International 22q11.2 Foundation

Newsletter June 2024

Photo above: 22q at the Zoo at Philadelphia Zoo, PA, USA.

Please check out p. 2-3 and our social media for more photos

To see a list of all the locations of the zoo events in 2024, visit our 22q at the Zoo page.

If you would like to host a zoo event locally next year, check out our hosting guide.

In this newsletter…

22q at the Zoo – Thank You!

The International 22q11.2 Foundation salutes you for once again bringing "22q at the Zoo—Worldwide Awareness Day" to hometowns - big, small, and in between all around the globe - so thank you from the bottom of our hearts!

The 14th Anniversary of this amazing worldwide awareness and friendship raising event has been brought to you by the International 22q11.2 Foundation (22q.org) in collaboration with our remarkable colleagues at family support organizations and 22q Centers of Excellence from all corners of the Earth. We acknowledge each and every one of you including the parents, grandparents, caregivers, relatives, friends, healthcare providers, and volunteers who gave so much time to bring this very special event to fruition. Particularly, we would like to give a shout out to all the coordinators from all over the world! We thank you for helping us raise awareness.

We are especially grateful to the children and adults with 22q11.2 differences, along with their siblings, parents, other relatives, and friends, who in some locations braved unfavorable weather to go to the zoo (or park, aquarium, backyard, or playground) for 22q! We are also so very grateful for the in-kind donations from Natera and the 22q and You Center at the Children's Hospital of Philadelphia, which contributed to another absolutely fantastic annual collaborative event!

We definitely went to the zoo for 22q… And we couldn't have done it without you!!!

Looking forward to seeing you next year on

May 18, 2025!!!

22q at the Zoo - Worldwide Awareness Day

A small selection of the photos from May 19, 2024

Zoo Planckendael, Mechelen, Belgium

St. Louis Zoo, MO, USA

Palić Zoo, Subotica, Serbia

Roller skating in North Tonawanda, NY, USA

Cleveland Metroparks Zoo, OH, USA –
3 mothers telling the public about 22q!

Nagano Zoo, Nagano, Japan

Dolphinarium Hardervijk, The Netherlands

Bronx Zoo, Bronx, NY, USA

Fota Wildlife Park, Cork, Ireland

Orange County Zoo, Orange, CA, USA

Como Park Zoo, St. Paul, MN, USA

See you next year on

May 18, 2025!

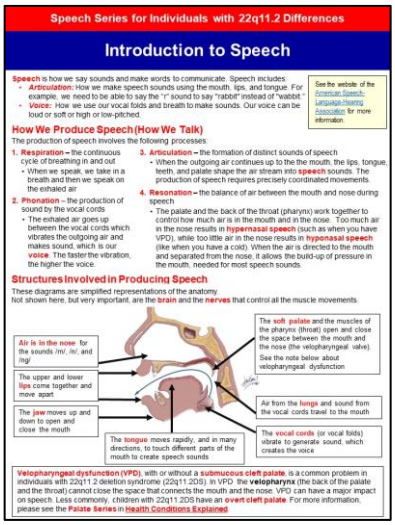
Visit our social media for more photos.

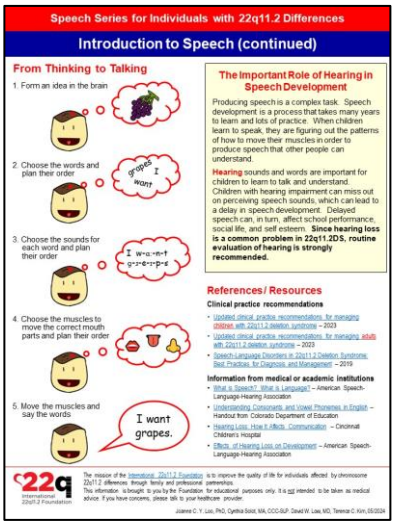
Speech Series

Information sheets now available on our website

We are happy to let you know that the 8 topics of the **Speech Series** are now available in the "**Health Conditions Explained**" section of our website.

Our newest series explains 22q-related speech and voice disorders. Special thanks to Cynthia Solot, Speech-Language Pathologist; Joanne Loo, Programmatic and Educational Tool Developer; Dr. David W. Low, Plastic Surgeon and Medical Illustrator; and Terence C. Kim, student.





SPEECH SERIES

- Introduction to Speech
- Consonants and Vowels in English
- Speech Disorders in Individuals with 22q11.2 Deletion Syndrome – an Overview
- Speech Disorders in Individuals with 22q11.2 Duplication Syndrome – an Overview
- Voice Disorders
- Speech Sound Disorders
- Motor Speech Disorders
- How Parents and Professionals Can Help

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.

Health Conditions Explained

HEART SERIES

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

DENTAL SERIES

Let's Talk 22q Teeth – Info for Families
Dental Health in Children with 22q – Info for Dentists

PALATE SERIES

The Velopharynx
Cleft Palate & Submucous Cleft Palate
Velopharyngeal Dysfunction: Introduction and Causes
Velopharyngeal Dysfunction: Diagnosis
Velopharyngeal Dysfunction: Surgery
Velopharyngeal Dysfunction: Speech Issues

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)

GI SERIES

GI Issues in 22q11.2 Deletion Syndrome
The Digestive System
Feeding Difficulties
Swallowing and Dysphagia
Constipation
Gastroesophageal Reflux Disease (GERD)
Esophageal Dysmotility
Gastroparesis
Cholelithiasis (Gallstones)
Nausea and Vomiting
Autoimmune Issues in the GI System
Inguinal Hernias
Nonalcohol Fatty Liver Disease (NAFLD)

22q GLOSSARY

Second Edition (217 entries)

Stress and the Risk of Psychosis in Individuals with 22q11.2 Differences

Associations between acute and chronic lifetime stressors and psychosis-risk symptoms in individuals with 22q11.2 copy number variants

Modasi J, Khachadourian V, O'Hora K, Kushan L, Slavich GM, Shields GS, Velthorst E, Bearden CE.
Psychol Med. 2023 Nov;53(15):7222-7231. doi: 10.1017/S0033291723000740. Article available for free

When a person's abilities do not match what the environment requires, the person may experience **stress**. In the general population, stress is a known risk factor for **psychosis**, a symptom in which the person has difficulty telling apart what is real and what is not. Researchers wanted to find out if stress is also associated with the risk of psychosis in people with 22q11.2 deletion syndrome (22q11.2DS) and 22q11.2 duplication syndrome (22q11.2DupS).

Study population

- Individuals who are involved in brain and behavior observational studies at the University of California Los Angeles (UCLA)
- 100 participants (65 had complete data), aged 6 to 42 years
- 46 with 22q11.2DS, 30 with 22q11.2DupS, 24 controls

Measurements

- Major life stressors – both acute life events and chronic difficulties. E.g. work, housing, financial etc.
- Symptoms of psychosis (see box on the bottom right)

Results

- 16% (7 people) in the deletion group had a psychotic disorder. No one in the duplication group had it.
- Both the deletion and the duplication groups had more counts of acute stressors compared to controls.
- The duplication group had more severe acute stressors than each of the deletion group and controls. These stressors involve housing, finance, role change, and physical danger.
- Lifetime chronic stress counts and severity were similar for the deletion and duplication groups.
- **The deletion group showed a strong association between chronic stressors and positive symptoms of psychosis** (see box on the right). **The duplication group did not show this association.**
- In the duplication group, lifetime stressors were related to the general symptoms (mood, sleep problems) .

Conclusions

- **Stress is a risk factor for psychosis in individuals with 22q11.2DS.** Minimizing the effect of stress may decrease the risk of psychosis in this group.
- Consistent with previous results, **22q11.2DupS appears to protect against psychotic symptoms, even in the presence of more severe stressful life events.**

Psychotic disorders (including schizophrenia) are **treatable** mental health conditions. These disorders often begin during teenage years or in early adulthood.

Individuals with 22q11.2DS have a higher risk of developing schizophrenia than those in the general population. It is important to identify changes in thinking, emotions, behavior, and functioning in order to intervene early for the best possible outcome.

For more information, please have a look at the information sheet on Psychotic Disorders in the Mental Health Series in the Health Conditions Explained section of our website.

Symptoms of Psychosis
(not a complete list)

Positive symptoms:
Changes in behavior or thoughts. E.g. hallucinations, delusions, grandiose ideas

Negative symptoms:
Social withdrawal. E.g. Avoiding people (even friends), losing interest, not feeling pleasure etc.

Disorganization symptoms:
Odd behavior, not taking care of personal hygiene, lacking focus etc.

General symptoms:
Sleep disturbances, mood changes, decreased tolerance to stress etc.

22q11.2 Deletions Greatly Increase the Risk of a Birth Defect in the Central Nervous System

Risk of meningomyelocele mediated by the common 22q11.2 deletion

Vong KI and others (See the above link for the complete list of authors)
Science. 2024 May 3;384(6695):584-590. doi: 10.1126/science.adl1624.
Download this article for free

Meningomyelocele (MM) is a severe type of neural tube defect (see box on the right), but the causes are unknown. A large group of researchers are working together to identify genetic differences that increase the risk of MM.

Searching for the genetic cause of MM

- 715 trios (child with MM, + parents) from USA, Mexico, Brazil, Canada, Italy, Georgia, Egypt, Nigeria, and Pakistan participated.
- The DNA of these 715 trios was sequenced.
- Researchers found 22q11.2 deletions in 6/715 patients with MM.
  - Deletion extent: 2 A-D, 2 B-C, 2 C-D (**C-D is deleted in all 6**)
  - 3/6 patients' mothers did not take folate before pregnancy
- Frequency of 22q11.2 deletions in controls = 4/10847
- Patients with MM are ~23 times more likely to have a 22q11.2 deletion than the general population

Searching for MM among patients with 22q11.2DS

- In 1522 patients with 22q11.2DS, 8 had MM
- Patients with 22q11.2DS have a 12.28 to 15.54 times risk of having MM compared to pre-folate and post-folate populations

Finding the gene responsible for MM

- The smallest extent of deletion is C-D, which contains 10 genes
- After analyzing the 10 genes in multiple aspects, researchers found *CRKL* to be the gene most likely responsible for MM.
  - *CRKL* codes for a protein involved in communications inside the cell when triggered by growth factors
  - *CRKL* is needed in neural tube cells in mice and humans
  - In both mice and humans, damaged CRKL proteins fail to do their job in intra-cell communications.

CRKL and folate in relation to neural tube defects

- Mice with a damaged *CRKL* have an increased risk of neural tube defects even if the mother is given folate before pregnancy.
- Mice with a damaged *CRKL* and where the mother is on a low folate diet have a notably higher risk of neural tube defect.

Conclusions

- 22q11.2 deletions greatly increase the risk of MM.
- The increase in the risk of MM may be due to the loss of 1 copy of the *CRKL* gene, which is needed in communications inside cells.
- The increase in the risk of MM in individuals with 22q11.2DS is partially lower if the mother takes folate before pregnancy.

Meningomyelocele

The **neural tube** normally forms the **brain** and **spinal cord**. The two sides of the neural tube usually join together within the first 28 days of pregnancy.

Spina bifida is a birth defect that occurs when the neural tube fails to close completely in the fetus, and the spinal cord and its covering (meninges) forms a sac. The backbone then needs to form around bulge. The incomplete closure of the neural tube and the backbone means that the sac protrudes out of the backbone.

Meningomyelocele (MM; also called myelomeningocele) is the most severe type of spina bifida. In babies with this condition, the liquid-filled sac sticks out from their back. This sac is not covered by skin, so the nerves and tissues are exposed. The affected babies likely have a buildup of spinal fluids on the brain, which must be removed. They also have problems with bladder and bowel functions as well as life-long disabilities on the lower body.

To assess if a fetus may have spina bifida, a blood test and ultrasound imaging can be done during pregnancy. Those with spina bifida may require surgeries before and after birth.

To minimize the chance of spina bifida in a child, it is recommended that women of childbearing age take **folate** supplements daily before becoming pregnant.

Image and info from the Cleveland Clinic

Meet Kendall

Kendall is a resilient artist who has been growing her small business locally across North Bay, Ontario, Canada. Having 22q11.2 deletion syndrome (22q11.2DS) does not keep her from pursuing her passion!

Kendall was diagnosed with 22q11.2DS at age 4. As a young child, she had an open-heart surgery and a palate surgery. Later, she had 3 major back surgeries to correct her curved spine. Although she often had to deal with multiple health issues, including major depression and anxiety attacks, she does not stop when things get hard. Kendall has overcome many challenges and has reached the milestones in her life.

As an adult who loves art, Kendall has been working multiple jobs, trying to get other people to notice her passion. In her business, she offers a great selection of products and services to her community. These includes face-painting, henna designs, and mixed media artwork. She also works as a nanny, an art teacher, and a guitar music publisher. She has been a published musician for a while now and is hoping to get her new album and artwork out there. The next goal that she has set for herself is to publish a book!

Kendall goes above and beyond in what she does, and this is obvious in her involvement with pickleball. She has been playing pickleball for almost 2 years, committing to weekly practices at her local park. She has joined Special Olympics, where she gets to go and compete across Ontario. With her fellow athletes, she showcases what pickleball is about – it is like a four-in-one game that is enjoyable, competitive, yet easy to catch on. She is proud of her collection of rackets and trophies – she has won three times since starting! In addition to playing pickleball, she has been swimming and engaging in other sports with Special Olympics over the years. It is a great way to get herself more active and to meet new people.

When she is not working on her art business or playing sports, she volunteers. She is an active part of many events hosted by her church. In particular, she is part of the Street Ministry, where she helps homeless people in her community. She has also volunteered with the Heart and stroke Foundation and assisted people with special needs.

Kendall often participates in community events. She especially loves appearing as the Dino Girl mascot, and she was even interviewed by the local news in 2022. She uses her talent in photography and publishing to document her projects. She is active in online social networking, particularly involved in the Wave Movement and bringing new community insights to the rest of the world.

Kendall loves sharing her passion with the world doing what she loves to do! She is very grateful to her mom, siblings, and close friends for all their support.

Xanthie Featured in the News in Australia

Despite having to deal with conditions associated with her 22q11.2 duplication syndrome (22q11.2DupS), Xanthie has a smile that brightens everyone's day. Because of her hypotonia (low muscle tone), she has increased flexibility. Her story was featured in 7NEWS in Australia on February 25th:

Adelaide toddler with rare genetic condition that makes her 'ultra-flexible'

Since the publication of the story, Xanthie's family was able to raise enough money to buy an assistive walker that helps her learn to walk.

More recently, in the June 15th edition of the Take 5 Magazine (available in print), Xanthie's mother Mikayla shared more details about the family's journey. While Xanthie's legs could spread out unusually wide, she was not meeting other milestones such as crawling. The pediatrician explained that Xanthie, then age 1, had hypotonia, and he **ran genetic tests to find the cause**. The wait for answers was long, but the family finally found out that Xanthie has 22q11.2DupS. She needed physiotherapy to build muscle strength and speech therapy to help with talking.

Mikayla shared that the National Disability Insurance Scheme (NDIS) initially did not approve their application for funding support, as people with 22q11.2DupS tend to have very mild symptoms. She and her partner Kurtis paid out of pocket for multiple assessments and treatment, and eventually the NDIS application was approved.

Xanthie loves to play with her big brother Arlo, and she is starting to talk more. Mikayla and Kurtis are happy that Xanthie can get the support that she needs to grow and thrive.

Donate Now

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!

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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