# 22q & Friends

## 2023 Newsletter

### Greetings from all of us at the Dalglish Family 22q Clinic!

Our team continues to maintain our outstanding quality of care to both patients in follow-up and those new to our Clinic. While most of the families we serve are affected by 22q11.2 deletion syndrome (22q11.2DS), we have also welcomed several families with 22q11.2 duplication syndrome (22q11.2DupS). Relatively little information is available on 22q11.2DupS, particularly in adults. We look forward to working with patients and their families and advancing the knowledge on this condition.

We have returned to a focus on in-person visits, with "virtual" care options available, depending on what is best for the individual situation. Patients and families continue to express their thanks for our team's wrap-around care and ability to provide the most up-to-date information about 22q11.2DS and 22q11.2DupS. Patients often come to the Clinic with one or more relatives, including parents, and/or other caregivers. We welcome all!

Over the past year we have seen a record number of new patients, includes several coming from pediatric care settings. We were delighted that in 2023, the SickKids 22q clinic team restarted in-person transition clinics with our Clinic for the first time since 2019.

This year marks a special milestone for our clinical research work to better understand 22q. In total, over the past three decades, we have now seen over 500 patients with 22q11.2DS! We continue to have more experience with adults with 22q11.2DS than any other centre in the world.

The leadership of our Dalglish Family 22q Clinic was front and centre in another major achievement of the past year – the publishing of three international clinical practice guidelines for 22q11.2DS. This multi-year effort involved dozens of colleagues from four continents, and included significant contributions from our staff, learners, and colleagues at UHN.

Our excellent research publication record continues, adding to other high-profile events such as 22q at the Zoo and the Lighting of the CN Tower, each helping to raise awareness about 22q and our Clinic.

In this newsletter you will find an update of our exciting work. On behalf of the entire team at the Dalglish Family 22q Clinic, thank you for letting us be part of your journey.

**If we have not seen you in a while, please call Radhika at (416) 340-5145 or email her at 22q@uhn.ca to book an appointment.**

Yours sincerely,

**Dr. Anne S. Bassett**
Clinic Director
Dalglish Family Chair in 22q11.2 Deletion Syndrome

Toronto ACHD PROGRAM

The Dalglish Family 22q Clinic

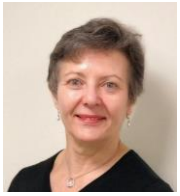
UHN Toronto General Toronto Western Princess Margaret Toronto Rehab Michener Institute

# Our staff

## 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

Physician for people with intellectual disability

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

## Lisa Palmer

Registered 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

## Radhika Sivanandan

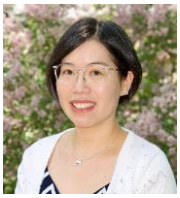
Patient Flow Coordinator

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

## Joanne Loo

Education and Communication Officer

- 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 is 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

## Residents

**Dr. Sarah (Voll) Malecki, MD**
Resident in General Internal Medicine

**Dr. Christina Blagojevic, MD**
Resident in Internal Medicine

## Students

**Sabrina Cancelliere, HBSc**
Medical student

**Nikolai Reyes, MD**
Neurologist, Graduate Student

**Adonis Ng, BMSc**
MPH Candidate

## Volunteers

Tala Maatouk

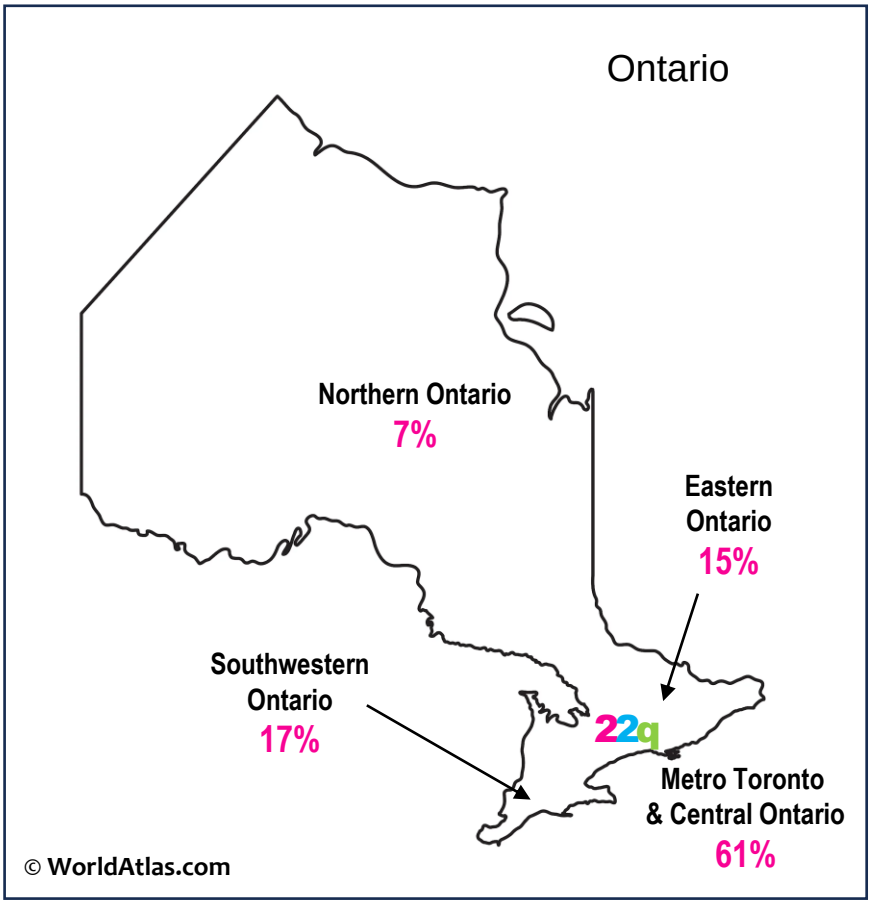
Fatima Khan

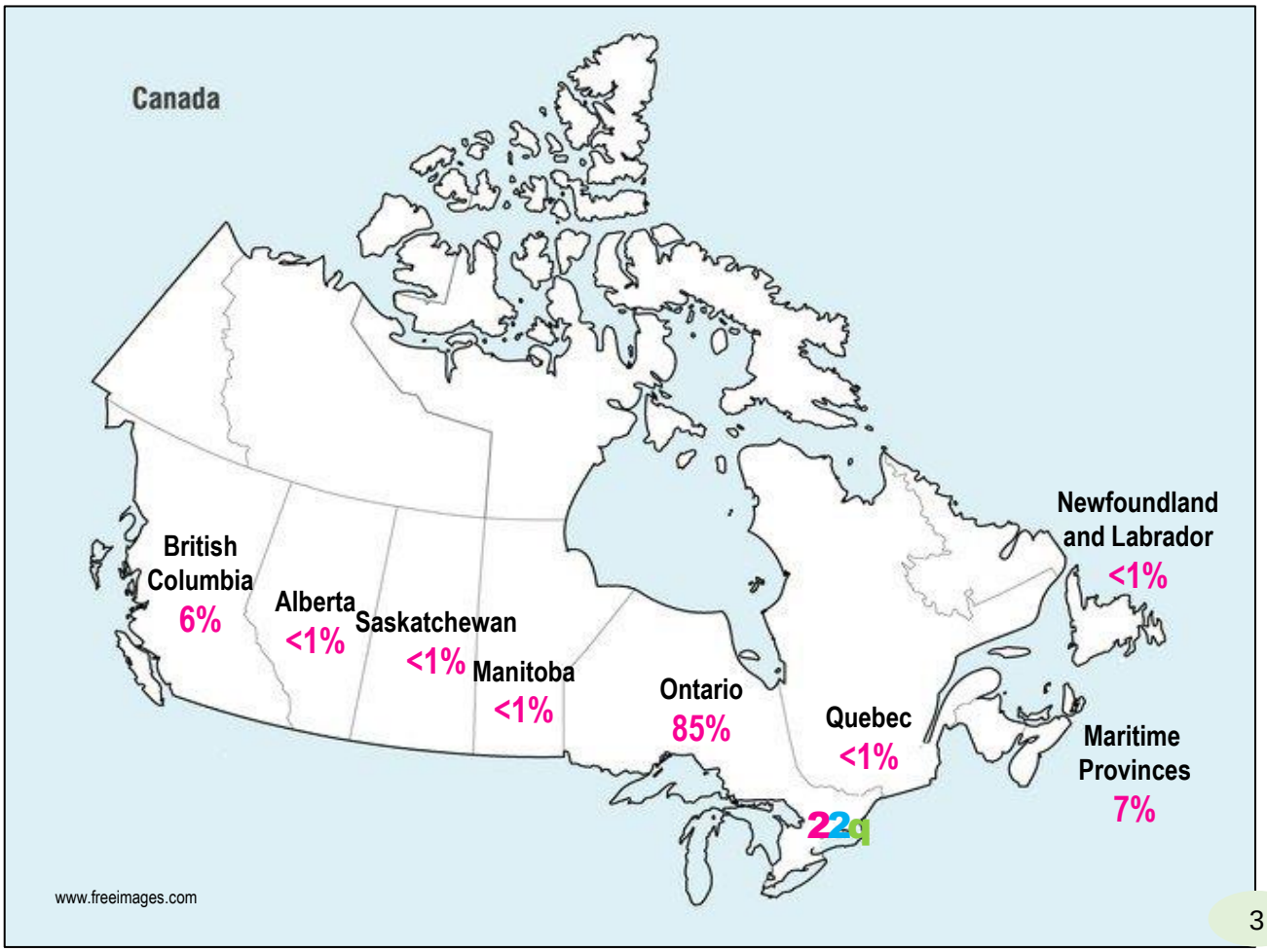
# Where our patients come from

While over 85% of the 500+ adults with 22q11.2DS we have seen over the years are from Ontario, our Clinic and our research program in fact serve Canadians from coast to coast. The maps show where our patients come from.

Most often patients and their families and caregivers come to our Toronto Clinic for in person appointments. Other times they may receive “virtual” care, usually by video conference.

We do our best to make sure that appointments match the needs of the individual.



# Our collaborators

**The Toronto Adult Congenital Heart Disease Program at the Peter Munk Cardiac Centre**

Dr. Rafael Alonso-Gonzalez, MD, MSc, FESC
Medical Director and Fellowship Director

Dr. Rachel Wald, MD, FRCPC
Research Director

Dr. Sara Thorne, MBBS, MD, FRCP
Director of ACHD Trainee Education

Dr. Erwin Oechslin, MD, FRCPC
Dr. Candice Silversides, MD, FRCPC
Dr. Lee Benson, MD, FRCPC, FACC, FSCAI
Dr. Jack Colman, MD, FRCPC, FACC
Dr. Eric Horlick, MDCM, FRCPC, FSCAI
Dr. Luc Mertens, MD, PhD
Dr. Mark Osten, MD, FRCPC
Dr. Lucy Roche, MD, MBChB, MRCPCH(UK)
Dr. Danielle Massarella, MD, MSc

## Collaborating doctors and researchers

### Ontario

Dr. Danielle Andrade, MD, FRCPC
Neurologist
Toronto Western Hospital

Dr. Anthony Lang, MD, FRCPC
Neurologist
Toronto Western Hospital

Dr. Connie Marras, MD, PhD, FRCPC
Neurologist
Toronto Western Hospital

Dr. Erwin Oechslin, MD, FRCPC
Cardiologist
Toronto General Hospital

Dr. Candice Silversides, MD, FRCPC
Cardiologist
Toronto General Hospital

Dr. Satya Dash, MD, PhD, FRCP (Edin), FRCPC
Endocrinologist
Toronto General Hospital

Dr. Christine Cserti-Gazdewich, MD, FRCPC
Hematologist
Toronto General Hospital

Dr. Jacob Vorstman, MD, PhD
Child Psychiatrist
Hospital for Sick Children

Andrea Shugar, Cheryl Cytrynbaum, Simone Fischbach, Paula Klaiman
22q Clinic & Cleft Palate Clinic
Hospital for Sick Children

Ryan Yuen, PhD, Christian Marshall, PhD, FACMG, FCCMG, and Stephen Scherer, PhD
Geneticists
Hospital for Sick Children

Dr. Greg Costain, MD, PhD
Medical Geneticist
Hospital for Sick Children

### Atlantic Canada

Dr. Sarah Dyack, MD, FRCPC
Medical Geneticist
Halifax, NS

Kathleen Hodgkinson, PhD
Geneticist
St. John's, NF

### International

**Europe**

Ann Swillen, PhD
Child psychologist
Leuven, Belgium

Joris Vermeesch, PhD
Geneticist
Leuven, Belgium

Dr. Therese van Amelsvoort, MD, PhD
Psychiatrist
Maastricht, Netherlands

**Asia**

Dr. Brian Chung, MBBS, MSc, MD, DCH, MRCPCH, FHKAM, FRCPCH, FCCMG
Medical Geneticist
Hong Kong SAR, China

### USA

Donna McDonald-McGinn, MS, LCGC
Genetic Counselor
Clinical Professor of Pediatrics
Director, 22q Clinic
Children's Hospital of Philadelphia (CHOP)
Philadelphia, PA

Dr. Linda Brzustowicz, MD
Geneticist
New Brunswick, NJ

Bernice Morrow, PhD
Geneticist
New York, NY

### South America

Dr. Gabriela Repetto, MD
Medical Geneticist
Santiago, Chile

# Knowledge translation in action: Updated clinical practice recommendations published!

We hope you are as excited about these 22q guidelines as we are, and we look forward to sharing further advances as we move forward.

**Knowledge translation** means taking what we have learned through research and applying this knowledge to clinical practice. And we have done exactly that!

In March 2023, we published new clinical practice recommendations for the care of adults and children with 22q11.2 deletion syndrome ("22q"). These are based on the most up-to-date knowledge available, and on the very first guidelines for 22q published in 2011 (for children and adults) and 2015 (for adults), initiatives also led by Dr. Bassett. You can find the two new papers in the premier clinical genetics journal, Genetics in Medicine (see the next page for links).

The 2023 publications are a result of major efforts across many years. Here are the numbers:

- 30 years of medical research reports on 22q were obtained and reviewed
- 2,318 articles about clinical care for 22q were selected and sent to experts in a variety of fields.
- Experts from 4 continents were involved in writing the new recommendations
- Also, 1 brand new paper was added to the two main papers – the first ever recommendations on prenatal care for 22q
- Together, these 3 papers will have an impact on clinical care for patients and families worldwide
- Over 95% of patients at our Dalglish Family 22q Clinic participate in clinical research. And the 2023 guidelines for adults with 22q are packed with new knowledge, much of it based on our pioneering research − in partnership with our patients and families over many years.

This is a great example of how research and clinical care come full circle in knowledge translation – we learn from every patient we see, and the insights gained benefit people with 22q and their families − at our Clinic, and around the world.

**UHN Team Members involved**

**5 Dalglish Family 22q Clinic team members**

- Anne Bassett, MD, FRCPC – Psychiatrist and Director, Dalglish Family 22q Clinic (project leader and senior author), Senior Scientist, Toronto General Hospital Research Institute (TGHRI)
- Erik Boot, MD, PhD – Former Dalglish Fellow (project leader and senior author)
- Lisa Palmer, BEd, MSW, RSW – Registered Social Worker, Dalglish Family 22q Clinic
- Ania M. Fiksinski, PhD – Psychologist, former graduate student, Dalglish Family 22q Clinic
- Joanne C.Y. Loo, PhD – Education and Communication Officer, Dalglish Family 22q Clinic

**7 UHN Specialists affiliated with the Dalglish Family 22q Clinic**

- Danielle M. Andrade, MD, FRCPC – Neurologist, Adult Genetic Epilepsy Program, Krembil Clinician Investigator
- Anthony E. Lang, MD, FRCPC – Neurologist and Director, The Edmond J. Safra Program in Parkinson's Disease and the Morton and Gloria Shulman Movement Disorders Clinic, Senior Scientist, Krembil Research Institute
- Nikolai Gil D. Reyes, MD – Clinical Fellow, The Edmond J. Safra Program in Parkinson's Disease and the Morton and Gloria Shulman Movement Disorders Clinic
- Erwin Oechslin, MD, FRCPC – Cardiologist, Toronto Adult Congenital Heart Disease Program
- Candice Silversides, MD, FRCPC – Cardiologist, Toronto Adult Congenital Heart Disease Program, Clinician Investigator, TGHRI
- Christine Cserti-Gazdewich MD, FRCPC – Hematologist and Transfusion Medicine Specialist, Clinician Investigator, TGHRI

**1 UHN information specialist**

- Ani Orchanian-Cheff, BA, MISt – Information Specialist, Library and Information Services

# Read our updated guidelines

We encourage you to bring or send a copy (or a link) of the relevant 22q11.2DS guidelines to your healthcare providers.

**How to download the guidelines**

On the electronic copy of this newsletter, you can click on the links below.

On the paper copy of this newsletter, use the camera of a smart phone to scan the QR code

If you need a paper copy, please email us at 22q@uhn.ca or call us at (416) 340-5145.

**Updated clinical practice recommendations for managing adults with 22q11.2 deletion syndrome**

**Traditional Chinese version (recommendations for adults)**

管理 22q11.2 缺失症候群成人患者的更新臨床實用建議

**Updated clinical practice recommendations for managing children with 22q11.2 deletion syndrome**

**Prenatal screening and diagnostic considerations for 22q11.2 microdeletions**

# At a glance:

# 22q11.2 deletion and duplication

## 22q11.2 deletion syndrome (22q11.2DS)

1. Caused by a missing piece on chromosome 22q. There is one copy (instead of 2) of several of the genes that usually tell the body how to form during early development and how to function throughout life.
2. Happens in about 1 in every 2,148 babies. Usually arises as a new event in a family. Fewer than 1 in every 10 patients has a parent who also has the deletion.
3. Not enough people (including those in healthcare) know about 22q11.2 deletion syndrome. The condition used to be known by other names, such as DiGeorge syndrome & velocardiofacial syndrome.
4. Common, often treatable features include:
   - Birth defects of the heart and/or palate
   - Hypocalcemia (low calcium levels)
   - Thyroid conditions, diabetes risk
   - Ear, nose, and throat conditions
   - Sleep apnea (see next page)
   - Scoliosis (curvature of the spine)
   - Learning disabilities
   - Anxiety, schizophrenia
   - Seizure risk
   - Early onset Parkinson's
5. Each person (even if in the same family) can have their own pattern of 22q11.2DS features and severity of each.
6. For the person who has the deletion, there is a 50% chance that the deletion will be passed on to a baby at each pregnancy.

## 22q11.2 duplication syndrome (22q11.2DupS)

1. Caused by an extra piece on chromosome 22. There are 3 copies (instead of 2) of several of the genes that usually tell the body how to form during early development and how to function throughout life.
2. Happens in about 1 in 4000 babies, but not many people know about it. Often runs in the family. The majority (well over half) of patients have a parent with the same 22q duplication.
3. Much less is known about the 22q11.2 duplication syndrome compared to the 22q11.2 deletion syndrome.
4. Common features include:
   - Birth defects of the heart
   - Developmental delays
   - Autism spectrum disorder
   - Attention deficit hyperactive disorder (ADHD)
   - Seizure risk
   - Vision and hearing problems
   - Palate issues
5. Each person (even if in the same family) can have their own pattern of 22q11.2DupS features and severity of each.
6. For the person who has the duplication, there is a 50% chance that the duplication will be passed on to a baby at each pregnancy.

A version of this page was published in the September 2023 Newsletter of the International 22q11.2 Foundation.

# Research spotlight:

## Adult-onset obstructive sleep apnea in 22q11.2 deletion syndrome.


**Cancelliere S**, **Heung T**, Fischbach S, Klaiman P, **Bassett AS**
Sleep Medicine 104:49-55, 2023 (doi: 10.1016/j.sleep.2023.02.010)

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### Obstructive Sleep Apnea (OSA)

OSA happens when muscles in the throat become too relaxed during sleep and prevent proper breathing. The brain is starved of oxygen, so the person wakes up every once in a while to breathe again. This leads to very broken sleep and tiredness during the day.

If you think you may have OSA, please check with your doctor to see if you need a sleep study.

**Obstructive sleep apnea (OSA)**, or upper airway blockage during sleep, is a fairly common sleep problem. Because of low oxygen levels, this condition can affect the way the brain and heart work. Often, the person with OSA is tired and may have mood changes. OSA is a treatable condition.

In a pioneering study, our team found that OSA was quite common in adults with 22q. At an average age of only 32 years, about 1 in every 10 adults had OSA. Of those who had had a sleep study, over half were found to have OSA.

All of the usual risks for OSA - such as obesity, older age, asthma, and male sex - were also risk factors in 22q. However, a new finding was that having had certain types of surgery of the palate may also add to the risk of adult-onset OSA in 22q. The palate is the roof of the mouth, and it separates the mouth from the nose. Many children with 22q have palate and speech problems, and surgery improves the way their palate works and make their words easier to understand. It appears that in some individuals this helpful childhood surgery may increase the risk of OSA in adult years. We found this was the case even when taking into account the effects of other risk factors. Next, we want to find out why most people with 22q who have palate surgery do not develop OSA.

**Sleep apnea is treatable with continuous positive airway pressure (CPAP).** In fact, our team found that the majority of adults with 22q who were prescribed CPAP were able to use this treatment in the long term. This helped to improve energy and mood, and often helped people lose weight, too.

## Adult height in 22q11.2 deletion syndrome

**Heung T**, **Conroy B**, **Malecki S**, **Ha J**, **Boot E**, **Corral M**, **Bassett AS**
Genes 13:2038 (pages 1-8), 2022 (doi: 10.3390/genes13112038)

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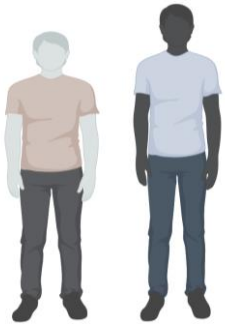

Research questions

- How tall are adults with 22q11.2DS compared to those in the general population?
- Are there features that might predict short stature in 22q11.2DS?

Study population

- 397 adults with 22q11.2DS
- Confirmed 22q11.2 deletion

Results

**Adult height** varied from person to person. Adults with 22q11.2DS are a bit shorter on average than those in the general population.

**Short stature** (shorter than 97% of the general population):

- Was found in about 1 in every 5 adults with 22q11.2DS
  - About 7 times more common than in the general population
- May be more common in adults:
  - With more severe intellectual disabilities
  - Born with more severe heart disease
- May be less common in adults with a smaller 22q11.2 deletion.

# Reproductive outcomes in adults with 22q11.2 deletion syndrome

**Palmer LD*, McManus Z*, Heung T*, McAlpine G, Blagojevic C, Corral M, Bassett AS**
(*These authors contributed equally.)
Genes 13:2126 (pages 1-9), 2022
(doi: 10.3390/genes13112126)

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Our research team studied the history of pregnancies in individuals with 22q, none with severe intellectual disability. There were a total of 156 pregnancies for 63 adults (15 male, 48 female), with 157 pregnancy outcomes because one pregnancy involved twins.

Pregnancy outcomes

- 94 (60.3%) live births
- 34 (21.2%) miscarriages
- 22 (14.1%) terminations
- 7 (4.5%) stillbirths

Main findings

- Pregnancy loss is an important health issue for adults with 22q11.2DS, and far more common compared to the general population.
- For a mother with 22q and severe heart disease, the chance of pregnancy loss may be greater.

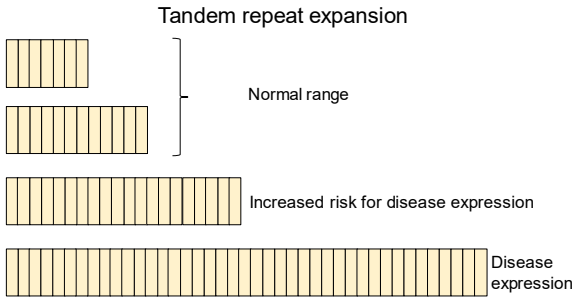
Having supports in place before, during, and after pregnancy is essential. Please call our Clinic if you have any questions related to reproductive health and/or family planning considerations.

# Genome-wide tandem repeat expansion is a new type of genetic risk for schizophrenia

Schizophrenia is a serious but treatable illness that affects about 1 every 100 people in the general population, and about 1 in every 4 to 5 individuals with 22q11.2 deletion syndrome (22q11.2DS). The illness involves major changes in thinking (e.g., being out of touch with reality), emotional state, and a decrease in the ability to function (e.g., at work or school, with other people, with self-care). Researchers are trying to learn more about the many genetic factors that affect the risk of developing this illness in order to further improve treatments.

Tandem repeats are a type of genetic sequence where there are stretches of repeated “letters” of DNA. Sometimes these repeats can get longer when they are passed from one generation to the next. As a repeat sequence expands, it may change a gene’s function and cause or increase the risk for developing an illness. This is a known mechanism for several brain diseases.

In two studies – the first Canadian, and the second international – our research group analyzed the DNA of adults with and without schizophrenia. The results show that individuals with schizophrenia have longer or expanded tandem repeats that are rarely found in those without this illness. The genes that are affected by these expansions are involved in communication between nerve cells in the brain. These findings show for the first time that the expansion of rare tandem repeats may be part of the genetic causes of schizophrenia in the general population. Next, we plan to study this in individuals with 22q11.2DS.




**Genome-wide tandem repeat expansions contribute to schizophrenia risk**

[Download this article for free]

Mojarad BA, Engchuan W, Trost B, Backstrom I, Yin Y, Thiruvahindrapuram B, Pallotto L, Mitina A, Khan M, Pellecchia G, Haque B, Guo K, **Heung T**, **Costain G, Scherer SW, Marshall CR,** Pearson CE, **Bassett AS**, **Yuen RKC**.
Mol Psychiatry. 2022 Sep;27(9):3692-3698.
(doi: 10.1038/s41380-022-01575-x)

**Rare tandem repeat expansions associate with genes involved in synaptic and neuronal signaling functions in schizophrenia**

[Download this article for free]

Wen J, Trost B, Engchuan W, Halvorsen M, Pallotto LM, Mitina A, Ancalade N, Farrell M, Backstrom I, Guo K, Pellecchia G, Thiruvahindrapuram B, Giusti-Rodriguez P, Rosen JD, Li Y, Won H, Magnusson PKE, Gyllensten U, **Bassett AS**, Hultman CM, Sullivan PF, **Yuen RKC**, Szatkiewicz JP.
Mol Psychiatry. 2023 Jan;28(1):475-482.
(doi: 10.1038/s41380-022-01857-4)

# What makes it more likely for adults with 22q11.2DS to have high levels of triglycerides?

**Polygenic risk for triglyceride levels in the presence of a high impact rare variant**
**Ying S**, **Heung T**, Thiruvahindrapuram B, Engchuan W, Yin Y, **Blagojevic C**, Zhang Z, Hegele RA, **Yuen RKC**, **Bassett AS**.
BMC Med Genomics, 2023. 16, 281 (doi: 10.1186/s12920-023-01717-2)

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**Triglycerides** are a type of lipid (fat). Your body stores and releases energy as triglycerides in fat cells.

**Hypertriglyceridemia** means having a triglyceride level in the blood that is too high. This condition increases the risk of:

- Obesity
- Diabetes
- Heart disease

Having a **22q11.2 deletion** and being male can increase the risk of having hypertriglyceridemia. These are not changeable. However, we can lower the triglyceride level. How? With exercise / activity, and with eating and drinking less of certain foods (especially sugars and starches). Here's to healthy eating!

In our 2022 publication, we reported that having a 22q11.2 deletion and being male increase the risk of having high levels of triglycerides (a condition called hypertriglyceridemia). Chromosome 22q11.2 does not contain any genes that are directly related to hypertriglyceridemia. So, what makes this condition to be more likely in adults with 22q11.2DS?

In this study, our team checked if certain common genetic variations outside of the chromosome 22q11.2 region can predict the risk of having higher levels of triglycerides. In 157 adults with 22q11.2DS and detailed genetic data, we found that these common variants, together with obesity and being male, could predict the risk of high triglyceride levels quite well.

Of course, lipid levels (including triglycerides) can easily be measured in a blood test. In the future, however, children found to have the variants that increase their risk of hypertriglyceridemia may be encouraged to adopt healthy diets and active lifestyles early on in life to lower the risk.

# Neurodevelopmental functioning in individuals with a 22q11.2 microduplication

Drmic IE, MacKinnon Modi B, McConnell B, Jilderda S, Hoang N, Noor A, **Bassett AS,** Speevak M, Stavropoulos DJ, Carter MT.
Am J Med Genet A. 2022 Oct;188(10):2999-3008.
(doi: 10.1002/ajmg.a.62916)

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Payment or subscription is required for this article. Please email us at 22q@uhn.ca if you need help.

There are very few studies on 22q11.2 duplication syndrome (22q11.2DupS) that include adults. The 8 adults (out of 22 participants) in this study with the duplication were all identified after a family member (the proband) was confirmed to have 22q11.2DupS.

This initial study examined how 22q11.2DupS individuals functioned. Even though the researchers found symptoms of attention deficit and anxiety, the affected individuals in this study functioned, on average, at a similar level compared to the general population.

Larger studies are needed to learn more about 22q11.2DupS, especially in adults, as associated health conditions may vary greatly from person to person.

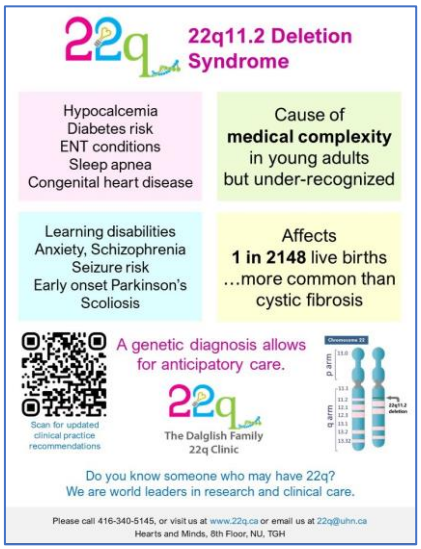
Individuals with 22q11.2DupS

- IQ and learning at school:
  - Within average range
  - Slightly lower in probands (individuals who are the first in the family to be diagnosed with 22q11.2DupS)
- Dealing with day-to-day needs
  - Comparable to same-aged individuals in the general population
- Feeling; acting and reacting to the environment and to people:
  - Attention deficit disorder and anxiety are more common than in the general population
  - Only one participant had autism spectrum disorder (a low rate compared to previous studies of 22q11.2DupS)

# Increasing Awareness


**22q at the Zoo**

- Annual social event for 22q11.2 deletion and duplication
- Most recent Toronto event took place on June 4, 2023
- Tracy, Sam, and Joanne L. shared our newsletter and the clinical practice recommendations with families




**Raising awareness for 22q11.2DS on UHN computer screens**

- November 15 to 18, 2023
- Staff at 5 UHN healthcare institutions saw this infographic
- This poster is available on our website.

**Raising awareness for 22q11.2DS by lighting the CN Tower in red**

- November 22, 2023
- To find out how to apply for the lighting of other buildings or sites across Canada on November 22, 2024, please email us at 22q@uhn.ca.

## Asking for an increase in Canadian research funding

Research is the key to understanding health conditions and developing new treatments. However, the Canadian Institutes of Health Research (CIHR), the federal agency that funds health research, does not receive enough year-to-year support. Canadian health research **has not been funded adequately for many years** and lags behind that of other G7 countries.

As a result, Canadian researchers spend more time and effort now than ever before in requesting CIHR grant funding. Our latest 22q grant took 4 tries, and over 2 and a half years of persistence, to be successful! But we will now be able to include siblings in our research to help us better understand 22q.

Researchers across the country began a letter-writing campaign for increasing research funding, and we joined in. **Dr. Bassett mailed a letter to the prime minister** to give him an **overview of 22q11.2DS** and to ask him to **increase funding to CIHR**.

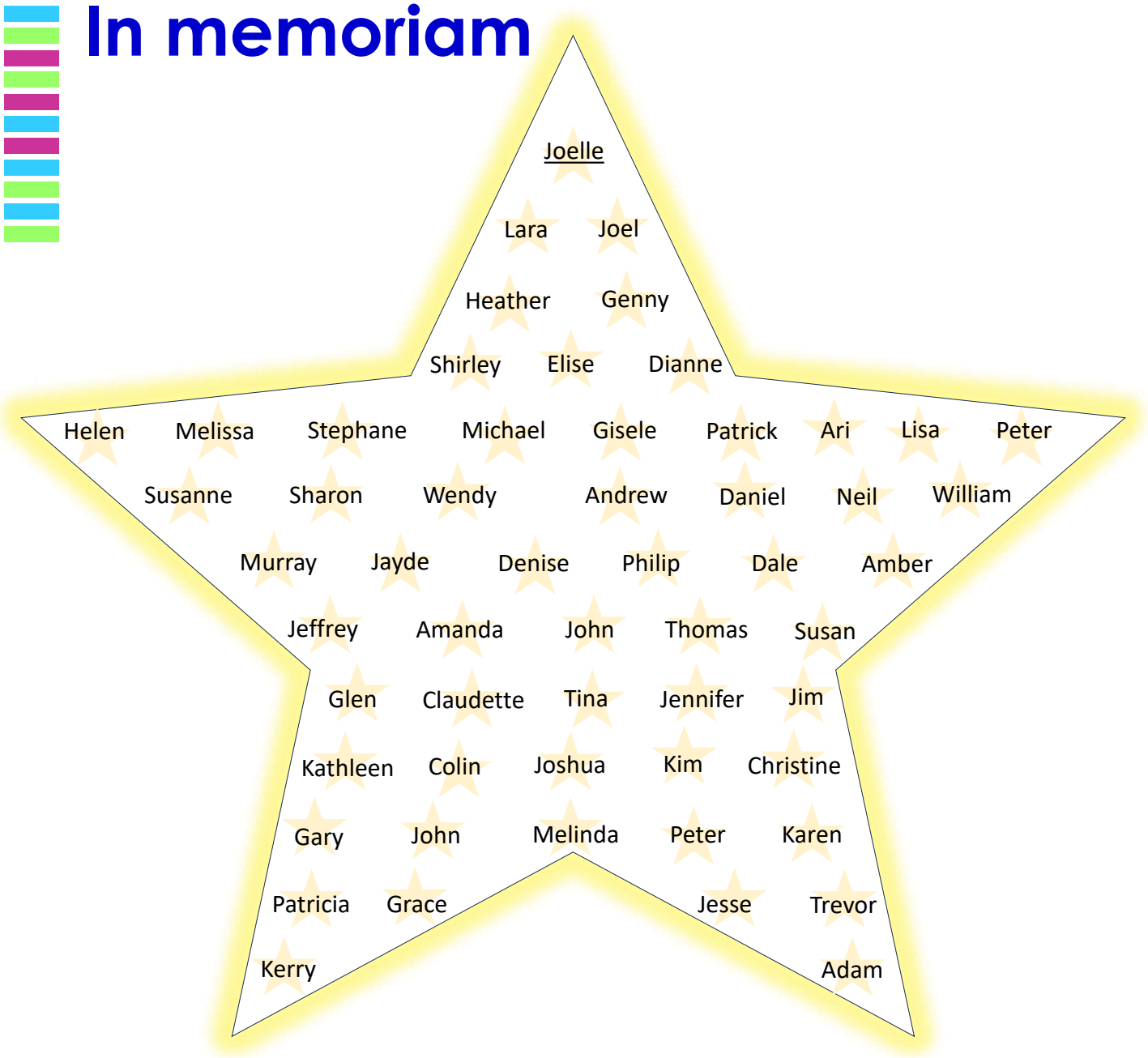
You can join this effort to ask for an increase in research funding by signing this petition on change.org using the link below.

Increasing CIHR research funding will help all families.

**Thank you for your support!**

**We Ask the Canadian Government to Increase CIHR's Budget**

# In memoriam




These are the members of our 22q family we have lost over the past 25 years, including 1 individual in 2023.

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 stars that will forever shine!

# We invite you to…

## Join our exciting new online nutrition programs!

Once again, this year our Clinic dietitian, Samantha D'Arcy, is running online nutrition programs for our patients, family members and caregivers. Below are some of the exciting topics discussed. Please note, at this time, these programs are only available for Ontario residents, due to hospital policy. We will send out the dates and times for these programs early in the new year, so please check your mail and email for more information.

### Meal Planning Support Group

(for individuals with 22q only)

This group program is about meal planning. We will meet online each week to learn a new tip and set a meal planning goal for the following week. There will be follow-up meetings after the first month, to help participants stay motivated.

### All About Appetite and Cravings

This online program is about high appetite and cravings: what we know about appetite in people with 22q and strategies to keep your cravings in check.

### All About Calcium

This online program is about calcium: why it is important for people with 22q, and how to get healthy levels from the foods you eat.

### Healthy Weight in 22q

This online program is about healthy weight: what do we know about weight and how it changes for people with 22q and how to lead a healthy lifestyle and have a positive outlook about your weight.

# Write to us about your experience

We always try to understand and help our patients and families, and we want to hear about your experience with 22q and the healthcare system. Please take some time to complete this survey to tell us what barriers you may be facing as you manage your health, and how you hope your healthcare providers can improve your care.

*Both patients and caregivers are invited to participate. Please scan the QR codes with your phone camera, or click the link to complete our short survey.*

**Survey for patients**

PATIENT SURVEY

**Survey for caregivers**

CAREGIVER SURVEY

**Thank you for your time!**

# We also invite you to…
# Donate and support our work

While the Ontario Ministry of Health supports the basic operations of our Clinic, we receive **no funding for the education, outreach, and many other things we do for patients with 22q and their families**. There are 3 options to donate, and the UHN Foundation manages all donations for our Clinic. **Thank you** for your support!

**Donate online**

**Donating by mail or fax Using this form**

**The Honour Your Hero program**


# Join our research

Without your help, we would know nothing about 22q11.2 deletion and duplication syndromes, and how best to care for people with these conditions. Over the years, **hundreds of our patients with 22q differences** – and others around the world – have helped doctors and scientists to learn about 22q differences.

We are one of the very few centres to focus on adults with 22q differences – so every person who participates is precious. We invite you to take part. And do let us hear from you about how we could make this process better.

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

# Contact us!

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22q
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**

The illustrations on pages 7, 8, 10, and 13 were created with BioRender.com. The pink/blue ribbon on page 9 was retrieved from Wikipedia.

To download a digital copy of this newsletter or previous ones, please scan the QR code to reach:
https://22q.ca/aboutus/our-newsletter/