International 22q11.2 Foundation

# Newsletter March 2022

## It's Zoo Time!!!!


Our annual 22q at the Zoo event aims to raise awareness about chromosome 22q11.2 differences while providing families, friends, and professionals a chance to socialize. See page 2 for the story behind this amazing event. Please visit our **22q at the Zoo** webpage to find a participating zoo in your area. We look forward to celebrating this most **AMAZING** day with you on **Sunday**, **May 22**!

## International 22q Family Meeting in Split, Croatia

**Registration is now Open!**

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. For the first time, the family meeting, intended for individuals aged 16 years or older interested in/affected by chromosome 22q11.2 deletion or duplication differences will include three full days of information sharing and friend raising with an intentional emphasis on Integrative Health and Wellness, featuring hands on demonstrations such as yoga, aromatherapy, meditation, nutrition, mindfulness, photography, art therapy, and writing for relaxation. Traditional lectures will focus on medical conditions, school challenges, behavioral differences, social considerations, and advances in cutting-edge basic science including developing novel treatments. Breakouts will focus on concerns by age (early childhood, school-aged, adults).


12th Biennial International 22q11.2 Conference

Family Meeting
June 26 – 28, 2022
Le Méridien Lav Hotel
Split, Croatia

For more information visit CHOP.cloud-cme.com

Families and presenters will be traveling from around the globe, representing numerous countries across four continents. A fantastic social program, inclusive of a conference dinner on June 27th, is certain to provide plenty of 1:1 time with invited speakers, while offering tremendous opportunity for creating lasting memories and lifelong friendships. All are invited and encouraged to attend. We look forward to seeing you there!

# History of 22q at the Zoo - Worldwide Awareness Day

22q at the Zoo – Worldwide Awareness Day® launched for the first time on May 22, 2011. It was originally planned as a Philadelphia (USA) event to promote friendship and raise awareness. Thanks to families, professionals, and other support organizations around the globe, this "local" function morphed into a worldwide phenomenon where people gathered at their closest zoo for a day of fun and solidarity, sporting our unique 22q at the Zoo T-shirts. While adults socialize, children enjoy the animals, face painting, music, dancing, parades, and craft activities. During all of this fun, teams of volunteers distribute fact sheets informing the lay public about 22q11.2 deletion and duplication syndromes.

Just 12 weeks prior to that first event in 2011, Prof. Donna McDonald-McGinn, the Founding Non-Parent Board Member of the International 22q11.2 Foundation and Director of the 22q and You Center at the Children's Hospital of Philadelphia, who prides herself on rhyming 22q related events, called one of our then, newest board members, Sheila Kambin, and said, "I was thinking…what if we made 22q at the Zoo a worldwide event?"… "I will contact my international colleagues and support organizations and you can generate interest with families using social media." That year, 65 cities around the world participated in the first ever 22q at the Zoo – Worldwide Awareness Day.

This success was certainly the result of dedicated families and professionals joining in and doing their part, combined with the amazing reach of social media. With the help of Facebook alone, we reached a massive audience around the globe, including those just waiting for an organization to take the lead in promoting solidarity around 22q.

Please visit our **22q at the Zoo** webpage to find a participating zoo in your area.

Every year, on 22q at the Zoo day, everyone is included. There are no barriers. All cities, states, provinces, counties, towns, nations, districts, and continents are welcome to participate. Those without a zoo go to a park or someone's backyard. Those who live in areas where it is too hot or too cold gather at indoor locations. Those who use the terms DiGeorge, VCFS, CTAF, Opitz G/BBB, Cayler, Cardiofacial, and 22q11.2 are all equally welcome. 22q at the Zoo, at last, is a day where everyone can join in making friends, having fun, promoting public awareness, and sharing a common bond.

Since our initial inception, additional locations from all around the globe have joined in the annual fun. In 2019, a record 115 cities celebrated our Worldwide Awareness Day ,posting photos from Down Under to the South Pacific and everywhere in between. Moving forward, it is our sincere hope that 22q at the Zoo will continue to grow each and every year. It is our hope that one day we will have a zoo event in every country around the world. It is our hope that our world will remain small – small enough that we can continue to support one another around 22q. 22q at the Zoo is a happy experience, fiercely battling isolation and hopelessness. 22q at the Zoo – Worldwide Awareness Day is a day to remember, a day to celebrate, a day to make friends while creating awareness, and most importantly, a day to stand together. Please join with us because we cannot do it without you!


**May 22, 2022 (Sunday)**

# The Biennial International 22q11.2 Conference

Our most recent conference took place in Whistler in 2018. Can you believe that it has been 4 years since we were able to come together?

Let these photos from our 2018 Conference bring back some wonderful memories!

Group photo – Professional Meeting


Booth in the foyer

Group photo – Family Meeting

22q experts – Family Meeting

**The 12th Biennial International 22q11.2 Conference**
**Split, Croatia**

**Family Meeting** [Registration Website]

- **June 26 (Sunday) to 28 (Tuesday), 2022**

International Brain and Behaviour Consortium (IBBC); Guidelines, Clinics, and Networks

- June 28 (Tuesday), 2022

Professional Meeting

- June 29 (Wednesday) to July 1 (Friday), 2022

## Let's get together in Croatia!

If you have any questions, please contact the conference planning officers at 22qmeeting@chop.edu.

# Updating the Practical Guidelines for 22q11.2 Deletion Syndrome

22q11.2 deletion syndrome (22q) is not a well-known genetic syndrome, even among healthcare providers. To help medical professionals care for individuals affected by chromosome 22q11.2 deletion syndrome, a panel of healthcare providers and researchers who are familiar with 22q published the first Practical Guidelines for Managing Patients with 22q11.2 Deletion Syndrome in 2011. In response to the needs of the growing adult patient population, the experts published the Practical Guidelines for Managing Adults with 22q11.2 Deletion Syndrome in 2015. In 2019, a group of experts published a set of specialty guidelines called Speech-Language Disorders in 22q11.2 Deletion Syndrome: Best Practices for Diagnosis and Management.

Likewise, a seminal overview paper has been published in Nature Review Primers.

Over the years, through clinical encounters and research, experts have learned a lot more about 22q. Some of the content in the 2011 and 2015 documents requires updating. So, our group has been working tirelessly together remotely to update these guidelines, and we expect to submit them for publication within the first quarter of 2022. The abstracts of both guidelines have been submitted for presentation during the 12th Biennial International 22q11.2 Conference in June-July in Croatia. We take this opportunity to thank the key leaders:

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

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

# A Way to Support Ukrainian Refugees

## Including Those with 22q Differences and Other Disabilities

We stand in solidarity with our European colleagues and families, and most importantly the Ukrainian people and those helping displaced persons across Europe.

If you are looking for a way to help Ukrainian refugees, here is an option. 22q11.2 Society advisor, member, and basic scientist, Dr. Beata Nowakowska, is housing families, feeding newly arriving refugees at the train station in Warsaw, and working closely with the Polish 22q Association (Stowarzyszenie 22q11 Polska) to **support families of children with disabilities, including those affected by chromosome 22q11.2 differences, as well as others arriving daily to Poland**.


Current estimates suggest that >1.2 million Ukrainians are now in Poland and numbers are expected to continue rising. To support these efforts, Stowarzyszenie 22q11 Polska has created a fundraiser, with a website in Polish, English and Ukrainian (the payment page is in English as well).

Please consider **making a donation**. Every little bit helps. Thank you.

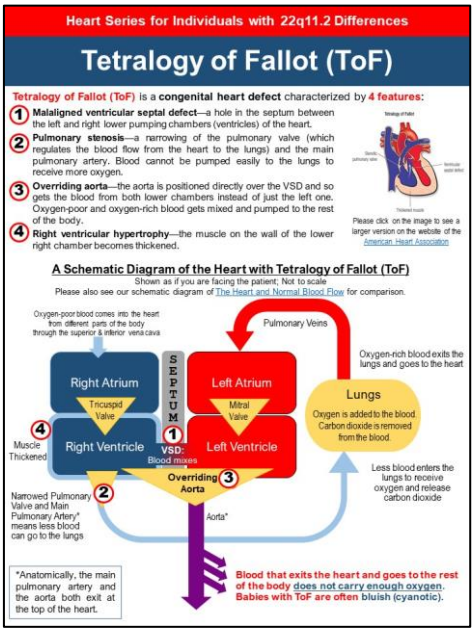
# Heart Series

## Information sheets now available on our website

Medical terms can be overwhelming, especially for families who are affected by multiple health conditions. To solve this problem, we have started a new page called "**Health Conditions Explained**" on our website. 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.

Our first series is all about the heart:

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




The series is available on: **https://22q.org/symptoms-care/health-conditions-explained/**

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

# Coming Soon: A Glossary for 22q Differences

Another way that we are helping families understand medical terms is the development of a glossary for conditions related to 22q differences. We are currently generating an alphabetical list of health-related words and their meanings. It is an ongoing project, and we will post updated versions on our website once in a while. Here is a preview:

**Autoimmune Disorders** – Our immune system can usually distinguish between foreign entities and our own body. Autoimmune disorders happen when our immune system attack components of our own healthy body by mistake. There are over 100 autoimmune disorders. The overall frequency of autoimmune disease may be as high as 25% in 22q11.2 deletion syndrome.

**Coloboma** – A coloboma is a condition in which the eye is missing some tissue at birth. There are different types of colobomas, depending on the exact type of tissue that is missing. A coloboma can result in a loss of vision as well as sensitivity to light.

**Seizure** – Electrical pulses are a normal part of brain cell activities that contribute to how people think and behave. A seizure happens when there is a sudden uncontrolled surge of electrical activity in the brain, and can affect the patient's behavior, movements or feelings, and even consciousness. There are multiple causes of seizures, with a wide range of severity. Some patients need medications or even surgeries to manage the seizures while others do not.

# COVID-19 Vaccination

## Did you receive your COVID-19 vaccines?

COVID-19 vaccines contain no virus capable of causing disease, so they are safe – and indeed recommended for people with immune deficiencies. If you are allergic to components of the vaccine, please check with your allergist for instructions.

### Our Medical Advisory Board's recommendations for Individuals who are eligible to receive COVID-19 vaccines:

It is recommended that **all persons with or without chromosome 22q11.2 differences receive the COVID-19 vaccine**, unless the person's treating physician/cardiologist determines otherwise.

It is also recommended that **all persons with immunodeficiencies receive four doses of the mRNA vaccines**, unless the person's treating physician/allergist determines otherwise.

## Eligibility for COVID-19 Vaccination

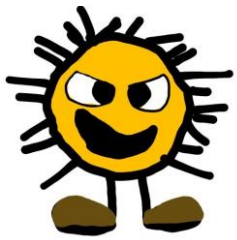
In the **USA**:

- Everyone aged 5 and over are eligible to receive the vaccine.
- Individuals aged 12 and over who received the Pfizer-BioNTech vaccine are eligible to get a booster.
- Adults aged 18 and over who received the Moderna vaccine are eligible to get a booster.
- See the **CDC website** for details about eligibility.

If you live **outside of the USA**, please check with the health authority in your area for vaccination eligibility.

## Why receive a booster?

- Vaccine effectiveness may decrease over time.
- Data so far shows that with a booster, there is good (if imperfect) protection from the omicron variant.
- See the **CDC website** for details about boosters.



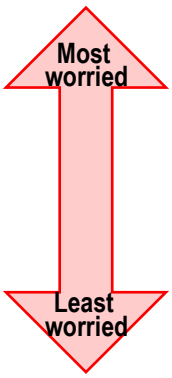
**If you are concerned about receiving COVID-19 vaccines or have any questions about your health, please check with your healthcare provider directly.**

# COVID-19-related Worries and Medical Disruptions in Families Affected by 22q11.2 Differences

The COVID-19 pandemic has changed many aspects of life, and the suffering and stress are affecting mental health as well. In addition, it has disrupted medical care for many individuals with 22q11.2 deletion and duplication syndromes, who often have a variety of healthcare needs. To better understand the impact of the pandemic has had on families with 22q differences, the Genes to Mental Health (G2MH) Network consortium sent out surveys that asked about COVID-19-related worries and disruptions. Most respondents were primarily female, and many were parents of patients. Here are the key points of the report.

- Respondents from the third wave (9/2020 to 2/2021) reported more worries compared to those from earlier waves.
- Care-related interruptions had a significant effect on health for 36% of respondents
- Rehabilitation appointments were cancelled for 20% and postponed for 45% of participants. Services with medical specialists or general practitioners were postponed for 44%.
- 44% of those who usually received primary care at hospitals avoided them for fear of catching COVID-19.
- 89% of participants reported no disruptions to the availability of medications and/or treatment, but 59% were worried that disruptions would happen if pandemic continued.
- Worries from families affected by 22q differences are similar compared to other population cohorts.
- Although the levels of medical disruptions are similar compared to other studies, the impact on perceived health on the 22q differences cohort may be higher.

## COVID-19 Stresses for Families with 22q Differences




Family getting COVID-19

Unknowingly infecting others

Self getting COVID-19

Financial burden from pandemic

Dying from COVID-19

Currently having COVID-19

**Higher levels of worries** reported by:

- Those with health effects due to medical disruptions
- Those avoiding primary care at hospitals
- Those with a higher level of fear of disruptions of medication or treatment

For details, please refer to the original article. Institutional access or a small payment is required.

**The COVID-19 pandemic's impact on worry and medical disruptions reported by individuals with chromosome 22q11.2 copy number variants and their caregivers**

https://pubmed.ncbi.nlm.nih.gov/35191118/

White LK, Crowley TB, Finucane B, Garcia-Minaur S, Repetto GM, van den Bree M, Fischer M, Jacquemont S, Barzilay R, Maillard AM, Donald KA, Gur RE, Bassett AS, Swillen A, McDonald-McGinn DM.

J Intellect Disabil Res. 2022 Feb 21. doi: 10.1111/jir.12918. Online ahead of print.

## Feeling stressed?

Here are some resources that may help:

**U.S. Department of Health & Human Services**
Mental Health and Coping during the Coronavirus (COVID-19) Pandemic

**Centers for Disease Control and Prevention**
Coping with Stress

**If you are concerned about COVID-19 or any aspects of your health, please check with your healthcare provider directly.**

# Clinical Features of 22q11.2 Duplication Syndrome

Although both 22q11.2 duplication syndrome (22q11.2DupS) and 22q11.2 deletion syndrome (22q11.2DS) affect the same area of the chromosome, 22q11.2DupS is not as well-studied as 22q11.2DS. A recent publication reports on the clinical findings of 42 children with 22q11.2DupS and compares them with those of 120 who have been described previously. Here is a summary of findings seen in both the current study and in existing medical literature.

## Birth Defects

Heart
Urogenital
Cleft lip / cleft palate
Bifid uvula
Brain

## Growth

Poor growth
Microcephaly
Macrocephaly

## Neurological

Intellectual disability
Learning disabilities
Developmental delay
Abnormal muscle tone
Seizures

## Eyes and Ears

Eye / vision anomalies
Hearing loss

## Behavior / Psychiatric

Autism
ADHD / ADD
Anxiety
Other diagnoses

## Speech

Speech delay
Velopharyngeal insufficiency (VPI)
Hypernasal speech

Additional findings in the 42 children in the study include (but are not limited to):

- Sleep apnea or suspected sleep disordered breathing
- Asthma or reactive airway disease
- Allergies and immunodeficiency
- Constipation, gastroesophageal reflux
- Kidney abnormalities and/or difficulty urinating
- Abnormalities of bones, joints, and muscles
- Low calcium levels, hormone deficiency
- Need for surgery (heart, ears, and/or others)

In many cases, the 22q11.2DupS is inherited from a parent. Nevertheless, affected members of the same family often have different clinical features. Affected parents who have no birth defects or only relatively mild symptoms themselves are sometimes not diagnosed until their children are.

## Recommended Routine Checks

**Cardiac screening (Heart)**

**Abdominal ultrasounds (Genitourinary)**

**Audiology evaluation (Hearing)**

**Ophthalmology evaluation (Vision)**

For details, please refer to the original article. Institutional access or a small payment is required.

**22q11.2 duplications: Expanding the clinical presentation**

https://pubmed.ncbi.nlm.nih.gov/34845825/

Bartik LE, Hughes SS, Tracy M, Feldt MM, Zhang L, Arganbright J, Kaye A. Am J Med Genet A. 2021 Nov 29.

# Talking to Children about Psychiatric Conditions – Feedback from Parents of Individuals with 22q11.2DS

Individuals with 22q11.2 deletion syndrome (22q11.2DS) have a higher risk of developing mental health problems. It can be challenging for parents to find suitable words and an appropriate time to discuss this stigmatized topic with their children, and the children may or may not understand. A group of researchers interviewed 10 parents of 9 children with 22q11.2DS to get their perspectives and to see how medical professionals can help. Here are some of their findings.

## What helps

- Parents choosing to be proactive and control what they can
- Parents having skills or experiences working with people with disabilities
- Parents being open with the children and taking the stigma away early on
- Parents drawing parallels between mental illness and other features of 22q11.2DS

## What makes it harder

- Parent wanting to protect the child from fears and worries of something that "may" happen
- Children understanding black-and-white situations but not an "increased risk" of something happening

## Depending on the situation…

- Parents who feel that their child are at a higher risk of developing mental health problems are likely to discuss the topics sooner.
- Parents who feel that 22q11.2DS is part of their child's identity are likely to talk about mental health in the context of their genetic diagnosis.

## When to talk

- Use the child's developmental age (not chronological age) and emotional maturity as a guide
- Talk about mental health as part of other important conversations as the tween/teen years approach

## How professionals can help families

- Discuss with the parents what the conversation about mental health with their child may look like
- Recommend suitable and positive words that the parents can use and the child can understand
  - One possibility is to use the mental illness jar analogy by genetic counsellor Dr. Jehannine Austin. To see the analogy adapted for 22q11.2DS, please see the original article of the current study (details below).
- Advise parents about potential dilemmas and miscommunications
- Maintain a trusting relationship with the family and provide consistent proactive support, even as the child gets older
- Personalize the support to suit the parents, the child, and the family
- **Be informed about 22q11.2DS**

For details, please refer to the original article. Institutional access or a small payment is required.

**Parents' perspectives, experiences, and need for support when communicating with their children about the psychiatric manifestations of 22q11.2 deletion syndrome (22q11DS).**

https://pubmed.ncbi.nlm.nih.gov/34783994/

Cook CB, Slomp C, Austin J. J Community Genet. 2021 Nov 16. doi: 10.1007/s12687-021-00558-9.

# 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!

There are so many options for **Team 22q Fundraising**! Take part in one of the Foundation's event or create your own. You can also have a **No Show Valentine's Ball** even after February is long gone.

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**.

# Thank you!

## Team 22q Fundraising

- **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**

The honor of your absence is requested by
The International 22q11.2 Foundation

at our 2022

*No Show Valentine's Ball*

*at* your *home*
*at* any *time*

It's a fabulous affair you will never attend.
We will proudly present
not a single speech, no entertainment,
no cocktail hour, no valet parking,
no need to be there.

Instead...
WE CORDIALLY INVITE YOU TO
JOIN US IN SPIRIT

By making a contribution to
The International 22q11.2 Foundation

22q International 22q11.2 Foundation

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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