAY
TREETOP CHECK-IN

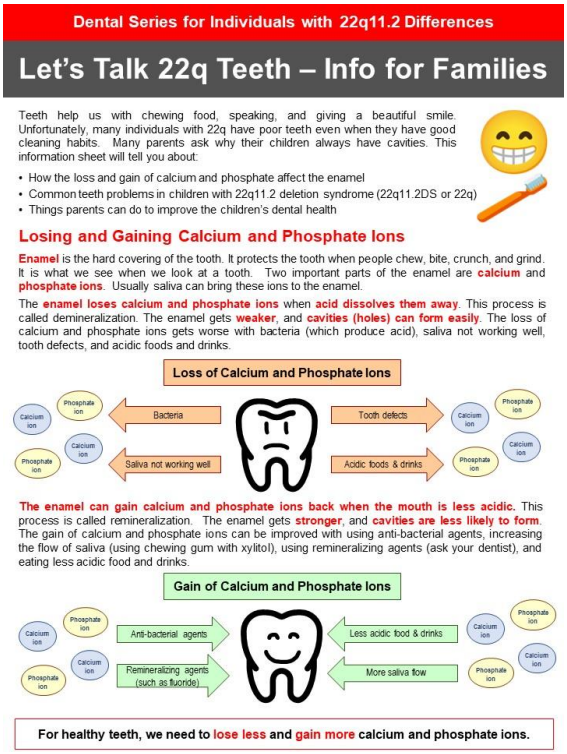
See you at the zoo next year!

Dental Series

Information sheets now available on our website

In February 2022, we launched a new section called "**Health Conditions Explained**" on our website. We began with a series of five information sheets about the heart and some congenital heart conditions that are common in children with 22q.

We have heard from families who are very frustrated with the poor teeth conditions of their children. No matter how hard they try to maintain good dental hygiene habits, the children still get a lot of cavities. The reality is that **individuals with 22q often have conditions that weaken the enamel, making it easier to have cavities**. It is also difficult to find dentists who are knowledgeable about 22q and understand what the families go through. To help both families and dentists, we have now published the **Dental Series**, which includes two information sheets (see below)



Dental Series for Individuals with 22q11.2 Differences

Let's Talk 22q Teeth – Info for Families

Teeth help us with chewing food, speaking, and giving a beautiful smile. Unfortunately, many individuals with 22q have poor teeth even when they have good cleaning habits. Many parents ask why their children always have cavities. This information sheet will tell you about:

- How the loss and gain of calcium and phosphate affect the enamel
- Common teeth problems in children with 22q11.2 deletion syndrome (22q11.2DS or 22q)
- Things parents can do to improve the children's dental health

Losing and Gaining Calcium and Phosphate Ions

Enamel is the hard covering of the tooth. It protects the tooth when people chew, bite, crunch, and grind. It is what we see when we look at a tooth. Two important parts of the enamel are calcium and phosphate ions. Usually saliva can bring these ions to the enamel.

The enamel loses calcium and phosphate ions when acid dissolves them away. This process is called demineralization. The enamel gets weaker, and cavities (holes) can form easily. The loss of calcium and phosphate ions gets worse with bacteria (which produce acid), saliva not working well, tooth defects, and acidic foods and drinks.

Loss of Calcium and Phosphate Ions

Bacteria | Tooth defects | Saliva not working well | Acidic foods & drinks

The enamel can gain calcium and phosphate ions back when the mouth is less acidic. This process is called remineralization. The enamel gets stronger, and cavities are less likely to form. The gain of calcium and phosphate ions can be improved with using anti-bacterial agents, increasing the flow of saliva (using chewing gum with xylitol), using remineralizing agents (ask your dentist), and eating less acidic food and drinks.

Gain of Calcium and Phosphate Ions

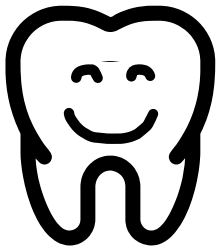
Anti-bacterial agents | Remineralizing agents (such as fluoride) | Less acidic food & drinks | More saliva flow

For healthy teeth, we need to lose less and gain more calcium and phosphate ions.

Information sheets from the Dental Series and the Heart Series can be downloaded from:

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

We will continue to add more information sheets on various topics as they become available.



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)

Coming Soon

Updated clinical recommendations for children and adults with 22q

WHY

22q11.2 deletion syndrome (22q11.2DS or 22q) is not well-known, even among healthcare providers. To help professionals care for individuals affected by the syndrome and their families, 22q experts published clinical guidelines in 2011 and 2015. We have since learned a lot more about the condition. Therefore, the guidelines needed to be updated.

WHAT

Two very important documents awaiting publication:

- Updated clinical practice recommendations for managing children with 22q
- Updated clinical practice recommendations for managing adults with 22q

WHO

- **The 22q11.2 Society**
- Expert 22q healthcare providers from approximately 10 countries, covering >20 subspecialities
- Patient advocacy organizations from 7 countries, representing over 7000 families affected by 22q
- Resource curators
- External reviewers

WHEN & WHERE

On May 3rd, 2022, the two sets of guidelines were submitted for publication.

The updated clinical recommendations will be presented during both the **Family Meeting and the Professional Meeting at the 12th Biennial International 22q11.2 Conference** in June-July in Croatia. If you would like to attend the Family Conference, please **register** by Friday June 10th.

HOW

Working together, 22q experts combined information from over 2300 studies and patient advocate survey results with their collective patient-care experiences. They generated consensus documents. Two external reviewers provided feedback and approval for the documents before they were submitted for publication.

ACKNOWLEDGEMENT

We thank the key leaders:

- Sólveig Óskarsdóttir, MD, PhD
- Erik Boot, MD, PhD
- Anne S. Bassett, MD, FRCPC
- Donna M. McDonald-McGinn, MS, LCGC

as well as everyone who has contributed to this very important project.

We will let you know when the guidelines are published!

Participating in Research

Have you ever wondered how healthcare providers learn about medical conditions? They find out a lot when they take care of patients, but they also read reports from medical research studies.

It takes time, effort, and financial support to conduct research, but **it is critically important to have enough participants** in a study in order for scientists to draw meaningful conclusions. No research studies are successful without participation by families.

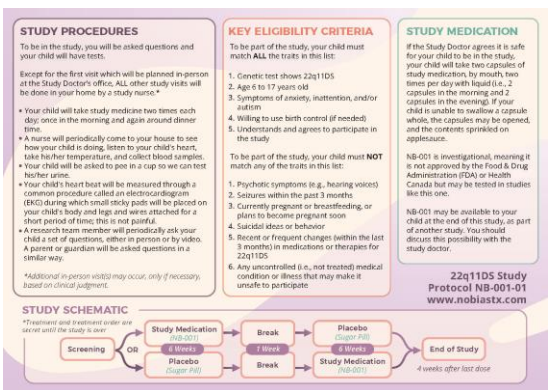
Over the years, many patients and families with 22q11.2 differences have signed up for research studies that helped healthcare providers and scientists learn about these conditions. To provide a perspective, experts who updated the 22q11.2 deletion syndrome clinical practice recommendations (see page 7) reviewed information from more than 2300 studies! Without patient and family participation, we would know little about 22q, and how best to care for people with 22q!

A number of studies are listed on the **Clinical Trials and Research page** of our website **www.22q.org**. One such study includes an international collaborative called the **Genes to Mental Health Network (G2MH)**, which is studying brain and behavior in individuals over the age of 6 years with 22q11.2 deletion and duplication syndromes. Individuals and families can participate in person or remotely, including performing standardized assessments by video and providing DNA by receiving and returning a kit to collect saliva if a sample has not already been collected. For information about the study or to find a location nearby, please visit : **https://genes2mentalhealth.com/people/**.

DISCLAIMER: The announcement below is provided for information purposes only. Unless otherwise noted, the research is not being conducted by or funded by the International 22q11.2 Foundation. The announcement and the parties conducting and sponsoring the research meet basic guidelines established by the Foundation regarding relevancy and appropriateness to 22q11.2 deletion and/or duplication syndromes. The Foundation takes no position and makes no claims as to the potential benefits of the studies, including those studies involving medications, and cannot be responsible for any outcomes, lack of outcomes or side effects. If interested in learning more about, or participating in, any research study, please contact the research study leads directly.

Research by Nobias Therapeutics

Nobias Therapeutics is launching a clinical trial of a new potential treatment for children and adolescents who have 22q11.2 deletion syndrome *plus* inattention, anxiety, or difficulty with social interactions. You can read more about the clinical trial in the **study brochure for families** or in the study profile at **ClinicalTrials.gov.**



Immunodeficiency in 22q11.2 duplication syndrome

There are relatively few research articles about 22q11.2 duplication syndrome (22q11.2DupS), but more are emerging. Here, we feature an article that reports on immunodeficiency in young people with 22q11.2DupS.

Original article: (Institutional access or payment required)
Immunodeficiency in 22q11.2 duplication syndrome
https://pubmed.ncbi.nlm.nih.gov/32949807/
Sun D, Lee J, Heimall J, Jyonouchi S.
J Allergy Clin Immunol Pract. 2021 Feb;9(2):996-998.e3.
doi: 10.1016/j.jaip.2020.09.005.

Immunodeficiency

Our immune system normally fights off things that are foreign (e.g. viruses, bacteria, fungi, etc.) without targeting healthy parts of the body.

Immunodeficiencies (or immune deficiencies) happen when the immune system is weakened. The patient may have too many infections, infections that are difficult to cure, unusually severe infections, or infections with unusual organisms. The immunodeficiency studied in the feature article is a primary immunodeficiency, meaning that the problem arises from the immune system itself (not from external factors such as chemotherapy).

To learn about the immune system and the types of immune deficiencies, please visit the website of the **Immune Deficiency Foundation**.

Patients in the Study

of patients: 7

Age: 13 months to 15 years

22q11.2 duplications: A-D, D-E, and A-E

Infection locations include:

- Ears
- Sinuses
- Respiratory tract

Other conditions include:

- Atopy (develops allergic diseases)
- Autism
- Developmental delay

A Side Note about B Cells and Antibodies

B cells mature into two main types of cells:

1. **Plasma cells** make a variety of **antibodies**, also called **immunoglobulins** (Ig). These antibodies "stick" to invaders, labelling them for destruction.
2. **Memory B cells** "remember" the invader, so the immune system can launch a faster response if the same invader comes again.

For individuals whose immune systems do not fight off infections well, **immunoglobulin replacement therapy** is a possible form of treatment. It involves injecting antibodies (Ig) either intravenously (into the vein) or subcutaneously (under the skin) to help people defend against infections.

Findings

- 3 patients have too few memory B cells.
- 3 patients had rapidly decreased protection from vaccines compared to expected levels.
- 4 patients continued to have repeated infections even with preventative antibiotics. After these patients were given immunoglobulin replacement therapy, their frequencies of infections decreased. Even when they needed antibiotics to treat infections, they needed them for a shorter time.

Main Message

22q11.2 duplication syndrome patients may develop antibody deficiencies. We are still learning about the immune system in people with the duplication of 22q11.2. **Those with recurrent infections should be referred to immunology**.

Immunoglobulin replacement therapy can significantly improve their quality of life.

Meet Teresa Elena

March 1, 2022

Teresa Elena Mataya was born on June 22, 2017, in Nashville, Tennessee, at Monroe Carrell Jr. Children's Hospital (Vanderbilt). She was diagnosed prenatally with the 22q11.2 microdeletion syndrome and an Interrupted Aortic Arch Type B. Four days after birth, she had surgery to repair the arch. They then had to go in again to stop bleeding later that night.

As part of the heart surgery, one of her vocal cords was paralyzed, so Teresa needed a feeding tube put in. During the G-tube implantation, the surgeon 'nicked' a major blood vessel and she nearly died; she was saved by the cardiac anesthesiologist.

Shortly after her discharge from the hospital, we moved to San Antonio, Texas. She has had several surgeries since, including surgery to correct a tethered spinal cord.

Teresa is currently being homeschooled due to her speech delays (due to VPI) and lower-than-normal immune system. Teresa loves counting, and she can count past ten in five languages (English, Spanish, French, Basque, and Latin)!

Before COVID-19, Teresa was involved with gymnastics with an organization called Kinetic Kids. Teresa loves her toy kitchen and has been "cooking" lots of food for her mommy while she's pregnant with Teresa's little sister. She's involved in hippotherapy (horse-therapy) and is improving daily with that. She also loves going to "ball-games" with her daddy (baseball, soccer, rugby, etc.).

While Teresa has had a lot of struggles in her short life so far, she is very positive about everything and loves being with her mommy and daddy. She loves her doctors at Children's Hospital of San Antonio, and she loves going on "adventures" with her daddy to places like the Spanish Missions in San Antonio.

For more inspirational stories, please visit:
https://22q.org/inspirational-stories/

Teresa Elena's photo was featured in the month of March in the 16th Annual Faces of Sunshine Calendar.

Would you like to share your story like Teresa Elena did?

We all need inspirations every now and then. If you or someone you know is living and thriving with 22q, please share your story! Please download this questionnaire and email your information to us at info@22q.org, along with two, high quality photos, and we will contact you for more information!

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.

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