

# 22q & Friends



The Dalglish Family  
22q Clinic



## 2021 Newsletter

### Hello and Happy Reading!

2021 has been an extraordinary year for all of us. Our Dalglish Family 22q Clinic has continued to care for patients and their families through in-person and virtual appointments through all the ups and downs we have faced with the pandemic.

In November, we raised awareness for 22q by putting an infographic on hospital computers and even lighting the CN Tower in red! You can read about these exciting initiatives on page 7. We published a landmark study about how common 22q is in newborn babies. You can read about our studies on pages 10-11.

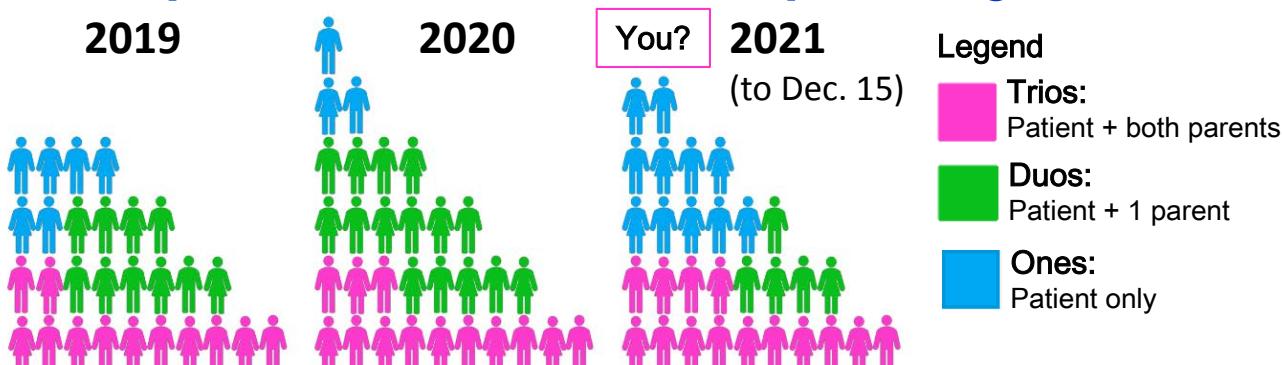
We will tell you how our team members have been involved in the 22q community locally and internationally. We also bring you lots of useful information. Happy Reading!



Our Clinic staff wore orange to observe the National Day for Truth and Reconciliation on Thursday September 30<sup>th</sup>, 2021.

Get a digital copy of this newsletter on:  
<https://22q.ca/aboutus/our-newsletter/>

### New patients seen in the past 3 years



Our 22q clinic is referred an average of 30 new individuals every year. All our **new referrals** (i.e. patient and their family members) are invited to participate in our 22q research study. We encourage **all** to participate, because together, we are making new discoveries. New findings for 22q help us help individuals with 22q and their families here and around the world.

In 2021, a total of 14 trios (patient + both parents) have joined our research family! If you and/or your parents have not yet signed up, what are you waiting for? Get in touch with us to find out more! We aim to make it as convenient for you as possible. You are making a difference for those with 22q and their families!

# Vaccination Against COVID-19



The COVID-19 pandemic has brought many worries and inconveniences, but the availability of vaccines has helped control the spread of the virus significantly.

All vaccines approved for use in Canada give excellent protection against the COVID-19 infection. **None of these contain any live virus. They cannot cause disease.** And they are safe for people with immune deficiencies, just as they are for everyone else in the general population. In fact, they are **especially necessary for people with 22q.** Individuals who have recovered from a COVID-19 infection should still receive the COVID-19 vaccine. If you have any questions about these vaccines, please talk to your doctor or healthcare provider.

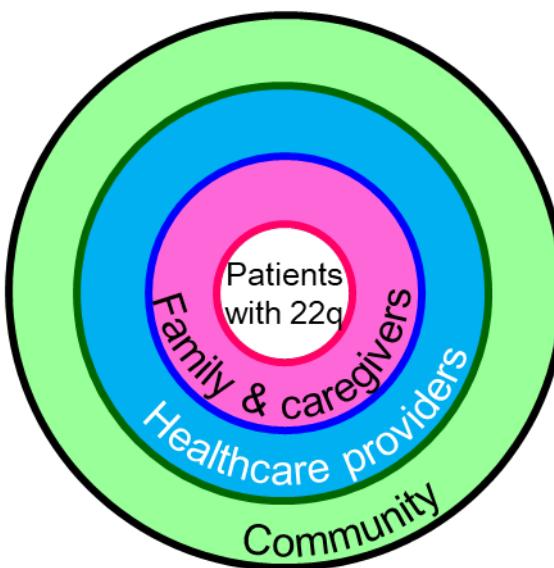
## The Dalglish Family 22q Clinic's Recommendations for Individuals who are eligible to receive COVID-19 vaccines:

All **individuals with 22q** should **receive the vaccine** - the two doses protect very well against the worst effects of the virus, including the Delta variant. Three doses appear to be needed for the Omicron variant.

All **close contacts** (e.g., **immediate family members**) of individuals with 22q should also **receive the vaccine**, in order to protect the person in the family likely to be most in danger of getting very sick from the virus (that is, the person with 22q).

## Rings of protection

The more rings of protection that are in place (people fully vaccinated against COVID-19), the harder it is for the virus to infect you.



## A third dose of the COVID-19 vaccine

A **third dose** is now available to individuals with moderate to severe primary immunodeficiency (e.g. DiGeorge syndrome) **at least 2 months (56 days)** after the second dose. "DiGeorge syndrome" is an older name for 22q11.2 deletion syndrome (22q). For patients with 22q, before you go for your third dose, you may need a **referral form or letter from your doctor or our Clinic.**

As of December 13<sup>th</sup>, **adults 50 years of age** and older can book an appointment for their third dose **at least 6 months (168 days)** after the second dose.

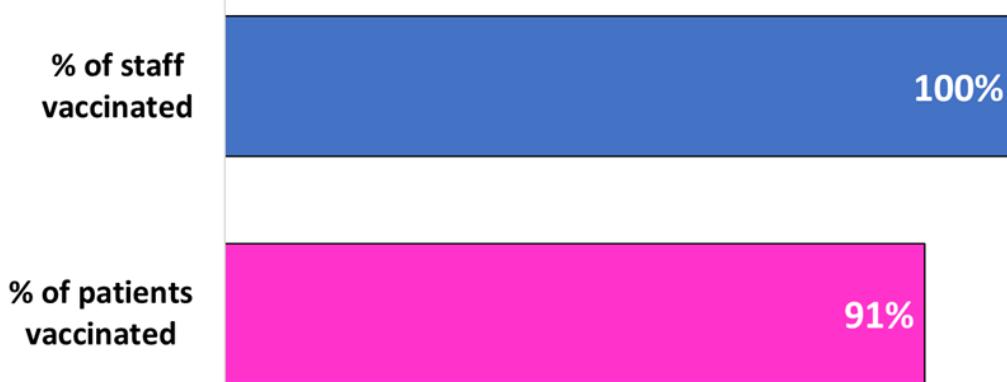
As of December 20<sup>th</sup>, **individuals aged 18 and over** will be eligible to schedule their third dose appointment **at least 3 months (84 days)** after a second dose.

For more information and to book your vaccination appointment (see next page), please visit:

<https://covid19.ontariohealth.ca/>

**Flu vaccine:** With flu season now here and the COVID-19 pandemic ongoing, it is important to get vaccinated against both COVID-19 and the flu now. **It is safe to receive the COVID-19 vaccine and the flu shot at the same time or close in time.**

# Vaccination Status of Our Patients & Clinic Staff Members



All of our staff are fully vaccinated. As of December 15<sup>th</sup>, 2021, 91% of our patients have received one or more doses of the vaccine. If you have not received the vaccine, book your vaccine now and help us get to 100% so that everyone is protected!

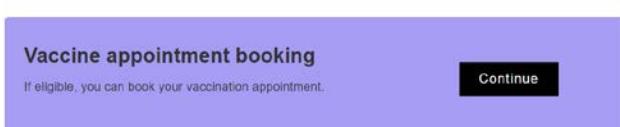
## Proof of Vaccination

Ontarians who are at least 12 years old need to **prove that they are vaccinated** (two doses plus 14 days) before entering or using non-essential businesses or services.

All **essential care partners** must show their proof of vaccination against COVID-19 when they enter the Toronto General Hospital and other UHN sites.

## Where to get your proof of vaccination (QR code) or book your vaccination appointment

1. Get your green OHIP card ready (It works even if it is expired). You will need the information on the front and the back. If you are booking a vaccination appointment, you will also need an email address and a modern browser.
2. Go to <https://covid19.ontariohealth.ca/> and read the instructions.
3. Scroll to this section. You will need to click the box to indicate that you have read the Terms of Use.
4. Enter your OHIP numbers (front & back), version code, date of birth, and postal code.
5. Depending on what you need, click on either "Proof of Vaccination" or "Vaccine appointment booking"



Click  To download your enhanced vaccine certificate or book your vaccine appointment

I confirm that I read, understood, and accept the [full Terms of use](#).

[Download your enhanced vaccine certificate or book your vaccine appointment](#)

If you need help, please call us at (416) 340-5145 or email us at [22q@uhn.ca](mailto:22q@uhn.ca) at the Clinic.

# Staff at the Dalglish Family 22q Clinic



## Dr. Anne Bassett

Clinic Director, Psychiatrist

- Dalglish Chair in 22q11.2 Deletion Syndrome
- Expert in the genetics of complex health conditions
- Has been caring for patients with 22q and their families for over 25 years



## Dr. Maria Corral

Psychiatrist

- Care provider on our team since 2016
- Worked 30 years as a psychiatrist, educator, and administrator
- Provides care in English and Spanish



## Dr. Erik Boot

Research Associate,  
Physician for people with  
intellectual disability

- Dalglish Fellow at our Clinic from 2014-16
- Lives in the Netherlands, but works closely with our Clinic



## Samantha D'Arcy

Registered Dietitian

- Works with patients and families to create nutrition plans that fit their needs and lifestyle
- Leads educational group discussions



## Lisa Palmer

Registered Social Worker

- Has degrees in psychology, education, and social work
- Helps patients and families find the best resources available



## Radhika Sivanandan

Patient Flow Coordinator

- Our front desk leader!
- Facilitator for your appointments and follow ups
- Prepares all documents for staff and patients



## Joanne Loo

Website Manager,  
Patient and Family Educational  
Tool Developer

- Develops content for Clinic website and pamphlets
- Generates medical information cards for patients



## Tracy Heung

Clinical Research Analyst

- Coordinates 22q-related research projects locally and internationally
- Ensures that the data are complete and runs statistical analyses



## Joanne Ha

Clinical Research Analyst

- Invites families to take part in initiatives offered at our Clinic
- Organizes logistics of research projects and clinical care



# Our Trainees

**Dr. Lily Van**

Resident in Psychiatry  
Clinician Scientist Program

**Dr. Sarah (Voll) Malecki**

Resident in Internal Medicine

**Dr. Spencer Van Mil**

Resident in Pediatrics

**Christina Blagojevic**

Medical student

**Brigid Conroy**

Medical student

**Grace McAlpine**

Medical student

**Sabrina Cancelliere**

Medical student

**Shengjie Ying**

Masters student

Recipient of the Joseph Wong Memorial Graduate Award from the Institute of Medical Sciences at the University of Toronto in 2021

**Adonis Ng**

Undergraduate Student

# Our Colleagues and Collaborators

## University Health Network

Toronto Congenital Cardiac Centre for Adults (TCCA),

Peter Munk Cardiac Centre

Dr. Erwin Oechslin

Dr. Candice Silversides

Dr. Rafael Alonso-Gonzalez

Dr. Lee Benson

Dr. Jack Colman

Dr. Eric Horlick

Dr. Luc Mertens

Dr. Mark Osten

Dr. Lucy Roche

Dr. Sara Thorne

Dr. Rachel Wald

Dr. Krish Nair

The Centre for Applied Genomics

Dr. Greg Costain

Medical Geneticist

Christian Marshall, PhD

Stephen Scherer, PhD, and

Ryan Yuen, PhD

Geneticists

Endocrinology

Dr. Satya Dash

Endocrinologist

Movement Disorders Clinic

Dr. Danielle Andrade

Neurologist

Dr. Connie Marras

Neurologist

Dr. Anthony Lang

Neurologist

Ear, Nose, Throat

Dr. John Rutka

Otolaryngology

World-wideAtlantic Canada

Dr. Sarah Dyack

Medical Geneticist

Halifax, NS

Kathleen Hodgkinson, PhD

Geneticist

St. John's, NF

South America

Dr. Gabriela Repetto

Medical Geneticist

Santiago, Chile

Asia

Dr. Brian Chung

Medical Geneticist

Hong Kong SAR, China

USA

Donna McDonald-McGinn, MS, LCGC

Genetic Counsellor

Director, 22q Clinic

Children's Hospital of Philadelphia

Philadelphia, PA

Dr. Linda Brzustowicz

Geneticist

New Brunswick, NJ

Bernice Morrow, PhD

Geneticist

New York, NY

Europe

Ann Swillen, PhD

Child psychologist

Leuven, Belgium

Dr. Therese van Amelsvoort

Psychiatrist

Maastricht, Netherlands

Peter Scambler, PhD

Geneticist

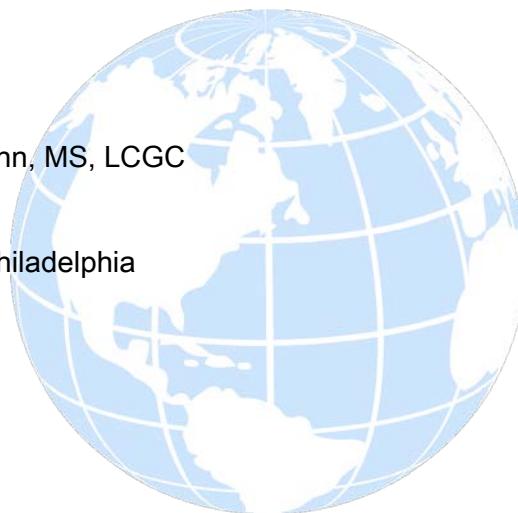
London, UK

Australia

Linda Campbell, PhD

Psychologist

Newcastle, Australia



# Our Clinic and the 22q Community

Team members of our Clinic are leaders in the 22q community. Here are some examples of our involvement in 2021.

## 22q11.2 Society

This society consists of international researchers and physicians who specialize in conditions related to the deletion and/or duplication in the chromosomal region 22q11.2, as well as their underlying biology.



**Dr. Anne Bassett** – Trustee and Treasurer  
Leader in the update of the International Clinical Practice Guidelines for 22q

**Dr. Erik Boot** – Advisor  
Leader in the update of the International Clinical Practice Guidelines for 22q

**Joanne Loo** – Website Editor  
Publication screener and coordinator for the update of the International Clinical Practice Guidelines for 22q

**Tracy Heung**  
Data analyst for research for the International Brain and Behaviour Consortium

## International 22q Foundation

This foundation's aim is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.

**Dr. Anne Bassett** – Medical Director

**Joanne Loo** – Programmatic and Educational Tool Developer

## 22q Parent Conference

Supported by the 22q Deletion Syndrome Clinic at SickKids in Toronto, this parent-organized conference (virtual in 2020 and 2021) is designed to connect families whose lives have been touched by 22q11.2 deletion syndrome (22q11DS).

**Dr. Anne Bassett** – Speaker  
Mental Health Matters

**Lisa Palmer** – Speaker  
Benefits of Peer and Other Supports

## Upcoming Conference

The 12<sup>th</sup> Biennial International 22q11.2 Conference will take place in-person at Le Méridien Lav Hotel, Split, Croatia in **2022**, we hope!

### **Family Meeting**

- June 26 (Sunday) to 28 (Tuesday)

International Brain and Behaviour Consortium (IBBC) and Network of the Americas

- June 28 (Tuesday)

### Professional Meeting

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



# Raising Awareness about 22q in UHN

22q is not a well-known condition, even among medical professionals. In order to raise awareness, we applied to display information about 22q on the computers of the University Health Network (which includes the Toronto General Hospital). From November 20<sup>th</sup> to 27<sup>th</sup>, 2021, healthcare professionals at the Toronto General Hospital, Toronto Western Hospital, Princess Margaret Hospital, and Toronto Rehab – University Centre saw this infographic. If you would like to download a pdf or a jpg version of the infographic, please go to:

<https://22q.ca/22q112ds-quick-facts/>

# 22q

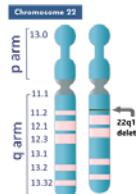
## 22q11.2 Deletion Syndrome

Affects  
**1 in 2148 live births**

Under-recognized cause of  
**medical complexity** in young adults

Learning disabilities  
Anxiety / Schizophrenia  
Seizure risk  
Early onset Parkinson's

Congenital heart disease  
Hypocalcemia  
Autoimmune diseases  
Diabetes risk



**A genetic diagnosis allows for anticipatory care.**

Please visit [www.22q.ca](http://www.22q.ca) or email us at [22q@uhn.ca](mailto:22q@uhn.ca).



## Lighting the CN Tower in Red!

The [CN Tower](#) was lit up in **red** to raise awareness for 22q on November 22<sup>nd</sup>!

The weather condition was excellent, and we could spot the red CN Tower even from 30 km away. Nevertheless, seeing the tower in close proximity was a real experience. The most recognizable Toronto landmark was majestic when lit in bright red. Of course, we keep in mind the true meaning of this event - we want to let Toronto and even the whole world know about 22q, an under-recognized condition that affects 1 in 2,148 live births.

We chose November 22<sup>nd</sup> ("22/11") as a play on the name of the condition, 22q11.2 deletion syndrome ("22q"). The lighting of the CN Tower was part of the "[Luces por el 22q/Lighting the 22q](#)" international awareness movement that began in Spain in 2018. Buildings in Spain, Germany, Belgium, Finland and the city of Philadelphia, United States, were lit up as part of the movement on the 22<sup>nd</sup> of November in previous years. Now joining this international awareness movement is Toronto – a national leader in providing care for families through clinics at the Toronto General Hospital and at SickKids Hospital, and in internationally renowned research into adult outcomes and genetics.

To see more photos of the CN Tower in red, please open this [Google Photo](#) folder.



# Celebrating Your Talents

Even though many of you face health-related challenges, you constantly amaze us with your talents and determination. Some of you paint artwork or play music, and many of you play various types of sports, even participating in Special Olympics. We are delighted to showcase a selection of your artwork and athletic endeavours.



If you would like us to share your artwork and accomplishments in future newsletters, please email us at [22q@uhn.ca](mailto:22q@uhn.ca). Thank you.

## We celebrate your talent in:

Badminton	Hiking	Weight-lifting
Baseball	Hockey	Writing
Basketball*	Horseback-riding	Yoga
Biking	Karate	
Bowling*	Knitting	Playing music instruments:
Boxing	Lacrosse	
Chess	Macramé	
Crochet	Painting	
Cross-country skiing	Photography	
Crafts	Pilates	
Curling	Running	
Dance	Skating	
Floor hockey*	Snowshoeing	
Football	Soccer	
Gaming	Softball*	
Golf*	Swimming	
Graphic design	Volleyball	
Gymnastics	Walking	
		And many more!

(\* = in Special Olympics)

# Topics Often Avoided...

## Planning Ahead for Care

As a parent or caregiver, “Who will take care of my loved one with 22q and complex care needs in the future” is a common question.

It is always a great idea to plan for the future of your loved one, as best as possible, when you are able to do so. Here are some general tips to help.

1. Have open, honest and developmentally appropriate **conversations** about wants and support needs with your loved one. Include, as appropriate, informal (e.g., family and friends) and formal (e.g., community support staff, medical professionals) supports (the “circle of care”) in the conversations.
  - Revisit and modify these advance care plans, as needed (e.g., as the “circle” changes over time).
2. Educate yourself about who your loved one’s **substitute decision maker** is now, and who would be best to take on that role in the future.
3. Educate yourself about community-based and government **funding, supports, and services** available to your loved one. Connect with local agencies, ask questions, and advocate for supports.
  - Revisit this periodically, as the supports and services change over time.
4. Connect with a **legal clinic** that specializes in supporting people with disabilities and their families.

Reassure the individual with 22q that **they will be cared for**. Remind them who is in their circle of care.

## Tips for Constipation

Constipation is a common problem, especially for many people with 22q, but there are 3 small changes you can make to help reduce constipation:

### Eat more fiber

Eat 1-2 more high fiber foods each day, like fruits, vegetables, and whole grains.



### Drink more fluid

Drink 250-500 ml more water each day. Check your pee: it should be clear or light yellow, not dark yellow.



### Do more physical activity

Add 15 minutes of walking or exercise to your routine each day.



# Our Research Publications



## How common is 22q?

### Estimate of the contemporary live-birth prevalence of recurrent 22q11.2 deletions: a cross-sectional analysis from population-based newborn screening

Christina Blagojevic, Tracy Heung, Mylene Theriault, Aoy Tomita-Mitchell, Pranesh Chakraborty, Kristin Kernohan, Dennis E. Bulman and Anne S. Bassett; CMAJ Open, 2021, 9 (3) E802-E809; PMID: 34404688  
[\[Full text\]](#)

#### Main Findings

- Among 30,074 newborns in Ontario, 14 had 22q deletions.
- This means **22q has a prevalence of 1 in 2148 live births**.
- 22q is more common than cystic fibrosis!
- Babies with 22q were:
  - More likely to be born to younger mothers;
  - Smaller on average;
- Babies with 22q also had lower counts of a blood test called TREC, but not low enough for 22q to be detected with this test.

With a prevalence of 1 in 2148 live births, 22q is more common than people think. We strongly recommend adding 22q to standard newborn screening programs. This would allow for early diagnosis and optimal treatment

## Can normal variability in unaffected parents explain some of the variability among individuals with 22q?

### Within-family influences on dimensional neurobehavioral traits in a high-risk genetic model

Fiksinski AM, Heung T, Corral M, Breetvelt EJ, Costain G, Marshall CR, Kahn RS, Vorstman JAS, Bassett AS. Psychological Medicine, (pages 1–9), e-published 14 January 2021 ahead of print; PMID: 33443009 [\[Full text\]](#)

We studied families where one person had 22q but neither parent had 22q.

#### Main Findings

- Within each family, the 22q11.2 deletion has by far the largest effect on the intellectual functioning of the person with 22q.
- The parents' IQ scores also have an impact on the IQ of their offspring with 22q.
- There was no significant relationship between the parents' social or motor functioning and the social or motor functioning of the person with 22q.



# Our Research Publications (continued)

## How much do you know about sexual health?

### Sexual knowledge and behaviour in 22q11.2 deletion syndrome, a complex care condition

Palmer LD, Heung T, Corral M, Boot E, Brooks SG, Bassett AS. Journal of Applied Research in Intellectual Disabilities (pages 1–10), e-published 21 July 2021 ahead of print. PMID: 34291536 [[Pubmed](#)]

- We have identified some sexual health knowledge deficits in adults with 22q. These deficits exist whether or not the person has intellectual disabilities.
- Adults with 22q are engaging in sexual activities.
- Some of these include high-risk sexual activities, such as:
  - Unprotected sex
    - Resulting in unplanned pregnancies
    - Acquiring sexually transmitted infections

- There is a need to provide:
  - More preventive sexual health measures
  - A safe space for discussions
  - Repeated education and counselling.

If you have any questions or concerns about your sexual health, please talk to your healthcare providers. All discussions are confidential.

### More of our 2021 publications:

#### Abnormal spirometry in adults with 22q11.2 microdeletion and congenital heart disease

Blagojevic C, Heung T, van Mil S, Oechslin E, Silversides CK, Granton JT, Bassett AS. International Journal of Cardiology: Congenital Heart Disease, 3:100085 (pages 1-7), 2021 [[Full text](#)]

#### A normative chart for cognitive development in a genetically selected population

Fiksinski AM, Bearden CE, Bassett AS, Kahn RS, Zinkstok JR, Hooper SR, Tempelaar W, the 22q11DS International Consortium on Brain and Behavior, Vorstman JAS, Breetvelt EJ. Neuropsychopharmacology (pages 1–8), e-published 29 March 2021 ahead of print; PMID: 33782512 [[Pubmed](#)]

# Our Research Funding

Multiple research grants are needed to help support our research studies, and we submit several applications every year. If and when these are funded, these grants support our research studies of 22q and related conditions.

We are grateful to these agencies for their research support.

In addition, we thank The Weston Family Foundation and the Dalglish Family for the renewal of the Dalglish Chair in 22q11.2 Deletion Syndrome.



#### Canadian Institutes of Health Research (CIHR)

- Five-year project grant, 2016-2021
- Five-year project grant, 2020-2025

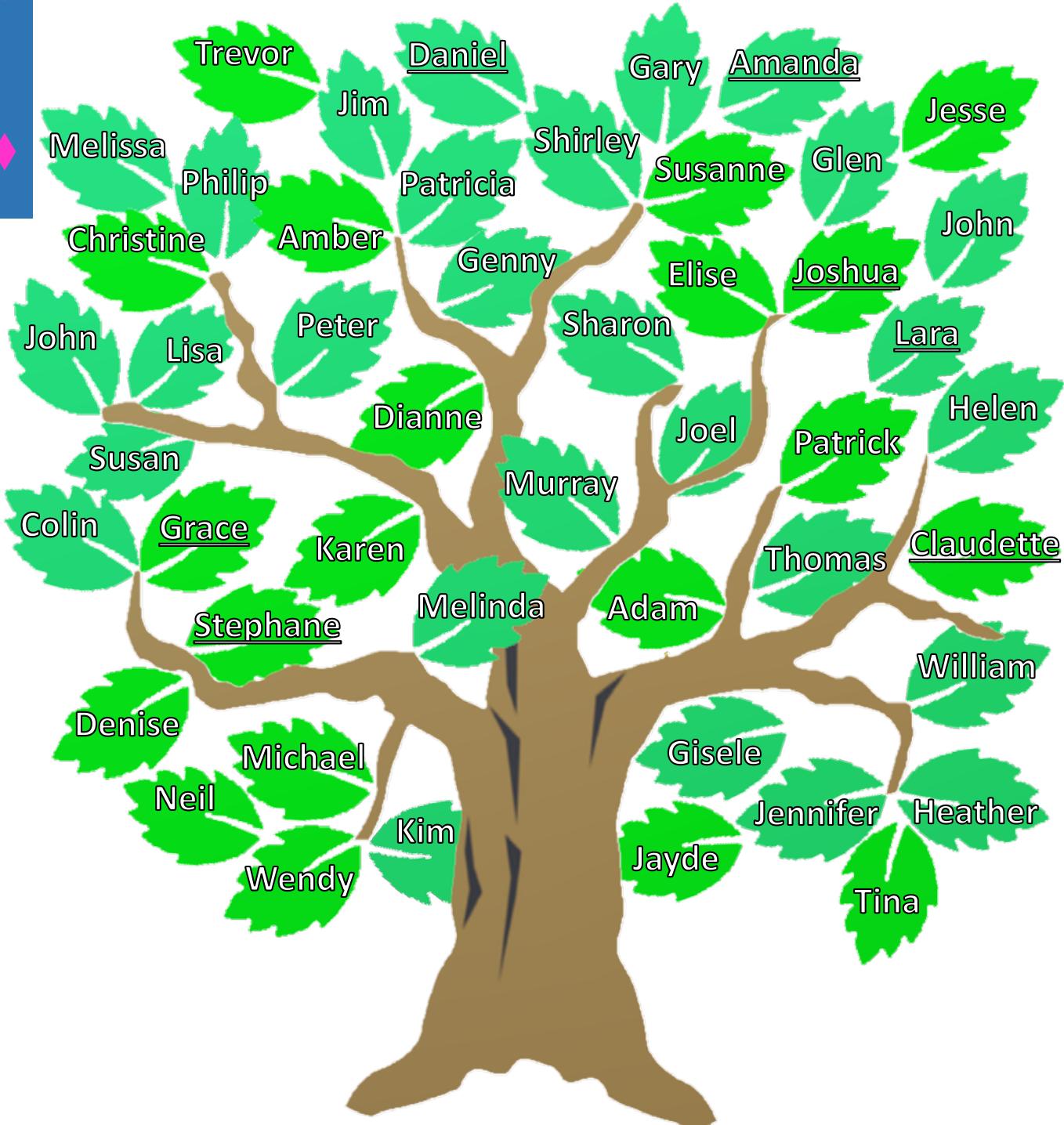
#### National Institute of Mental Health (NIMH)

- Small five-year sub-contract, 2019-2024



Weston Family Foundation

# In Memoriam



These are the members of our 22q family we have lost over the past 25 years, including those individuals in 2021 (one who was 76 years of age). They will never be forgotten, and remain close to our hearts. Their research contributions live on. Through the knowledge generated, they continue to help others with 22q. They are true angels!

# How can you help?

Your most precious gift is helping with our research to understand 22q!

The Dalglish Family 22q Clinic is a world leader in 22q, providing:

- **Specialized, individual-centred care** for adults with 22q and their families
- **Excellence in education** for patients, families, as well as trainees and health care providers
- **World-class research** to advance our understanding of all aspects of 22q and improve care and outcomes.

The Ontario Ministry of Health supports some of the basic operations of our Clinic but funding is not secure for our Clinic.

Also, we receive no financial support for the education, outreach, and many other things we do for patients with 22q and their families.

Please consider a donation to support these efforts.



Donations to the Dalglish Family 22q Clinic support patient care, education, and research that benefit adults with 22q and their families.

The [UHN Foundation](#) manages all donations for these important areas. Please visit our [Donate](#) page (<https://22q.ca/donate/>) for more information.

## Three ways to donate

### Donating online – easy to do!

- Please visit our [Donate](#) page (<https://22q.ca/donate/>) to donate online.

### Donating by mail or fax

- Please download the form from our [Donate](#) page (<https://22q.ca/donate/>)
- Make the cheque payable to the UHN Foundation
- Be sure to write **22q** in the memo field of the cheque.

### The [Honour Your Hero](#) program

- Honour a hero (a staff member or a care team) by making a donation.
- The hero will receive a certificate and a thank-you message from the donor.
- Please specify that the donation goes to the **Dalglish Family 22q Clinic**.

**Thank you** for supporting what we do for individuals with 22q.

# Why am I asked to participate in 22q research?

Without your help, we would know nothing about 22q, and how best to care for people with 22q!

Over the years, **hundreds of our patients with 22q** – and others around the world – have helped doctors and scientists to learn about 22q.

We are one of the very few centres to focus on adults with 22q – so every person who participates is precious.

## What do I have to do?

The short answer is **not very much!**

A research analyst will tell you about the study, and answer your questions.

We make our 22q studies as person-friendly and convenient as possible. So there is little of your precious time involved.

We invite you to take part. And always want to hear from you how we could make this process even better.

Ask us what studies we are working on. We are happy to explain!

## Why do we include family members in our studies?

Family members are the very best comparison group!

Please contact us if you have not joined and are interested to know more.

We are happy to answer your questions!

**Thank you** for supporting our efforts to help people with 22q. Your participation will improve the lives of those with 22q now and in the future!

To download a digital copy of this newsletter or previous ones, please scan the QR code to reach:



<https://22q.ca/aboutus/our-newsletter/>

Visit our website at  
[www.22q.ca](http://www.22q.ca)

## Contact us!

**The Dalglish Family 22q Clinic**  
**Toronto General Hospital**

8 NU, Room 802 (8th Floor)

200 Elizabeth Street

Toronto, Ontario M5G 2C4

Phone: (416) 340-5145

Fax: (416) 340-5004

E-mail: [22q@uhn.ca](mailto:22q@uhn.ca)



The Dalglish Family  
22q Clinic

Please call or email Radhika for an appointment if we haven't seen you in a while!