International 22q11.2 Foundation

Newsletter December 2023

This year, we celebrated 20 years of changing and improving the lives of individuals with 22q differences. We are especially proud that many of our Medical Advisory Board members played major roles in the publication of three sets of clinical practice recommendations for 22q11.2 deletion syndrome (see p. 5). Our Health Conditions Explained information sheets continue to help families understand various medical conditions (p. 4). We highlight the need for prenatal diagnosis (p. 2-3) as well.

On November 22, we raised awareness for 22q differences by lighting buildings in red (p. 6). We also bring you the story of a parent who explained 22q11.2DS on live TV in Belgium (p.8) and the story of Clara, a courageous young woman with 22q11.2DS (p. 7-8).

On behalf of the International 22q11.2 Foundation, thank you to all our families, friends, and professionals for your continued support. Happy holidays!

18th Annual Faces of Sunshine Calendar

On behalf of the International 22q11.2 Foundation, Inc., it is our pleasure to announce the featured children for each month of our **2024 Calendar**. Many thanks to all who submitted photos. We very much hope you enjoy the calendar as well as all of the beautiful faces of sunshine throughout the New Year! This calendar is a great way to show your 22q pride. Additionally, this item makes a lovely holiday gift; so **order via our website** now to ensure all orders can be filled. All proceeds benefit the work of the International 22q11.2 Foundation, Inc.

January – Teresa M *from San Antonio, Texas*
February – Fallon R *from St. Libory, Nebraska*
March – Jaxson P *from Hudson, North Carolina*
April – Eden P *from San Diego, California*
May – Katie L *from Milltown, Wisconsin*
June – Lee Michael *from Land O Lakes, Florida*
July – Celeste & Alexia M *from Marysville, California*
August – Wilson M *from Katy, Texas*
September – Collins H *from Brecksville, Ohio*
October – Kaylee A *from Benton, Pennsylvania*
November – Kruze S from *Alberta, Canada*
December – Elijah C *from Manteca, California*

...and the ***2024 Cover*** *goes to*
Brie H *from Boonsboro, Maryland*

The Importance of Prenatal Diagnosis of 22q11.2 Deletion Syndrome (22q11.2DS)

22q11.2 deletion syndrome (22q11.2DS) is not a well-known condition, even among healthcare providers. Getting the diagnosis while the affected baby is still in the womb gives them the best chance to receive the critical care they need during and after birth. Here, we present the basics of prenatal diagnosis of 22q11.2DS. For more information, please read Prenatal screening and diagnostic considerations for 22q11.2 microdeletions.

What is prenatal diagnosis for 22q11.2DS?

- 22q11.2DS is a genetic condition that is caused by a missing piece on 1 of the 2 copies of chromosome 22. It affects many body systems throughout life.
- The only way to get a definitive diagnosis is to go through **genetic testing**, which checks the DNA and see if a piece is missing on chromosome 22.
- Experts recommend that diagnosis for 22q11.2DS happens prenatally, which means receiving genetic testing during pregnancy. The genetic testing should be performed if there are clinical indications such as an abnormal ultrasound or an older child with the deletion.

Why is prenatal diagnosis important?

- The family can prepare themselves.
- The family can obtain information relevant to 22q11.2DS and better manage the pregnancy.
- If the baby will be born with complex heart problems or other anomalies, the family can make plans for the mother to give birth in a tertiary hospital with all the necessary support, including a higher-level neonatal (newborn) intensive care unit.
- Once the baby is born, the medical team can be ready to detect and treat 22q11.2-related issues such as feeding problems, heart issues, immune dysfunction, hypocalcemia (low calcium levels in the blood), laryngeal web, etc.
- Unfortunately, many children and adults affected by 22q11.2DS receive care from many healthcare providers for years without understanding the root cause of their health issues. A prenatal diagnosis of 22q11.2DS helps the family avoid this prolonged and confusing diagnostic odyssey.
- The diagnosis allows for timely and coordinated care from a team with specialists from relevant fields. The team can anticipate the necessary care and reduce suffering and risk of death.

How is prenatal diagnosis done for 22q11.2 deletion or duplication?

Chromosomal microarray analysis

- Also called array or **CMA**, it is the recommended test for 22q11.2DS.
- It is an invasive method but can potentially yield a definitive result.
- CMA can check for deletions and duplications in chromosome 22q and elsewhere in the genome. It can also determine the size of the deletion or duplication.
- The test needs a DNA sample from fetus via chorionic villus sampling (CVS) at 10 to 13 weeks of pregnancy OR amniocentesis at 15 weeks.
- For more info about CMA, see this video by Myhealth.Alberta.ca or the website of Dalglish Family 22q Clinic.
- Older methods such as multiplex ligation-dependent probe amplification (MLPA) or Fluorescence in situ hybridization (FISH) can miss certain deletions in 22q11.2 area, and cannot detect changes elsewhere in the genome.

Noninvasive Prenatal Screening (NIPS)

- NIPS that screens for specific deletions (including 22q11.2) is available
- The American College of Medical Genetics and Genomics (ACMG) issued a conditional recommendation for **NIPS** for 22q11.2DS to be offered. [summary]
- The test should be performed after 9 to 10 weeks of pregnancy. It needs the mother's blood sample, which contains DNA from the fetus.
- **NIPS is not a diagnostic test**. It can detect some of the deletions, but not all.

Wanted: Expanded Access for Prenatal Diagnosis of 22q11.2DS

Prenatal diagnosis of 22q11.2DS can provide the much-needed anticipatory care for affected babies and support for their families, and 22q experts have been lobbying for expanded access for prenatal diagnosis for years. In January, 2012, our Foundation's Board Member Donna McDonald-McGinn and Chair of the Medical Advisory Board, Dr. Anne Bassett were part of a group that presented before the US Secretary of Health's Committee on Heritable Disorders and Newborn Screening to rally support for newborn screening for 22q11.2.

Unfortunately, as of December 2023,
general access to prenatal screening of 22q11.2DS is still NOT available
in the Uniform Screening Panel in USA or anywhere in the world.

Let's ask for it.

Advocating for expanded access to early detection of 22q11.2DS

Dr. Kecia Gaither, the Director of Perinatal Services and Maternal Fetal Medicine at NYC Health + Hospitals/Lincoln in the Bronx, NY, authored an opinion piece in RealClearHealth to explain how limited access to prenatal screenings contributes to maternal health inequities in Black and Hispanic communities.

Dr. Gaither calls on the American College of Obstetricians and Gynecologists (ACOG) to update their guidelines to formally recommend increased access to prenatal screenings for common conditions like 22q11.2 deletion syndrome (22q11.2DS) in order to produce better health outcomes for families. Please click on the link on the right to read Dr. Gaither's full article.

Read the full article

A new grassroots petition urges the American College of Obstetricians and Gynecologists to recommend 22q11.2DS screening for all pregnancies

The International 22q11.2 Foundation is elevating the voices of patients and families by organizing a grassroots signature petition that urges health care leaders to expand access to 22q11.2 deletion syndrome (22q11.2DS) screenings. These screenings, which are approved for all 22q11.2 deletions and, in some cases, suspected duplications, allow families the chance to deliver their baby in a center with high level neonatal care best equipped to offer comprehensive evaluations and appropriate interventions prior to birth hospital discharge. Please click on the link on the right to sign the petition to support 22q11.2DS screening for all pregnancies!

Sign the petition

Gastrointestinal (GI) Series

Information sheets on selected topics now available on our website

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

Our newest series on the "**Health Conditions Explained**" section of our website focusses on gastrointestinal (GI) issues. Special thanks to Dr. Maria Mascarenhas, Pediatric Gastroenterologist and Nutrition Pediatrician; Joanne Loo, Programmatic and Educational Tool Developer; and Dr. David W. Low, Plastic Surgeon and Medical Illustrator.

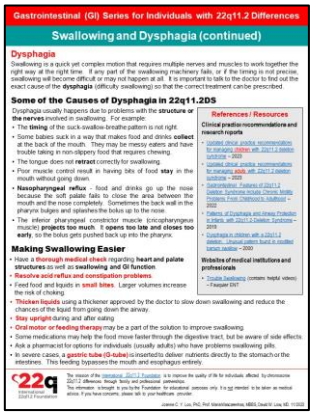
We have so far published information sheets on 4 GI-related topics:

- **The Digestive System**
- **Feeding Difficulties**
- **Swallowing and Dysphagia**
- **Constipation**


Gastrointestinal (GI) Series for Individuals with 22q11.2 Differences

Swallowing and Dysphagia

Four stages of swallowing


Gastrointestinal (GI) Series for Individuals with 22q11.2 Differences

Swallowing and Dysphagia (continued)

We are currently developing information sheets on additional topics in the GI Series. We are also planning to develop a Speech Series in the near future.

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 concerns, please talk to your healthcare provider.

Health Conditions Explained

The topics published previously are:

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)

22q GLOSSARY

- Second Edition (217 entries)

Clinical Recommendations and Resources for Conditions Related to 22q11.2 Differences

22q11.2 Deletion Syndrome (22q11.2DS)

22q11.2DS is caused by a missing piece on chromosome 22q. It used to be known by other names such as DiGeorge syndrome & velocardiofacial syndrome. This genetic condition happens in about 1 in every 2148 babies and usually arises as a new event in a family. The affected individual has 1 copy (instead of 2) of several of the genes that usually tell the body how to form and function during early development and throughout life. Each person (even if in the same family) can have their own pattern of 22q11.2DS features and severity of each.

Clinical recommendations (all published in 2023)

- 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

Subspecialty guidelines (published in 2019)

- **Speech-Language Disorders** in 22q11.2 Deletion Syndrome: Best Practices for Diagnosis and Management

Common Features of 22q11.2DS

- Birth defects (e.g. heart, palate, & kidney problems)
- Low calcium
- Difficulty fighting infections
- Feeding and swallowing differences
- Seizures
- Developmental delay - particularly in speech
- Learning differences
- Behavioral problems
- Other differences

22q11.2 Duplication Syndrome (22q11.2DupS)

22q11.2DupS is caused by a extra piece on chromosome 22q. This genetic condition happens in about 1 in every 4000 babies and usually runs in the family. The affected individual has 3 copies (instead of 2) of several of the genes that usually tell the body how to form and function during early development and throughout life. Each person (even if in the same family) can have their own pattern of 22q11.2DupS features and severity of each. There are currently no official clinical recommendations for this genetic condition. Here are some research reports that may be helpful.

- **Neurodevelopmental** functioning in probands and non-proband carriers of 22q11.2 microduplication – 2022
- 22q11.2 duplications: Expanding the **clinical presentation** – 2021
- **Cardiac evaluation** of patients with 22q11.2 duplication syndrome – 2021
- **Immunodeficiency** in 22q11.2 duplication syndrome – 2021
- 22q11.2 duplication syndrome: elevated rate of **autism spectrum disorder** and need for medical screening – 2016

Common Features of 22q11.2DupS

- Birth defects (e.g. heart, palate)
- Vision and ear problems
- Difficulty fighting infection
- Developmental delay
- Learning problems
- Autism
- ADHD
- Other differences

We Lit up the Night for 22q!

Here are some of the buildings that were lit up in **red** to raise awareness for 22q11.2 differences on November 22.

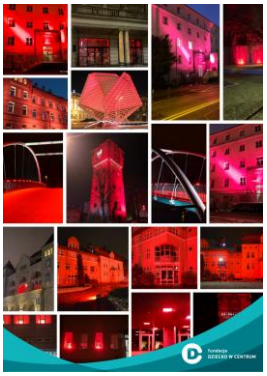
CN Tower
Toronto, ON, Canada

Children's Hospital of Philadelphia
Philadelphia, PA, USA

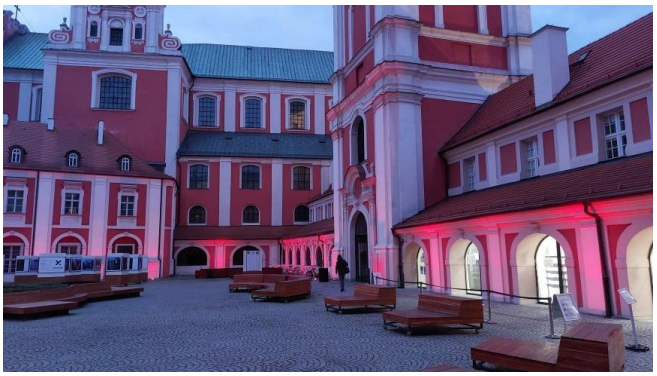
Havenhuis Antwerpen (Harbor House)
Antwerp, Belgium

Materialise
Leuven, Belgium

Institute of Mother and Child
Warsaw, Poland



Multiple buildings
Pila, Poland



City office building at Kolegiacki Square
Poznań, Poland

Meet Clara

(posted on November 2nd, 2023)

Hi! My name is Clara Von Feldt, and I grew up with 22q deletion syndrome also known as velo-cardio facial syndrome. I was born and raised in Minnetonka, Minnesota and I am currently 22 years old. Overall, I had a very happy childhood. My parents made it a goal for me to be able to succeed as an adult from the moment I was diagnosed with 22q. They allowed me to be "normal" (even though we don't really know what to consider as "normal"). My family includes 3 sisters, 2 pups, my mom and dad, plus a large extended family. Having a big family really helped me increase my social and emotional skills! My mom and dad were my biggest cheerleaders throughout my 22q affiliated life. They were the most supportive people when it came to school/sports/friendships. I could always come back to them and ask for help and encouragement. The only trouble I had was being compared to my siblings where my parents would say "oh you're not ready for that", and "maybe next time/year". This made me feel frustrated and I started some activities at a slower pace than I wanted to. All in all, I had a great upbringing while living with 22q deletion syndrome.

I was diagnosed with 22q at 6 weeks of age when I was in the hospital for failure to thrive due to issues with my ability to take in an adequate amount of calories. While at the hospital they discovered that I only had one kidney and I had a hole in my heart (which eventually closed). Due to my feeding issues, I had a feeding tube until around nine months. My main health issues have been related to my ears. I have had recurring ear infections, and my hearing was continuing to decline. Recently I had my left eardrum reconstructed and that has been a very exciting change! As a result of such an early diagnosis, I was able to receive therapy in areas such as speech, occupational and physical therapy from a very young age. I believe this made a big difference in who I am today.

Along with my 22q diagnosis I have learning disabilities which affects my time in school greatly. As long as I can remember I've had an IEP (Individualized Education Plan) which has helped me enormously even today! The only hard thing about having 22q and being in school was worrying what my peers thought about me. I went to a great school which had lots of disability services. My accommodations were extra help (para) in classrooms, extended time on tests, extended time on assignments, note cards for exams, limited questions on homework and some extra help in specific subjects. Being a student in special education made things harder for me to make friends. I did get bullied (I won't lie about that) but being in a school where bullying was taken seriously and where when I talked to a teacher about it, it stopped, helped a lot. Overall school was a positive experience for me, and I encourage all families to have their children go to a school that has the resources needed for your child with 22q differences!

Well to start off there are little accomplishments I had like the milestones' children have (crawling, walking, talking etc..) and I passed everyone with flying colors! But I would have to say my biggest accomplishment was to graduate from high school, it was 12 years of dedication and hard work. I also received a few college scholarships through my high school, which was a real honor. I was very fixated on getting a job, so after convincing my parents I was ready, I started working at a local grocery store at the age of 14. It was during this time that I realized my true love was working with children and that is what I have done ever since. I also enjoy music and have been playing the piano for about 15 years. For a long time, I was self-conscious about my voice, so a few years ago I started taking voice lessons and singing in front of an audience. It has been both challenging and rewarding and has given me a lot more confidence. I also have a passion for theatre and have been in a few plays as well as a part of many costume crews.

Meet Clara (Continued)

If a doctor says it, doesn't mean it's true! Prove them wrong. You can do so much more than what they are telling you. Believe in your dreams and pursue them! My mom was once told by a physical therapist that I would never be able to walk or talk but here I am today doing so much more. Yes, doctors know a lot but there's also a lot they don't know as well!

My plan for the future is to become a 1st/2nd grade teacher because I love working with kids due to the way it makes me feel inside! It brings me so much joy to know that I CAN be the person who helps a child become someone in this world no matter who they are. I would also like the opportunity to raise a family with a romantic soulmate!

I am in college now finishing up my junior year. College has been a struggle so far due to the fact I graduated from high school during the peak of COVID. One thing that has helped me find success in college has been working with the school's student accessibility services. Some professors in college don't understand why students with disabilities need extra help, so it's great to get support from the people that do understand the struggles disabled people go through with learning. It has been hard to make friends with my social skills not being 100% (but who has social skills at 100% – no one that I know of). I have a close group of friends that I hang out with, and we've known each other since elementary school but I have had a hard time expanding beyond that group and making new friends. I keep getting involved in things I love, and I know that is how I will grow my circle.

The International 22q11.2 Foundation would love to hear personal stories about friends and family persevering and thriving with 22q! Because everybody has those days when you need something inspiring, something that gives us hope. If you know someone, or would like to share your own story, we want to hear from you!

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 look forward to hearing from you!

Read more inspirational stories on

https://22q.org/inspirational-stories/

A Parent Explains 22q11.2DS on National Television in Belgium

Liv is a happy and fun 2-year-old girl who happens to have 22q11.2 deletion syndrome. She had already faced a lot of health issues such as surgery for a congenital heart defect, and major infections.

Liv's dad, Thomas Smolders, talked about life with 22q11 and raising awareness in the program "de Tafel van Gert" (Gert's Table). This TV program airs during prime time on national television in Belgium, so it is a great channel to tell many people about the syndrome.

In the interview, Thomas mentioned Professor Ann Swillen, an educational psychologist from the Center for Human Genetics in Leuven (UZ Leuven) who has been providing multidisciplinary care for patients with 22q11.2DS and doing research on this condition since 1994.

Thank you, Thomas, for your testimony and for spreading awareness for 22q11.2 deletion syndrome!

This 8 min 54 sec interview (in Dutch/Flemish, **with English subtitles**) can be viewed on Facebook page of Vecarfa 22q11ds Vlaanderen.

https://www.facebook.com/Vecarfa/videos/717905553779992/

The Honor of your absence is requested by **The International 22q11.2 Foundation** at our

No Show Valentine's Ball

on Wednesday the fourteenth of February 2024 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

Donate here

The International 22q11.2 Foundation

Donation Menu

• Table of 10 friends I do not have to persuade to attend!	$5222.00 USD
• Providing this gift in lieu of his & her new outfits!	$2222.00 USD
• Please accept this donation in place of raffle tickets!	$1222.00 USD
• It is not that I don't enjoy eating out!	$522.00 USD
• Saving on a Valentine's Day babysitter and Uber.	$222.00 USD
• Home before midnight – priceless!	$122.00 USD
• Cozy by the fire! – I'll take that any day of the week!	$52.00 USD
• Helping families in need!	$22.00 USD

22q at the Zoo Worldwide Awareness Day

Sunday May 19th, 2024

22q at the Zoo will kick off in Sydney, Australia on **Sunday, May 19th, 2024** and continue westward at zoos around the world, giving families, friends and professionals a chance to socialize, network and raise public awareness of 22q11.2 syndromes. Please **save the date**. More info to follow.

Developed by the International 22q11.2 Foundation, volunteers spread awareness at their local or regional zoo, park, playground or aquarium by handing out 22q Fact Sheets and educating guests and zoo visitors. Participants are recognizable by wearing official "22q at the Zoo" red T-shirts and "Ask Me About 22q" buttons.

If there were no 22q at the Zoo events in your area in 2023, you can host one in 2024! Please visit the Host a 22q at the Zoo Event section on our website for more information. We even have an event planner letter ready for you! No zoo? No problem. Consider gathering at a park, a farm, or even an aquarium.

If you have any questions, please email us: info@22q.org. Thank you.

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!

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**!

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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