



International  
22q11.2 Foundation

# Newsletter June 2025



22q at the Zoo 2025 at San Rossore Park, Pisa, Italy.

## Celebrating “22q at the Zoo—Worldwide Awareness Day

The International 22q11.2 Foundation extends heartfelt thanks to everyone for once again hosting “[22q at the Zoo—Worldwide Awareness Day](#)” in communities—big, small, and everything in between—around the world!

This year marked the 15<sup>th</sup> anniversary of this extraordinary global awareness and friendship-building event, brought to you by the International 22q11.2 Foundation ([22q.org](#)) in partnership with our incredible colleagues from family support organizations and 22q Centers of Excellence across the globe. We appreciate each and every one of you, including parents, grandparents, caregivers, relatives, friends, healthcare providers, and volunteers who devoted their time to make this special event a reality.

We are particularly thankful for the children and adults living with 22q11.2 differences, along with their siblings, parents, relatives, and friends, who, in some areas, faced challenging weather to attend the zoo (or park, aquarium, backyard, or playground) for 22q! Our heartfelt thoughts go out to those in middle America whose zoo day was affected by terrifying tornadoes. We know they were with us in spirit and look forward to celebrating with them in 2026.

We also deeply appreciate the in-kind donations from [Tolerance Bio](#) and the [22q and You Center](#) at the Children’s Hospital of Philadelphia, which helped make this year’s collaborative event truly fantastic!

WE DEFINITELY WENT TO THE ZOO FOR 22q... AND WE COULDN’T HAVE DONE IT WITHOUT YOU!!!

Check out the zoo day photos in this newsletter, and visit our social media sites for more!



We look forward to seeing you again next year on **Sunday May 17<sup>th</sup>, 2026!**



# 22q at the Zoo - Worldwide Awareness Day

A small selection of the photos from May 18<sup>th</sup>, 2025





## 22q at the Zoo (Photos Continued)



Visit our social media sites  
for **additional photos** from  
22q at the Zoo



See you next time on  
**May 17<sup>th</sup>, 2026!**



# Donna McDonald-McGinn Celebrates 40 Years at the Children's Hospital of Philadelphia



Congratulations to our Foundation's Founding Board Member, Donna McDonald-McGinn, MS, CGC, who recently celebrated her 40<sup>th</sup> anniversary at the Children's Hospital of Philadelphia (CHOP)!

Donna first arrived at CHOP as a Genetic Counseling student in 1983 and officially joined the Division of Human Genetics in 1985. Shortly after, she founded the [22q and You Center](#), a pioneering holistic care program for individuals and families affected by chromosome 22q11.2 differences, a program that continues to be a trailblazer in the field.

Donna has always supported education, multidisciplinary care, and collaborative research involving 22q11.2 deletion syndrome. She has published more than 300 research reports and book chapters. In 2003, she was part of the team that started the International 22q11.2 Deletion Syndrome Foundation, and soon she started to lead international initiatives including establishing clinical practice guidelines. All through her career, she shares her knowledge and inspires future chromosome 22q11.2 experts.

**Thank you, Donna, and congratulations!**

## Celebrating our Foundation's 22<sup>nd</sup> Anniversary!



Our Foundation is celebrating its 22<sup>nd</sup> anniversary! We invite you to join our **Around the World with 22q Gala** on Saturday September 6<sup>th</sup>, 2025.

Please save the date. More information will be available on our social media sites soon.



# Healthcare Costs for Adults with 22q11.2DS

[Young adults with a 22q11.2 microdeletion and the cost of aging with complexity in a population-based context.](#) Malecki SL, Heung T, Wodchis WP, Saskin R, Palma L, Verma AA, Bassett AS. Genetics in Medicine 26:101088, 2024. PMID: 38310401

It is common for individuals with 22q11.2 deletion syndrome (22q11.2DS) to have **multimorbidity**, which means they deal with two or more long-term health issues that need treatment. In particular, adults with 22q11.2DS have to deal with conditions that they have been born with as well as those that they develop as they grow up. The question is: **What are the healthcare cost and resource use for adults with 22q11.2DS?**

## Setting

- Place: Ontario, Canada – where healthcare services are covered by a universal healthcare system ([OHIP](#))
- Time period: April 2002 to March 2020

## Study Population

- 365 adults with 22q11.2DS who were followed at a specialty clinic in Toronto
- For each adult with 22q11.2DS, 10 controls with matching birthdate, sex, and socioeconomic status were selected from the 15 million people in the general population. There were 3650 matched controls in this study.

## Methods

- Analyzed 18 years of health administrative data
- Compared the use of the healthcare system by adults with 22q11.2DS vs. controls from the general population
- Summed up costs covered by OHIP. These include (but are not limited to):
  - In-hospital care
  - Visits to doctors
  - Laboratory tests (blood, urine etc.)
- Cost is standardized (adjusted for inflation) to Canadian dollars in 2019

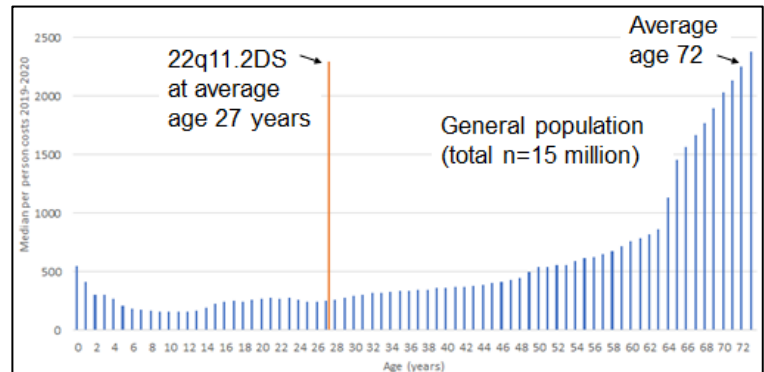


Figure above: Health care costs for 15 million people in Ontario, Canada, shown for each age from 0 to 73 years (blue bars), and the health care costs for persons with 22q11.2DS (orange bar) at the average age of 27 years. (Adapted from Figure 2 of the research article)

## Main Findings

- Only ~2% of adults with 22q11.2DS in the study had no multimorbidity, vs. ~41% of controls.
- The average healthcare costs for adults with 22q11.2DS were over **8 times higher** than those for the controls.
- Healthcare costs at the average age of **27** years for adults with 22q11.2DS were about the same as those for individuals who were **72** years old in the entire general population (see figure above).
- The likelihood of being within the top 1<sup>st</sup> percentile of healthcare costs for the entire Ontario population was nearly **18 times greater** for adults with 22q11.2DS.
- Healthcare costs were higher in adults with 22q11.2DS than in controls **across all healthcare sectors** analyzed in this study.
- Predictors of higher healthcare costs were having **22q11.2DS**, being **>45 years** old, and having a serious disorder that involves the **brain** (including **mental health**).

This study shows the high healthcare costs of adults with 22q11.2DS, which reflects their substantial medical complexities and care needs.

# Sleep Study Findings in Children with 22q Differences

[Polysomnographic findings in children with 22q deletion & duplication syndrome: relationship to genetic diagnosis, parent-reported symptoms, and calcium levels](#)

Ingram DG, Tracy M, Noel-MacDonnell J, Ghulmiyyah JM, Arganbright JM.

Sleep Breath. 2024 Mar;28(1):489-494. doi: 10.1007/s11325-023-02897-y. Epub 2023 Aug 1. PMID: 37526804.

**Polysomnography (sleep study)** is a diagnostic test that examines a patient's body systems as he/she sleeps. During the sleep study (at home or in a sleep lab), the equipment monitors the heart, lungs, brain activity, movements, and oxygen levels. It also assesses if the sleep involves the expected stages and duration.

Sleep problems have been reported in individuals with 22q11.2 differences, and polysomnography has been recommended for both [children](#) and [adults](#) with 22q11.2 deletion syndrome (22q11.2DS). In this study, the researchers ask:

1. Do sleep study results of children **match the sleep-related symptoms reported by their parents?**
2. Are sleep study results related to:
  - **Genetic diagnosis** (22q11.2 deletion syndrome (22q11.2DS) or 22q11.2 duplication syndrome (22q11.2DupS))
  - **Calcium** level in the blood
  - **Ferritin** level in the blood (which relates to iron levels in the body)

## Setting

- Missouri, USA, from 2015 to 2021

## Study Population

- 60 children with either 22q11.2DS or 22q11.2DupS who were followed at a specialty clinic in Kansas City, MO

## Methods

- Review of medical records of children who have undergone polysomnography and whose parents have completed the Childhood Sleep Habits Questionnaire (CSHQ) involving symptoms like snoring, stoppage of breathing, snorting/gasping



Left: A child being prepared for a polysomnogram by a respiratory therapist

[https://commons.wikimedia.org/wiki/File:Pediatric\\_polysomnogram.jpg](https://commons.wikimedia.org/wiki/File:Pediatric_polysomnogram.jpg)

## Main Findings

- Polysomnography results showed that 76.4% of children had **obstructive sleep apnea** (upper airway becomes blocked during sleep and the person cannot breathe properly). Other sleep problems included central sleep apnea, low oxygen in the blood, periodic limb movement etc.
- Comparing 22q11.2DS vs 22q11.2DupS:
  - No significant differences in the rate of obstructive sleep apnea
  - More children who have 22q11.2DS had periodic limb movement compared to children with 22q11.2DupS. However, this difference was in fact due to the low calcium levels in many children with 22q11.2DS.
  - Children with 22q11.2DupS had more minor differences in the duration of sleep stages compared to those with 22q11.2DS
- Comparing polysomnographic results vs. parent survey: **Parent-reported symptoms do not predict the presence or severity of obstructive sleep apnea**
- **Low calcium levels were associated with periodic limb movements**
- Ferritin levels were not associated with periodic limb movements

Sleep studies may be needed to check all children with 22q differences. For those with periodic limb movements during sleep, checking calcium levels may be helpful.

**Check out our new [Sleep Series](#) in [Health Conditions Explained](#)!**



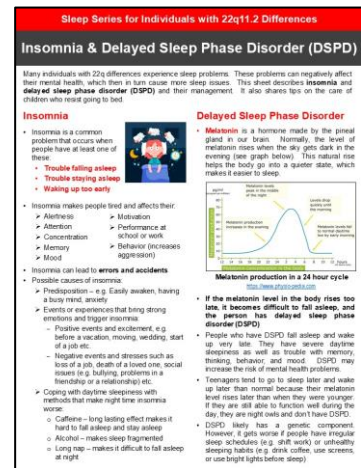
# Now Available: Sleep Series

## Information sheets now available on our website

We are happy to let you know that the **Sleep Series** is now available in the "[Health Conditions Explained](#)" section of our website.

Sleep is very important for health, but it is an area that is sometimes overlooked. Our Sleep Series provides background information on sleep conditions that are common in individuals with 22q differences and offer some tips on possible management. Five topics are available. Special thanks to Joanne Loo, Programmatic and Educational Tool Developer; Dr. David Ingram, Pediatric Sleep Physician; and Dr. Jill Arganbright, Pediatric Otolaryngologist (Ear-Nose-Throat specialist).

- [Introduction to Sleep](#)
- [Obstructive Sleep Apnea \(OSA\)](#)
- [Insomnia & Delayed Sleep Phase Disorder \(DSPD\)](#)
- [Restless Legs Syndrome \(RLS\) & Periodic Limb Movement Disorder \(PLMD\)](#)
- [Parasomnias](#) (focusing on Sleep Terrors, Confusional Arousals, and Sleep Walking)



## Recently Published: The Immune System Series

- [The Immune System](#)
- [Vaccination](#)
- [Immune Deficiency Related to T Cells](#)
- [Immune Deficiency Related to B Cells](#)
- [Autoimmune Disorders](#)
- [Allergies and Asthma](#)
- [Immune System Issues in 22q11.2DS](#)
- [Immune System Issues in 22q11.2DupS](#)

Be sure to check out all the topics in the "[Health Conditions Explained](#)" section of our website!

- **Heart Series**
- **Mental Health Series**
- **Gastrointestinal (GI) Series**
- **Dental Series**
- **Speech Series**
- **Immune System Series**
- **Palate Series**
- **22q Glossary (3<sup>rd</sup> ed.)**
- **Sleep Series**

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.

# About Measles



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- **Measles is a serious respiratory infection caused by a virus**
- Symptoms include:
  - High fever
  - Coughing
  - Aches and pains
  - Red eyes
  - Runny nose
  - Rash
- Complications include:
  - Ear infections
  - Deafness
  - Pneumonia
  - Impact to the immune system – having to re-learn how to fight bacteria and viruses
  - Swelling of the brain, often resulting in permanent brain damage, in 1 in 1000 people
  - Lung and neurologic problems in 3 in 1000 people
  - Degenerative nervous system disease 7 to 10 years following infection
  - Death
- Measles **spreads very easily** through coughing and sneezing
  - 9 out of 10 people who meet someone with measles will become infected
  - The measles virus can stay in the air for 2 hours
- **Measles is preventable through vaccination (MMR or MMRV)**
  - Two doses of the MMR vaccine are ~97% effective at preventing measles
  - One dose of the MMR vaccine is ~93% effective
  - **MMR and MMRV vaccines are safe and effective**
  - Vaccination schedule
    - [\[Travelling internationally\]](#) | [\[Not Travelling\]](#)
- For individuals who are **immune compromised**:
  - **Four studies confirmed the safety of the MMR vaccine for individuals with 22q11.2DS.**
  - Please consider consulting with your healthcare provider if you or your child has not been vaccinated against measles

## For additional information about measles please visit:

- Centers for Disease Control and Prevention (CDC) – [Measles \(Rubeola\)](#)
- Canadian Paediatric Society (CPS) – [Measles](#)

## Research reports about the MMR vaccines and 22q11.2DS:

[Vacinas Vivas em Crianças com Síndrome de DiGeorge/ Deleção 22q11.2](#)

(Portuguese)

[Live Vaccine in Children with DiGeorge/22q11.2 Deletion Syndrome].

Miranda M, Martins AT, Carvalho S, Serra-Caetano A, Esteves I, Marques JG. Acta Med Port. 2019 Aug 1;32(7-8):514-519. doi: 10.20344/amp.9802.

[Live vaccine use and safety in DiGeorge syndrome.](#)

Hofstetter AM, Jakob K, Klein NP, Dekker CL, Edwards KM, Halsey NA, Baxter R, Williams SE, Graham PL 3rd, LaRussa P. Pediatrics. 2014 Apr;133(4):e946-54. doi: 10.1542/peds.2013-0831.

[Live viral vaccines in patients with partial DiGeorge syndrome: clinical experience and cellular immunity.](#)

Moylett EH, Wasan AN, Noroski LM, Shearer WT. Clin Immunol. 2004;112(1):106-112. doi: 10.1016/j.clim.2004.02.008

[Safety of live viral vaccines in patients with chromosome 22q11.2 deletion syndrome \(DiGeorge syndrome/velocardiofacial syndrome\).](#)

Perez EE, Bokszczanin A, McDonald-McGinn D, Zackai EH, Sullivan KE. Pediatrics. 2003 Oct;112(4):e325. doi: 10.1542/peds.112.4.e325.

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# Kim van Bekkum Shared Her Experience

Kim van Bekkum is the Founder and Director of [Stichting Steun 22q11](#) in the Netherlands. She started this organization to provide support and information to affected families and to raise awareness for those who are affected by 22q11.2 deletion syndrome (22q11.2DS). Her determination stems from the difficult times after she gave birth to her son, Mika. Back then (in the early 2000s), healthcare providers did not know much about 22q11.2DS, and she had to figure things out by herself.



In early April, Kim shared her experience in the lifestyle section of RTL Nieuws, a Dutch news service. She talked about her pregnancy, the birth, the surgeries, and the traumatizing experience of almost losing Mika.

You can read her story in **Dutch** on:

[\*\*Kims zoon wordt nooit meer beter: 'Langzaam stikte hij in zijn eigen slijm'\*\*](#)

You can use your browser to translate the Dutch text into **English**:

[\*\*Kim's son will never get better: 'He slowly choked on his own mucus'\*\*](#)

We thank Kim for sharing her story and raising awareness for 22q11.2DS!

## 2025 Lighting Request Letter Now Available

### Let's raise awareness for 22q differences!

We invite you and your town to join an international movement of lighting up buildings and monuments in red on the evening of **November 22<sup>nd</sup> (22/11; or 11/22 in North America)** as a play on the name of the 22q11.2 deletion and duplication syndromes.

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 Saturday, November 22<sup>nd</sup>, 2025**. Click on image of the letter on the right to access the pdf file of the letter. **Download** the file to your own drive or cloud. Fill in the name of the building and the city or town, then email or mail the letter to the government or organization that manages the building.

On the night of **November 22<sup>nd</sup>, 2025**, take pictures of yourself with the illuminated building and share them on social media.

**Together, we will light up the night for 22q!**



[\*\*Lighting Request Letter\*\*](#)

# Seeking Identical Twins for Research

Dr. Beata Nowakowska, a geneticist and 22q11 specialist, and her research team are conducting a study to better understand how this syndrome works at the genetic level. They are currently looking for **identical (monozygotic) twins** (of any age) diagnosed with a **22q11 deletion or duplication** to take part in this innovative study.

## Why identical twins?

Because they share the same DNA, yet often show different symptoms. This fascinating difference offers a unique opportunity to explore how genetic and epigenetic factors influence the syndrome.

For more information, contact: **Dr. Beata Nowakowska** – [beata.nowakowska@imid.med.pl](mailto:beata.nowakowska@imid.med.pl)  
You can see her profile on the [website of the 22q11.2 Society](#).

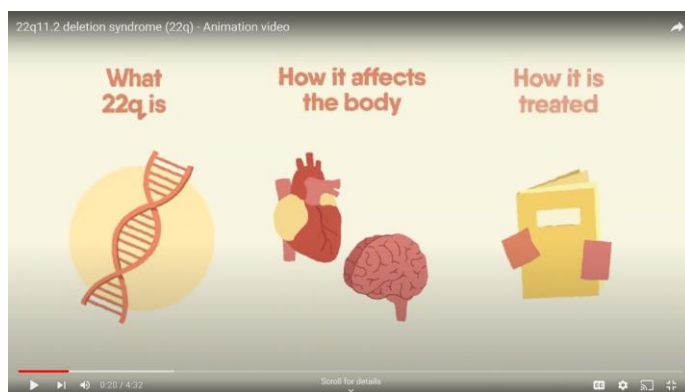
Please feel free to **share this message** with your networks, support groups, friends, and any families affected by 22q11. **The deadline to participate is August 2025.**

Together, we can make a impact on the future of 22q11 research! Thank you for your support.



# Animation video for Adults with 22q11.2DS and Their Families

Dr. Anne Bassett and the team at the [Dalglish Family 22q Clinic](#) previously worked with a Masters student in Biomedical Communications at the University of Toronto (Emily Tjan) to create an educational tool that was designed to help patients and caregivers better understand 22q. In the process, they asked the families for input in terms of content and the type of media that we should use. Ultimately, Emily created a [short animation video introducing 22q](#), and Lisa Palmer, the clinic's Social Worker, presented this at the International 22q11.2 Conference in July 2024. They have now added subtitles and uploaded the video to YouTube, so it's available to all!



Dr. Bassett and her team would love to know what you think. Please:

- [watch the video](#) and
- [complete the short survey](#).

Thank you.





# 22q Resources



**22q Overview:** <https://22q.org/what-is-22q/>

## Family Support and Inspiration

**Family Support Network:** <https://22q.org/resources/for-families/family-support-network/>

**Other 22q Organizations:** <https://22q.org/resources/for-families/other-22q-organizations/>

**Book Lists:** <https://22q.org/resources/for-families/book-lists/>

**Inspiration:** <https://22q.org/inspirational-stories/>

## Healthcare

**Symptoms and care:** <https://22q.org/symptoms-care/>

**Clinical Practice Recommendations:** (Each available in multiple languages; Checklist available in English)

**Adults** (recommendations + checklist): <https://22q.org/symptoms-care/adult-guidelines-checklist/>

**Children and Youth** (recommendations + checklist): <https://22q.org/symptoms-care/pediatric-guidelines-checklist/>

**Health Conditions Explained and 22q Glossary:** <https://22q.org/symptoms-care/health-conditions-explained/>

**Growth Charts:** <https://22q.org/symptoms-care/growth-charts/>

## Education / Development / Life Planning

**Educational Resources for Parents:** <https://22q.org/resources/for-families/school-and-learning/>

**Developmental Skills:** <https://22q.org/symptoms-care/developmental-skills/>

**Behavioral and Mental Health Resources:** <https://22q.org/resources/for-families/behavior-and-mental-health/>

**Life Planning and Legal Support:** <https://22q.org/resources/for-families/life-planning-and-legal-support/>

## Clinics

**USA:** <https://22q.org/resources/for-families/medical-clinics-usa/>

**International:** <https://22q.org/resources/for-families/medical-clinics-international/>

## Awareness Campaigns

**22q at the Zoo Worldwide Awareness Day (Annual Event):** <https://22q.org/get-involved/22q-at-the-zoo/>

**22q Awareness Month (November):** <https://22q.org/get-involved/november-is-22q-awareness-month/>

## Connect with us

Sign up for our **newsletter** and stay up-to-date on the latest research news and events.

<https://22q.org/news/newsletter/>

For more information, please visit our **website** at [www.22q.org](http://www.22q.org)

or **email** us at [info@22q.org](mailto:info@22q.org).



# 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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PO Box 532, Matawan, NJ 07747

**www.22q.org** (877) 739-1849 **info@22q.org**