

22q & Friends

2020 Newsletter



Hello and happy reading from all of us

2020 has certainly been an unusual year for all of us. Because of the COVID-19 pandemic, our Dalglish Family 22q Clinic has increased Ontario Telehealth Network (OTN) and phone (virtual) care for patients. We now offer appointments both online and in person. Also, our [Transition Program](#) and the annual “[22q at the Zoo](#)” event took place virtually. Please go to page 5 to see what our Clinic is doing to continue to care for you and your family safely.

We have also restarted our active clinical research activities to suit the COVID-19 realities. And we continue to publish our important results—all thanks to you and your participation! We invite you to go to pages 9-10 to see our publication highlights and page 11 for an interview with one of our colleagues.

Happy reading!

Dr. Anne Bassett receives the 2020 Lieber Prize for Outstanding Achievement in Schizophrenia Research



Congratulations to Dr. Anne Bassett, our Clinic Director, who is the 2020 winner of the international [Lieber Prize for Outstanding Achievement in Schizophrenia Research](#) from the Brain and Behavior Research Foundation.

Established in 1987, the prize aims to bring public recognition to the outstanding discoveries being made to understand this important brain disease.

Dr. Bassett is only the second Canadian ever to receive this award. She is hopeful that the future will bring further advances in prevention and treatment.

Inside you will find...

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|--------|--|------------|---|
| Page 2 | ➤ Staff biographies | Pages 9-10 | ➤ Our publications—A direct result of your help |
| Page 3 | ➤ World-firsts at the Clinic | Page 10 | ➤ Upcoming publications |
| Page 4 | ➤ Staying healthy and calm during the pandemic | | ➤ Our research funding |
| Page 5 | ➤ What we are doing at the Clinic to keep you safe | Page 11 | ➤ BIG data and 22q |
| | ➤ Reliable info about COVID-19 | Page 12 | ➤ In memoriam |
| Page 6 | ➤ Telling apart allergies, the cold, the flu, and COVID-19 | | ➤ Inspiration |
| Page 7 | ➤ Sleep apnea and CPAP | Page 13 | ➤ How can you help? |
| Page 8 | ➤ Resources from our Clinic | | ➤ Three ways to donate |
| | | Page 14 | ➤ 22q research participation |
| | | | ➤ Contact us! |



Staff at the Dalglish Family 22q Clinic

Dr. Anne Bassett

Clinic Director, Psychiatrist



- Expert in the genetics of complex health conditions
- Has been caring for patients with 22q and their families for over 25 years
- Medical Director for the [International 22q Foundation](#)
- Treasurer of the [22q11.2 Society](#)

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

Radhika Sivanandan

Patient Flow Coordinator



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

Lisa Palmer

Social Worker



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

Samantha D'Arcy

Registered Dietitian



- Works with patients and families to create nutrition plans that fit their needs and lifestyle
- Leads educational group discussions
- On maternity leave until February 2021

Ada Duperrouzel

Registered Dietitian



- Provides great advice and tips on healthy eating
- Shares fun ways to bring nutritious snacks and meals into your daily routine
- Is with us until Sam returns from maternity leave

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

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



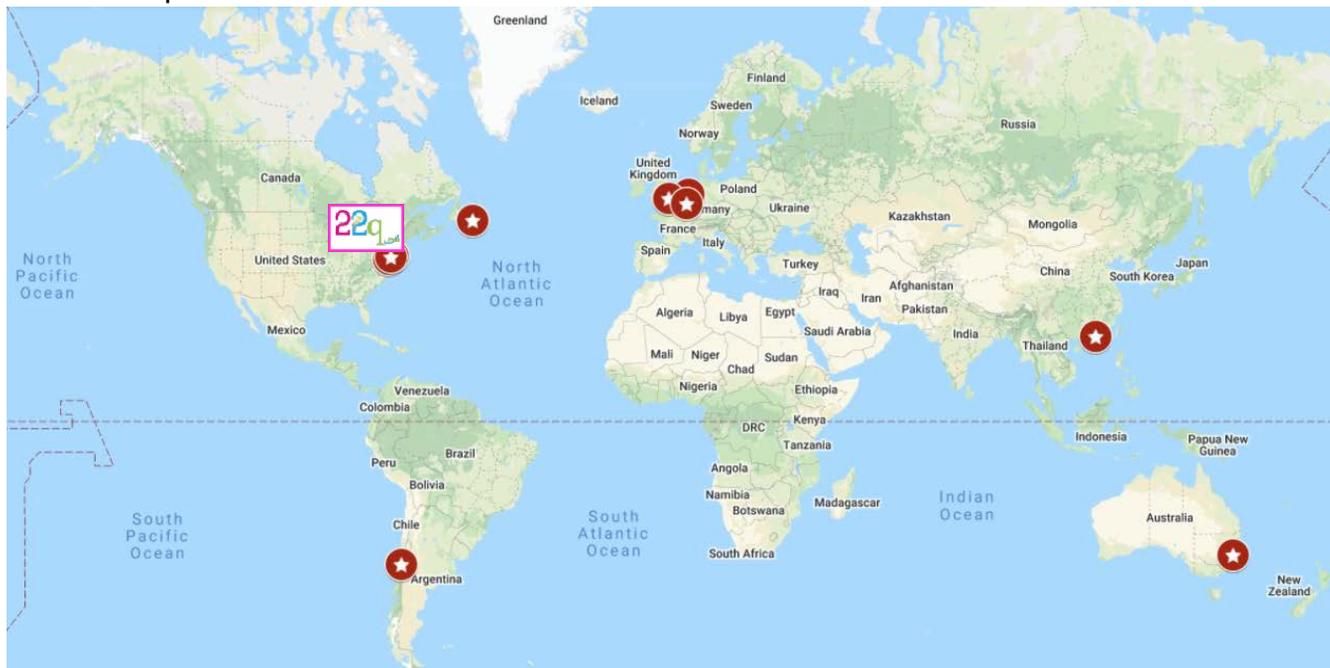
- Invites families to take part in initiatives offered at our Clinic
- Organizes logistics of research projects and clinical care

We are leaders in the international community!

Did you know that the Toronto General Hospital in the University Health Network, was recognized as the **fourth best hospital in the world** by [Newsweek](#) in 2020?

Our Clinic is the first of its kind in the world, specializing in adults with 22q. We are leaders in the international community, and have collaborators all around the world.

Our **world-first** research contributions have helped improve the quality of life for individuals and families affected by 22q. Can you name all the locations of our collaborators? Check out our website for the [list of worldwide collaborators](#). Here is a map of some of their locations.



First whole genome sequencing (WGS) studies

Possible features of 22q:

- * Type 2 diabetes
- * Obesity
- * Hypocalcemia
- * Multi-morbidity

We're the **1st** in the **WORLD** to discover

Early-onset Parkinson's disease in 22q

Connection between 22q and scoliosis in congenital heart disease

Clozapine as an effective treatment of schizophrenia (SZ) in 22q

Diagnostic odyssey of 22q persists

Publication of the first international clinical practice guidelines for adults with 22q

Additional genome-wide rare copy number variants ↑ risk of expression of SZ

We are making new discoveries at a great rate, thanks to your support! See page 14 and find out how you can join our research family.

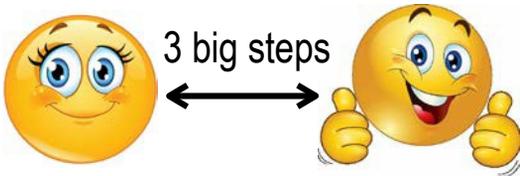
Staying healthy and calm during the pandemic

The COVID-19 pandemic has brought along some stresses and uncertainties. Although there are many things that you cannot control, here are some things you can do to stay healthy and calm, while we await the vaccine!

Adapted from "[Staying calm during COVID-19](#)" in the COVID-19 series from the International 22q11.2 Foundation

Do your best to stay healthy

- Stay 6 feet away (3 big steps) from others



- Wear a mask when out of your home
See [video for using a mask](#)



- Wash hands properly and frequently
See [video for handwashing](#)



- Avoid touching the face



Don't forget to:

Eat, sleep, exercise well

Stay hydrated

Avoid smoking, drugs, and alcohol

Write down how you feel and see what you can do about it

- How did you cope with hard things before?
- Does worrying about this help at all?
- Could meditation and breathing exercises help?
- Can you talk to a friend or relative to stay positive?
- Do you need to talk to your doctor?

Get information from trustworthy sources

- Some things you see on social media may not be accurate. Please get your information directly from hospital and government websites (see page 5)
- Only look at COVID-19 info once a day. Do something else afterwards

Do something to keep yourself busy

- Check out online tours for zoos, museums, aquariums, sports hall of fame etc.
- Read a book, listen to music, or watch a movie
- Prepare a healthy snack
- Learn a new language or new skill online
- Do a chore
- Help someone
- Take care of your pet

**Let's support one another.
We will get through these
uncertain times.**

If you have any concerns, please contact us at the Clinic at 416-340-5145 or 22q@uhn.ca.

What we are doing at the Clinic to keep you safe

COVID-19 or not, your health is important.

Our Clinic staff have found ways to ensure we take care of patients and families while minimizing contact during the pandemic. Please let us know how we can help you.

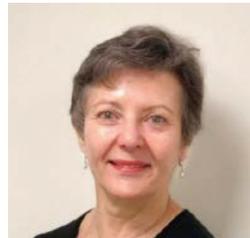


Caring remotely

- Our Clinic staff have been seeing some of our patients through the **Ontario Telehealth Network (OTN)**. This is easy to arrange, and is a secure online health service in your own home.
- Depending on the individual situation, our Clinic may be able to provide telephone appointments for non-urgent concerns. This may include prescription renewals, and new, non-urgent concerns.
- Patients can often get their blood work and other investigations done at an approved centre near their home.

At the Clinic

- There is now a **plexiglass screen** at the reception area.
- Caregivers can still come to the Clinic with patients.
- If you or your caregivers test positive for COVID-19, let us know. We can help you make a list of your contacts during a visit.
- **All patients and caregivers visiting our Clinic must wear masks.**
- All Clinic staff seeing patients wear **masks and face shields**. Below is a photo of Dr. Corral with, and without the mask and face shield.



- **Hand sanitizers and extra masks** are available to patients.

Reliable Information about COVID-19

There is a lot of information on the Internet about the current pandemic, but **not everything is accurate**. Please visit the following sites for the **most up-to-date** and **reliable** info.



- Our Clinic (www.22q.ca)
 - [COVID-19 webpage](#) & [leaflet](#)
- Our hospital organization (www.UHN.ca)
 - [COVID-19 webpage](#)
- From the Ontario Medical Association
 - www.virusfacts.ca
- Government of Canada (www.canada.ca)
 - [COVID-19 webpage](#)

COVID-19 Series from the International 22q11.2 Foundation www.22q.org
(Click below for info sheets)

[What is COVID-19?](#)

[Why should we stay home?](#)

[Staying calm during COVID-19](#)

[False information and scams](#)

[Immunodeficiencies and COVID-19](#)

[Heart conditions and COVID-19](#)

[When can we return to work/school?](#)

[PCR vs. Antibody testing](#) (Testing methods)

[Masks: Why, When, and How](#)

[Allergies, Cold, Flu or COVID-19?](#)

[COVID-19 Vaccines](#)

Telling apart allergies, a cold, the flu, & COVID-19

COVID-19 shares some symptoms with allergies, the cold, and the flu. Sometimes, it is a little difficult to figure out what a person has. Here is a simplified guide to help you. **If you have any concerns about your health, please check with your healthcare provider.**

Source: "[Allergies, Cold, Flu, or COVID-19](#)" in the COVID-19 series from the International 22q11.2 Foundation

FEVER is one of the main symptoms of COVID-19.

If your body temperature is above 37.8°C or 100°F, please get tested for COVID-19.

Fever is also a common symptom of the flu.

However, fever is NOT a common symptom of a cold and NOT a symptom of allergies.



The **loss of taste or smell** is a common symptom of COVID-19.

However, loss of taste or smell can also happen in rare cases of colds, the flu, and allergies.



Dry coughs, headaches, sore throat and fatigue are all possible symptoms of COVID-19.

However, they can also happen with colds, the flu, and allergies.

Runny or blocked nose, itchy eyes, and sneezes are very unlikely to be symptoms of COVID-19.

A runny/blocked nose can be due to a cold, the flu, or allergies.

If you have itchy eyes, you may have allergies.

Sneezing can be due to a cold or allergies.

COVID-19 testing is important for you and those around you.

To get more information about testing and to find a testing centre, please visit the [Government of Ontario's COVID-19 website \(https://covid-19.ontario.ca/\)](https://covid-19.ontario.ca/).

The COVID-19 Assessment Centres closest to our Clinic are located at Women's College Hospital and Mount Sinai Hospital.

Sleep Apnea: What it is, and How CPAP helps

Sleep apnea is an illness in which a person **stops breathing** many times when sleeping. Here, we will talk about **obstructive sleep apnea**, which is the more common type of sleep apnea in 22q.

Symptoms of sleep apnea

These symptoms can be signs of sleep apnea:

- Feeling tired even after sleeping
- Waking up often due to choking or gasping
- Others say that you snore loudly or stop breathing when you sleep
- Difficulty staying asleep
- Morning headaches
- Daytime sleepiness
- Poor concentration
- Easily irritable (feeling grouchy)



How does sleep apnea happen?

Sleep apnea happens when certain muscles in your throat relax and narrow your airway.

Then you cannot get enough air, and the oxygen level in your blood goes down.

Your brain briefly wakes you up to breathe, but you do not remember this.

You keep waking up to breathe, and you don't sleep well.

What can increase the risk of sleep apnea?

- Excess weight
- Smoking
- Men
- Women after menopause
- Older age
- Others in the family have obstructive sleep apnea
- Nasal congestion
- A narrowed airway



Many of our patients report improved mood when they use their CPAP regularly!

More info about sleep apnea & CPAP

- In the *Canadian Medical Association Journal*:
 - [Diagnosis and treatment of obstructive sleep apnea in adults](#)
 - [Appendix 2](#) lists potential solutions for problems using CPAP.
- The [Sleep apnea](#) webpage from the Mayo Clinic is also helpful
- Ontario's [Assistive Device Program](#) may cover part of the cost of your CPAP system. Please visit the Government of Ontario's [Respiratory equipment and supplies](#) page for more information.

Complications of sleep apnea

You should not ignore sleep apnea. It can lead to complications including:

- Feeling very tired, drowsy, and irritable
- Increased risk for accidents
- Less able to do well at school or work, even some behaviour problems
- Increased risk of heart problems like high blood pressure, heart attack, stroke

Diagnosis: Sleep study

Ask your doctor if you think you may have sleep apnea.

A sleep study is often helpful.

While you sleep, your heart, lungs, brain, movements, and oxygen levels are checked.

This makes it possible to “see” if there is sleep apnea and how severe it is.

Treatment: Using the CPAP

A little **CPAP (Continuous positive airway pressure)** machine can help you keep your airways open. This decreases snoring, the number of times you stop breathing, and related problems.

CPAP is the most reliable way to treat sleep apnea. And treating sleep apnea is very important for your overall health.

Losing weight and quitting smoking also help.

Resources from our Clinic

Did you know that our Clinic website offers a variety of topics related to 22q? We have sections for patients and families, health professionals, educators, and the police. Please visit our Clinic website at: www.22q.ca

[Our Pamphlets](#) page (22q.ca → Our Clinic → Our Pamphlets) contains the most up-to-date versions of our educational materials:

Our pamphlets

- Leaflet on 22q and the Dalglish Clinic
- Dalglish Family 22q Clinic Pamphlet
- Pamphlet for Health Professionals
- Information for Educators and School Resource Teams

Transition factsheets for teenagers and young adults

- Alcohol
- Mental health
- Sexual health
- Internet safety
- Healthy eating
- Emergency preparedness

Helpful tips

- Preparing to see a doctor or other healthcare professionals
- What to bring when you come to your 22q Clinic appointment

Did you know...

469 distinct households/organizations visited our Clinic website within a span of 10 full days in March 2020. This number does not include any automated computer entities.

Here are some topics on the website in the section for our patients and families

- What is 22q? Genetics Q&A
- Natural health products
- Recreational drugs
- Healthy sexuality and pregnancy
- Preparing for a medical appointment
- Vaccination
- Cancer screening
- Staying safe on the Internet
- Interacting with the Police
- Connecting to services
- Financial and legal considerations

22q info in other languages

<https://22q.ca/resources/22q-info-in-other-languages/>

[Español](#)

[Nederlands](#)

[Italiano](#)

[中文](#)

External resources

The International 22q11.2 Foundation Inc.
www.22q.org

Schizophrenia Society of Canada
www.schizophrenia.ca

Canadian Congenital Heart Network
www.cachnet.org

Our Publications – A Direct Result of Your Help

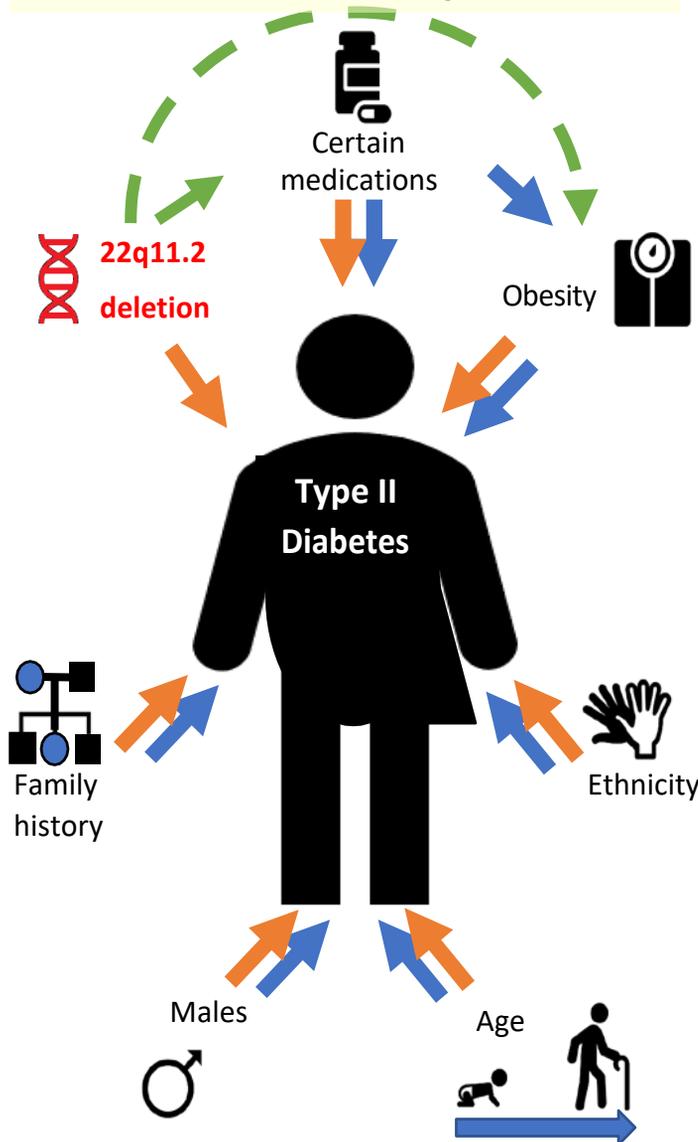
Check out our recent publications in the [Research](https://22q.ca/research/) section (<https://22q.ca/research/>) of our website! Our publications are only possible because of your generous contributions, which help us learn more about 22q. You are helping people with 22q here in Canada and worldwide! Thank you!

22q11.2 microdeletion and increased risk of type 2 diabetes

Van L, Heung T, Malecki SL, Fenn C, Tyrer A, Sanches M, Chow EWC, Boot E, Corral M, Dash S, George SR, Bassett AS. *E Clinical Medicine*. 2020; 26; 100528 (DOI: 10.1016/j.eclim.2020.100528)

A few years ago, we reported that the risk of obesity (being overweight) in adults with 22q was higher than in the general Canadian population. In our current study, we studied the possible effect of 22q on the risk of type 2 diabetes (T2D).

A proposed model of increased risk for type 2 diabetes in adults with 22q



For 314 adults with 22q, we were able to compare data to similar data from a survey of 11,874 Canadian adults.

We found that individuals with 22q were more likely to develop T2D compared to the general population, even when we took into account other risk factors for diabetes, like older age, obesity, medications, and family history of diabetes.

Also, the average age at diagnosis of diabetes was younger in adults with 22q.

This new knowledge means that we are changing when we start to monitor for diabetes in 22q. This will allow us to put in place measures to help prevent diabetes!

- Significant findings from our 22q diabetes study.
- Relationships suggested by previous studies of 22q.
- Diabetes risk factors in the general population

The green and blue dashed lines indicate results from other studies.

Our Publications – A Direct Result of Your Help (Continued)

Personalized medical information card for adults with 22q11.2 deletion syndrome: An initiative to improve communication between patients and healthcare providers

22q
The Dalglish Family
22q Clinic

The Dalglish Family 22q Clinic
Formerly The Dalglish Family Hearts and Minds Clinic For Adults with 22q11.2 Deletion Syndrome
Toronto General Hospital
Phone: (416) 340-5145 Fax: (416) 340-5004
Website: www.22q.ca/

MEDICAL INFORMATION CARD

Name: _____ Date of birth (dd/mm/yyyy): _____
Phone #: _____
OHIP number: _____ Medical Records # (MRN): _____
Emergency contact: _____
Family Physician: _____
Pharmacy: _____

For further information, please call The Dalglish Family Hearts and Minds Clinic at (416) 340-5145.
For after hours assistance, please call the Health Records Services at: (416) 946-4501 extension 4711
This card was last updated (dd/mm/yyyy): _____

Loo JCY, Boot E, Corral M, Bassett AS.
J Appl Res Intellect Disabil. 2020;00:1–7.
DOI: 10.1111/jar.12747; PMID:32407568

Many adults with 22q11.2DS and their family members have a hard time providing crucial information to those who try to help. To solve this problem, our Clinic has been offering [personalized medical information cards](#) for patients.

Card users have found the card to be useful in multiple ways. They provide necessary information, speed up interactions with professionals, and help avoid repeat storytelling. The image on the left shows what the front and the back of the card look like.

As of November 2020, we have generated medical information cards for 69 patients. If you would like to receive a personalized medical info card, please speak with one of our staff members.

has **22q11.2 Deletion Syndrome**
(Previously known as DiGeorge or Velo-cardio-facial syndrome).

Primary health concerns: _____

Medications: _____

Note: The information on this card is subject to change

Allergies or sensitivities: _____

Upcoming publications

- Population-based newborn screening estimate of the live birth prevalence of 22q11.2 deletions
- Pulmonary dysfunction in adults with 22q11.2 microdeletion and congenital heartdisease
- Can we identify what makes the human genome prone to having a 22q11.21 deletion?

To see more publications, please visit [Our Research \(https://22q.ca/research/\)](https://22q.ca/research/)

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.



McLaughlin Centre Accelerator Grant

- One-year operating grant, 2019-2020

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

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

THE W. GARFIELD WESTON
FOUNDATION

BIG data and 22q!

We recently interviewed Dr. Sarah (Voll) Malecki, who has been working with Dr. Bassett and studying individuals with 22q since 2014. She told us some exciting findings from her recent scientific papers, both involving the use of “big data” – **Canadian population data** to help us understand 22q.



Could you tell us what “multimorbidity” is.

Multimorbidity means having several important health conditions all at once. This seems to happen often in 22q. So we wanted to find out more about how the health status of 22q compared to the Canadian general population.

We used medications as an indirect way to count the health conditions. At the [Statistics Canada Research Data Centre](#) (StatsCan RDC), we got data from the [Canadian Health Measures Survey](#) (CHMS). This allowed us to compare the health situation of our 22q group relative to that of the general population. We found that young adults with 22q (aged 20-44 years) had health situations that were like people age 65 in the general population. And at age 45-64, people with 22q had about twice the number of health conditions as in the general population at that age. These results give us an idea about the burden of illness for people with 22q, and how we could start to lower this burden. “Big data” could help.

What about the recently published diabetes paper, what did you find?

After finding that a higher proportion of our 22q patients have obesity (being overweight) compared to Canadians in general, we wondered about risk for diabetes in 22q. I was involved in the planning and then the later stages of the Type 2 diabetes (T2D) study.

What we found was that quite a few of our 22q patients were on metformin, a medication for diabetes. And that the 22q deletion on its own is an important risk factor for diabetes. Also, adults with 22q were diagnosed with diabetes at a much younger age than other Canadians.

Sarah’s publications involving “big data”

[Obesity in adults with 22q11.2 deletion syndrome](#)

Voll SL, Boot E, Butcher NJ, Cooper S, Heung T, Chow EW, Silversides CK, Bassett AS.
Genetics in Medicine. 19(2):204-208, 2017. DOI: 10.1038/gim.2016.98

[A genetic model for multimorbidity in young adults](#)

Malecki SL, Van Mil S, Graffi J, Breetvelt E, Corral MG, Boot E, Chow EWC, Sanches M, Verma AA, Bassett AS.
Genetics in Medicine 22:132-141, 2020. DOI:10.1038/s41436-019-0603-1

[22q11.2 microdeletion and increased risk of type 2 diabetes](#)

Van L, Heung T, Malecki SL, Fenn C, Tyrer A, Sanches M, Chow EWC, Boot E, Corral M, Dash S, George SR, Bassett AS.
E Clinical Medicine. 2020; 26; 100528. DOI: 10.1016/j.eclinm.2020.100528

Earlier, you mentioned “big data”. How do you think using “big data” could help people with 22q in the future?

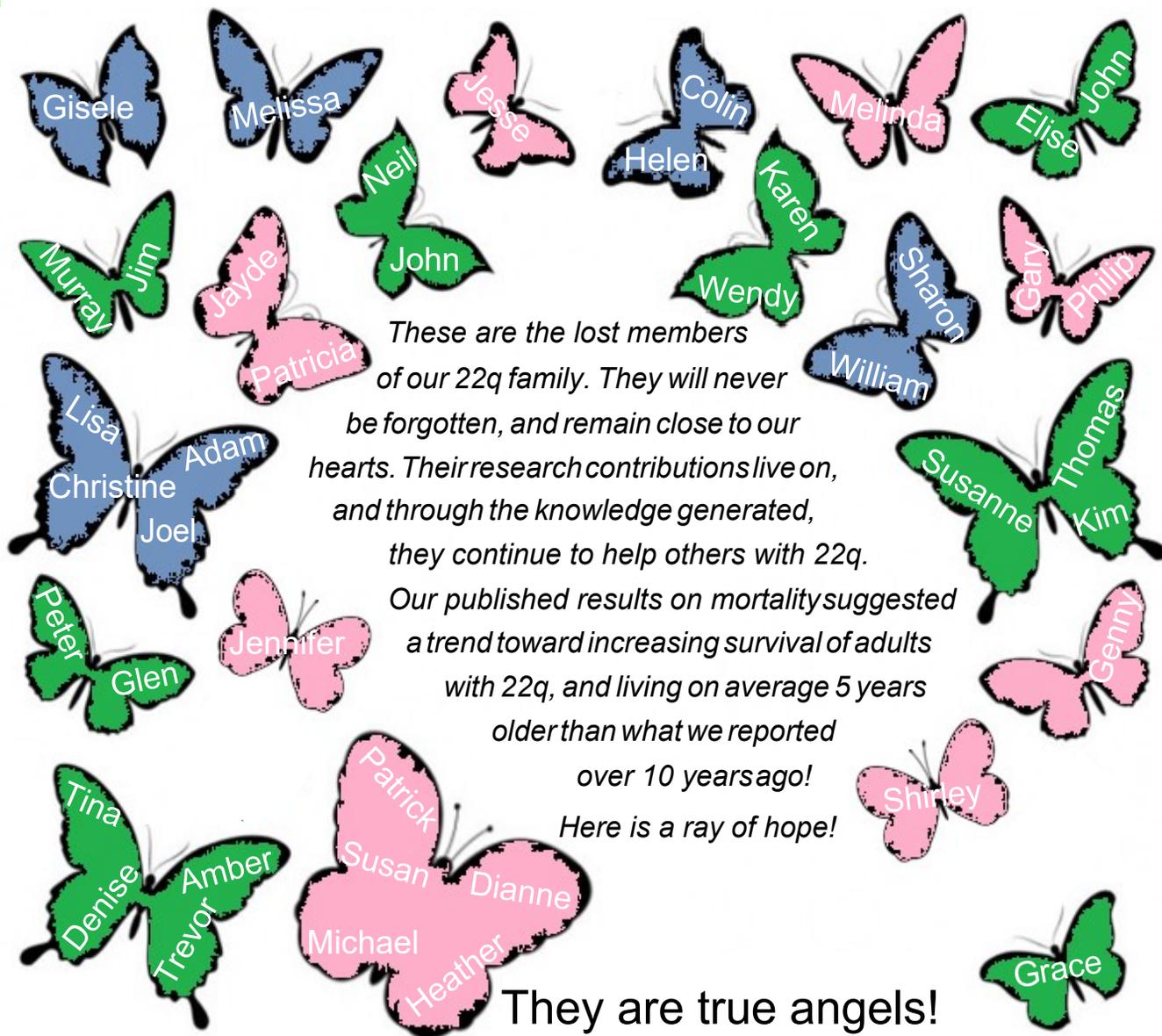
The term “big data” means population data or other large data sets. It might be able to give us a better sense of the true number of people in the population who actually have 22q.

It is already helping us understand the health conditions that people with 22q struggle with. Big data could also allow us to spot risk factors and diseases that can be managed earlier. If we can get a better picture of the disease patterns of 22q, it could help doctors identify people who may benefit from genetic testing, and help them get diagnosed earlier.

Any hints on your next 22q project?

I’m keen to know if big data can help us to better understand 22q, and reduce the burden of health problems. We have some ideas already, so stay tuned for our upcoming publications!

In Memoriam



Inspiration

Our patients with 22q, and their families, inspire us every day.

Even facing many daily physical and other challenges, they keep going.

22q is not a well-known medical condition, but it has a big impact on affected individuals and families.

We are working to improve knowledge about 22q for health care professionals and the general public – but we need to do more.

In memory of one of our patients, Adam, who encouraged people to “think positive”, the [International 22q11.2 Foundation](#) has established an advocacy award (a plaque of recognition) that will honour patients and family members who make a significant impact in the area of advocacy about 22q.

We can each do our little part to raise awareness about 22q so that people can better understand this condition.

How can you help?

Your most precious gift is helping with our research to understand 22q!
But if you have the means, a donation could help.

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 the basic operations of our Clinic so that we can care for our patients' health needs.

However, **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** benefiting adults with 22q and their families.

The [Toronto General and Western Hospital Foundation](#) manages all donations. Please visit our [Donate](#) page (<https://22q.ca/donate/>) for more information.

Three ways to donate

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**.

Donating by mail or fax

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

Donating online – easy to do!

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

Thank you for supporting what we do for people 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!

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



The Dalglish Family
22q Clinic



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

Visit our website at www.22q.ca