

# 22q & Friends

## 2024 Newsletter

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

2024 overall has been a glorious year for our small but mighty team! We continued to care for adults with 22q11.2 differences and their families and caregivers. Mostly we see those with 22q11.2 deletion syndrome (22q11.2DS), but also those with 22q11.2 duplications or other complex genetic conditions. This past year, we welcomed 50 new patients to our Clinic, and we continue to follow hundreds of others. We provide the time to listen to - and address - the concerns of patients and caregivers.

Our efforts have not gone unnoticed. I was honoured to be named to the Order of Canada at the end of December 2023 (page 3). Our Clinic was also honoured this year - by an international **Special Service Award** from the 22q11.2 Society at the 13<sup>th</sup> Biennial International 22q11.2 Conference in July (page 6). At this meeting, our team members also received a Junior Investigator Award and multiple Top Presenter Awards. In addition, two of our trainees received prestigious grants this year (page 7).

Our Consultant Psychiatrist, Dr. Maria Corral, retired at the end of June this year. We will all miss her, as she has contributed so much to our Clinic! Dr. Corral has shared some of her thoughts about her many years with us in an interview (pages 4 to 5).

We made further progress with our research and have published several important papers this year. In this newsletter, we are highlighting our report on the healthcare burden of adults with 22q11.2DS, as well as a special commentary about our proposed healthcare model for adults with complex genetic conditions (page 8).

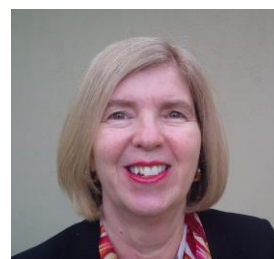
In 2023, we published three international clinical practice guidelines for 22q11.2DS. Our team members led the multi-year effort to develop these. In 2024, we focused on translating the guidelines to make them more accessible for local and international audiences. The guidelines for adults are now available in French, Spanish, and Traditional Chinese (page 9), with versions in other languages in progress.

We are also delighted to bring you a new educational video about 22q11.2DS, as well as special programming for 22q11.2DS from our dietitian, Samantha D'Arcy (page 10).

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

Yours sincerely,

**Anne S. Bassett,**  
CM, MD, FRCPC  
Clinic Director and  
Inaugural Dalglish  
Family Chair in  
22q11.2 Deletion  
Syndrome



# Our Clinic and affiliated staff



[Dr. Anne Bassett](#)  
Psychiatrist and Clinic Director



[Dr. Maria Corral](#)  
Psychiatrist (Retired)



[Dr. Erik Boot](#)  
Physician for people with  
intellectual disability  
Research Associate



[Dr. Nikolai Gil D. Reyes](#)  
Neurologist  
Masters Student



[Lisa Palmer](#)  
Registered Social Worker



[Samantha D'Arcy](#)  
Registered Dietitian



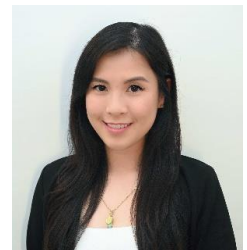
[Radhika Sivanandan](#)  
Patient Flow Coordinator



[Joanne Loo](#)  
Education and  
Communication Officer



[Tracy Heung](#)  
Clinical Research Analyst



[Joanne Ha](#)  
Clinical Research Analyst

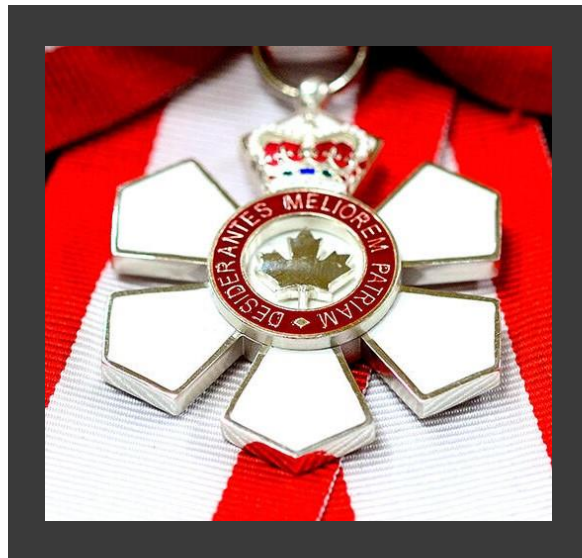
Please visit the [About Us](#) section of our Clinic website for a complete list of our staff, residents, students, and collaborators.

# Dr. Anne Bassett named to the Order of Canada

Our Clinic Director, [Dr. Anne Bassett](#), was appointed as a [Member of the Order of Canada](#) on December 28<sup>th</sup>, 2023. She was honoured “for her research and clinical contributions to advancing our understanding of the genetics of schizophrenia, particularly in the identification and characterization of genetic subtypes of the disorder”. This includes her three decades of clinical care of adults with 22q11.2 Deletion Syndrome (22q11.2DS, or “22q”), and her pioneering research to better understand this important genetic condition.

Dr. Bassett is the inaugural Dalglish Chair in 22q11.2 Deletion Syndrome and the Director of the Dalglish Family 22q Clinic at the Toronto General Hospital. Our Clinic provides ongoing care that is devoted to adults with 22q11.2DS of any age, from late teens through older age ranges. Along with many other possible associated conditions, an adult with 22q is about 20 times more likely than someone in the general population to develop schizophrenia, a serious but treatable psychiatric illness. Our multi-disciplinary and family/caregiver approach focuses on anticipatory care tailored to the individual, and partnered with clinicians in the community. Our clinical research enables us to learn from our patients their families, so that we can apply the new knowledge gained as we move forward. The aim is to help adults with 22q achieve the best health and quality of life possible.

We are very excited that Dr. Bassett’s contributions are being recognized by this special honour! We are currently waiting for the Office of the Governor General to announce the date of the investiture ceremony. These ceremonies are still back-logged due to ongoing delays related to the COVID-19 pandemic.



The motto of the Order of Canada is: **DESIDERANTES MELIOREM PATRIAM** – Latin for “**They desire a better country**”.

## More information

- [Official Announcement on December 28<sup>th</sup>, 2023](#)
- [List of Appointees](#)
- [U of T News](#)
- [UHN News](#)
- [Order of Canada – Official website of the Governor General of Canada](#)

Dr. Bassett is hopeful that this honour will help to increase awareness about 22q – and that everyone in our 22q community shares in this!

# Dr. Maria Corral's Retirement Interview

After 8 years as a key member of our Dalglish Family 22q Clinic team, and after 40 years of working as a leading Canadian psychiatrist, our colleague, **Dr. Maria Corral**, retired at the end of June 2024. We will all miss her, and the outstanding skills, dedication, and wisdom she has brought to patient care! Here is our interview with Dr. Corral soon after her retirement.



**Interviewer:** What were some of the most memorable / impactful moments from your practice in our clinic?

**Dr. Corral:** There are many memorable moments that come to mind; so many that I could not list them all. However, at the top of the list is having the opportunity to work with the talented, cohesive and professional staff at the Clinic. Each individual member is an expert in their field, and generous in sharing their knowledge. The nature of our work requires cooperation, and I would say this is the most unified and unstinting team with whom I have had a chance to work.

The most impactful moments from my time at the Clinic have been witnessing the improvement in our patients' health and

quality of life. So many patients and caregivers have said they felt tremendous relief and reassurance after meeting with members of the Clinical Team, who described the nature of 22q11.2 deletion syndrome (22q11.2DS or 22q) in language they understood. The Teams' ability to inspire confidence in patients and their caregivers helped ensure treatment and management recommendations were more likely to be followed over the patient's lifespan.

**Interviewer:** What changes have you witnessed in the past 8 years?

**Dr. Corral:** I have witnessed a tremendous increase in knowledge about 22q because of international clinical research, and our



The text on the crystal testimonial reads:  
Dr. Maria Corral – On the occasion of your retirement  
Thank you for being the greatest gift to our Team,  
our Patients, and their Families  
2016 – 2024 The Dalglish Family 22q Clinic

Clinic's leading contributions to that knowledge, such as finding that Type 2 diabetes occurs on average 20 years earlier in individuals with 22q. This research finding can then be incorporated into treatment guidelines for clinical care. Similarly, knowing that anxiety and schizophrenia are common in persons with 22q, we can screen patients regularly, and treat these conditions early, resulting in tremendous improvements in patients' quality of life.

Another big change at the Clinic has been the implementation of an electronic healthcare system at the Clinic, as part of the Toronto General Hospital and University Health Network. We look forward to this one day being connectable to other Ontario hospitals.

**Interviewer:** What future changes do you think are necessary in the field of caring for adults with 22q?

**Dr. Corral:** Most of our patients with 22q and their caregivers need greater medical, social and financial support. This is a complex, lifelong condition that requires long term support from family physicians, specialist physicians, allied health professionals in the hospital and community, and greater financial resources to enable patients to live with this condition throughout their lives. Medical students and residents

need to be taught about this condition, given that it is a relatively common genetic condition. And they need clinical exposure to people with 22q early in their studies. Hopefully, this would increase their interest and ability to care for patients with 22q.

**Interviewer:** What message would you like to share with your patients as you retire?

**Dr. Corral:** It has been an absolute honour and privilege to have worked with you over the years.

**Interviewer:** What message would you like to share with your colleagues as you retire?

**Dr. Corral:** I will miss your collegiality, support and weekly presence. It has been an honour to have worked and learned side by side with you over these last eight years.

**Interviewer:** What advice would you share with current psychiatry / medical students starting their careers?

**Dr. Corral:** I would advise them to choose elective rotations that expose them to patients with complex and chronic conditions while they are students and can benefit from the clinical experience, knowledge and supervision of senior clinicians. While caring for patients with chronic conditions can be challenging, it is also extremely rewarding work.

I also strongly recommend that they see and follow as many patients as they can. Strive to know your patients. Most mental health conditions are chronic and/or recurrent, so long term follow-up of patients provides the best understanding of the course of these conditions over a person's lifetime. The competencies required to pass an exam in medical school and residency are not the same as those needed to effectively help patients recover and remain well. Be curious, understanding and forgiving.

\*\*\*\*\*

We thank Dr. Corral for sharing her thoughts with us, and we wish her all the best!

# Awards at the International 22q Conference

Our participation in the [13<sup>th</sup> Biennial International 22q11.2 Conference](#) in Óbidos, Portugal in July was a major highlight. Our team delivered 18 presentations, demonstrating our leadership and commitment to sharing the latest advances in the field. Our aim now is to publish papers reporting our findings for the international community. We were honoured to receive multiple awards at the meeting.

## Special Service Award

**Dr. Anne Bassett** and our Clinic team received the Special Service Award at the conference in Portugal. The award was inaugurated in 2018 to commemorate the 20<sup>th</sup> anniversary of the International 22q11.2 Society and to recognize the long-term dedication of a group. This award, only given once before, is presented to a centre that demonstrates outstanding, longstanding, exemplary and unwavering commitment, and contributions to the chromosome 22q11.2 community.

This recognition reflects not only our clinical achievements but also our dedication to the broader 22q community. We are honoured to receive this international award, and we thank all of our patients and families for your support.



Dr. Erik Boot, Samantha D'Arcy, Lisa Palmer, and Dr. Anne Bassett received the Special Service Award on behalf of our whole team. The plaque is now on the wall of our Clinic!

## Junior Investigator Award

One of our residents, **Dr. Christina Blagojevic**, was one of the three international recipients of the 2024 Junior Investigator Award for the most outstanding presentation by a junior faculty member or trainee. Our former graduate student, Ania Fiksinski, and our research analyst, Tracy Heung, were previous recipients of this award in 2016 and 2018, respectively.

Who will be next from our team?



From left to right: Beata Nowakowska (Trustee of the 22q11.2 Society); Recipients of the Junior Investigator Award: Christina Blagojevic (2024), Daniella Miller (2024 & 2022), and Jente Verbesselt (2024); 2022 recipient Daniel McGinn

## Top Presenter Award

Four of our team members received the Top Presenter Award in their sessions at the meeting:

**Lisa Palmer**      **Dr. Nikolai Gil D Reyes**  
**Shengjie Ying**    **Dr. Christina Blagojevic**

We look forward to the next international 22q conference in 2026 in Greece!

# Dr. Nikolai Reyes receives NARSAD Grant

[Dr. Nikolai Gil D. Reyes](#) is a movement disorders neurologist who is pursuing graduate studies at our Clinic. Dr. Reyes is the recipient of a [2024 Young Investigator Grant](#) from the Brain and Behavior Research Foundation (BBRF) that will help support his research.

It is increasingly recognized that patients with 22q can experience several types of movement disorders. These conditions can involve having too few or too many movements. Together with Dr. Bassett's team at the Toronto General Hospital and Dr. Anthony Lang's team at the Toronto Western Hospital, Dr. Reyes is identifying the various ways adults with 22q are experiencing movement disorders. He presented his early findings at the 13<sup>th</sup> Biennial International 22q11.2 Conference in Óbidos, Portugal in July. Dr. Reyes will be publishing his research work in several papers that are now under way.

Adults with 22q are at an increased risk of developing early-onset Parkinson's disease, which is a treatable condition that is mainly characterized by motor symptoms. These symptoms include slowed movements, limb or body stiffness, tremor at rest, and trouble with balance and coordination, among others.

Dr. Reyes' work aims to investigate potential genetic factors that affect the risk of Parkinson's disease in individuals with 22q. This work promises to help those with 22q and other forms of movement disorders like Parkinson's disease in the general population.



Dr. Nikolai Gil D. Reyes is a graduate student of the University of Toronto Institute of Medical Science. He has also been a clinical fellow at the Toronto Western Hospital's Edmond J. Safra Program in Parkinson's Disease and the Morton and Gloria Shulman Movement Disorders Clinic.

# Dr. Sarah Malecki receives the Vanier Canada Graduate Scholarship



[Dr. Sarah Malecki](#) is the [recipient](#) of a [Vanier Canada Graduate Scholarship](#) in 2024. This prestigious scholarship from the [Canadian Institutes of Health Research \(CIHR\)](#), awarded to only 55 individuals from across Canada, provides 3 years of support for PhD studies. Here is the [official news release](#) from the Government of Canada. Sarah's project focuses on the Adult Impact of Complex Childhood-Onset Diseases – featuring 22q as a key model.

Sarah has been working with Dr. Anne Bassett for several years. Together, they have published world-leading research findings on several important aspects related to adults with 22q11.2 deletion syndrome. These include topics such as metabolic conditions (e.g., hypertriglyceridemia and type 2 diabetes), multimorbidity, aging and health care burden, and mortality. Most recently, Sarah led the publication of an important commentary for the Canadian Medical Association Journal (CMAJ) that proposes a healthcare delivery model for adults with complex needs (see page 8 of this newsletter).



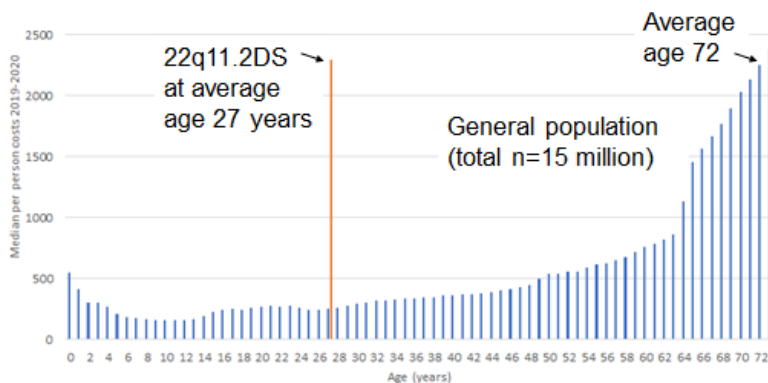
Dr. Sarah Malecki is a Resident in General Internal Medicine and PhD student at the University of Toronto

# Healthcare Burden for Adults with 22q

[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

In this report we show the breadth of inpatient and outpatient care needs, and the substantial medical complexity, faced by adults with 22q and their families and caregivers.

This is illustrated by the healthcare costs at the average age of 27 years for adults with 22q – these costs were about the same as the costs for individuals who were 72 years old in the general population, using health administrative data for the 15 million people in Ontario. 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 individuals with 22q.



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

## Proposing a Healthcare Model for Adults with Complex Genetic Conditions

[Closing gaps in care and knowledge for adults with complex genetic conditions](#)

Malecki S, Cohen E, Verma AAI, Bassett AS

CMAJ 2024 November 4;196:E1267-8. (doi: 10.1503/cmaj.240782) PMID: 39496350

In this brief commentary, we point out the great need to provide long-term all-round care to adults with complex genetic conditions (such as 22q).

We propose using models of pediatric care in adult medicine, combining multiple specialties and elements of existing programs such as the Dalglish Family 22q Clinic, and the well-resourced [Toronto Adult Cystic Fibrosis Centre](#) at St. Michael's Hospital. Multidisciplinary teams led by generalists but including experts from various medical and allied fields will better support the adult patients and decrease the burden on caregivers and primary care professionals.

We reinforced the importance of obtaining genetic diagnoses for adult patients. Not only do genetic diagnoses provide answers and emotional relief for the patients and their family, they allow for providing care at early stages of health issues. We suggest that increased genetic testing should be followed by improved gathering of population-level data on the genetic conditions and their adult outcomes, which would enable better understanding and management.

We point out that providing care for adults with complex genetic conditions requires substantially more support than is currently provided by our publicly-funded healthcare system.



# Clinical Recommendations for 22q Now Available in More Languages

In May 2024, Dr. Bassett received a \$15,000 “Planning and Dissemination Grant – Institute Community Support” from the Canadian Institutes of Health Research (CIHR) to translate the 2023 clinical practice recommendation papers for 22q into French, which is not only the other official language in Canada, but also a major language around the world.

We are delighted to let you know that the 22q clinical recommendations document for adults is now available in French, Spanish and Traditional Chinese. We are continuing to translate both the adult and pediatric guidelines papers into other languages as an ongoing project.

The original English documents and all of the translations are posted on our [Clinic website](#) and on the [22q11.2 Society website](#), as they become available. This provides free access to all!

## Clinical Recommendations Documents

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

- [English](#) (Original)
- [French – Français](#)
- [Spanish – Español](#)
- [Traditional Chinese 繁體中文](#)

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

- [English](#) (Original)
- French – in progress
- [Spanish – Español](#)
- Simplified Chinese – 简体中文 – in progress

Prenatal Screening and Diagnostic Considerations for 22q11.2 Microdeletions

- [English](#) (Original)

Help us spread the word!

## Barriers to Healthcare Access for Adults with 22q11.2 Deletion Syndrome

Ten years ago, we published a report entitled, [Perceived burden and neuropsychiatric morbidities in adults with 22q11.2 deletion syndrome](#). Before our Clinic existed, the patients and caregivers who completed the questionnaire used in this report noted the lack of medical and social and other services for adults with 22q, particularly at the time of transition from pediatric to adult care.

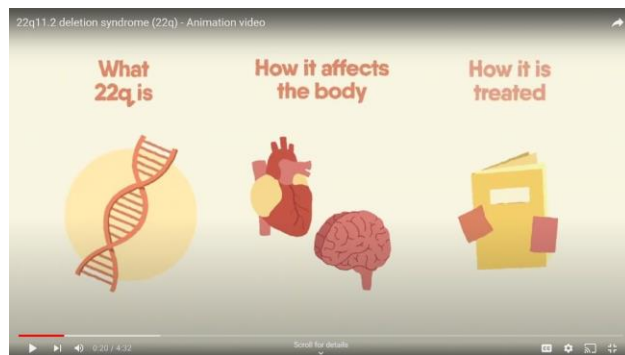
In 2023-24, we once again surveyed adults with 22q and their caregivers about their health and health care experiences. We are delighted to let you know that the results

indicate that there have been significant improvements in terms of care at transition care as well as access to medical and social services, as reported by our Clinic patients and caregivers. The survey results have confirmed our belief that a multidisciplinary clinic like ours is helpful to individuals and families affected by 22q.

Many thanks to all of the patients and caregivers who have taken part in our surveys. We have a manuscript reporting our research findings, that will be submitted for publication soon. Once published, we can share the details with everyone.

# An Educational Video about 22q

We 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 to better understand 22q. In the process, we asked our 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, our Social Worker, presented this at the International 22q11.2 Conference in July 2024. We have now added subtitles and uploaded the video to YouTube, so it's available to all!



## 22q Animation video

<https://www.youtube.com/watch?v=kIkX1xLIRko>

## What's Cookin' with Our Dietitian this year?

### Nutrition Webinars

Eating a healthy diet reduces the risk and severity of some of the medical conditions related to 22q.

To learn more about healthy eating for adults with 22q and their families, check out these recordings on YouTube from your 22q Dietitian, Samantha D'Arcy.

- [All about Blood Sugars and Cholesterol](#)
- [All About Calcium](#)
- [All About Appetite and Cravings](#)

### Join us in our 22q series "All About..."

Join us for some online workshops about 22q and nutrition. You can participate and ask questions, or keep your video and microphone off. Topics will include:

- All About Healthy Weight in 22q
- All About Iron and Vitamin B12
- All About Mindful Eating

We note that we have added **vitamin B12** levels to our annual bloodwork for 22q.

### Meal Planning Support Program

This group program is about meal planning; we meet virtually each week to learn and set meal planning goals. There are also follow-up meetings after the first month, to help you stay motivated.

### New Programs and Initiatives

- Mindful Eating Monthly: An online program where we practice mindful eating strategies and try different ones each month.
- How to Make a Healthy Snack: An online workshop where we make a simple, no-cook healthy snack. You'll receive an ingredients list ahead of time to buy the ingredients and participate right at home!

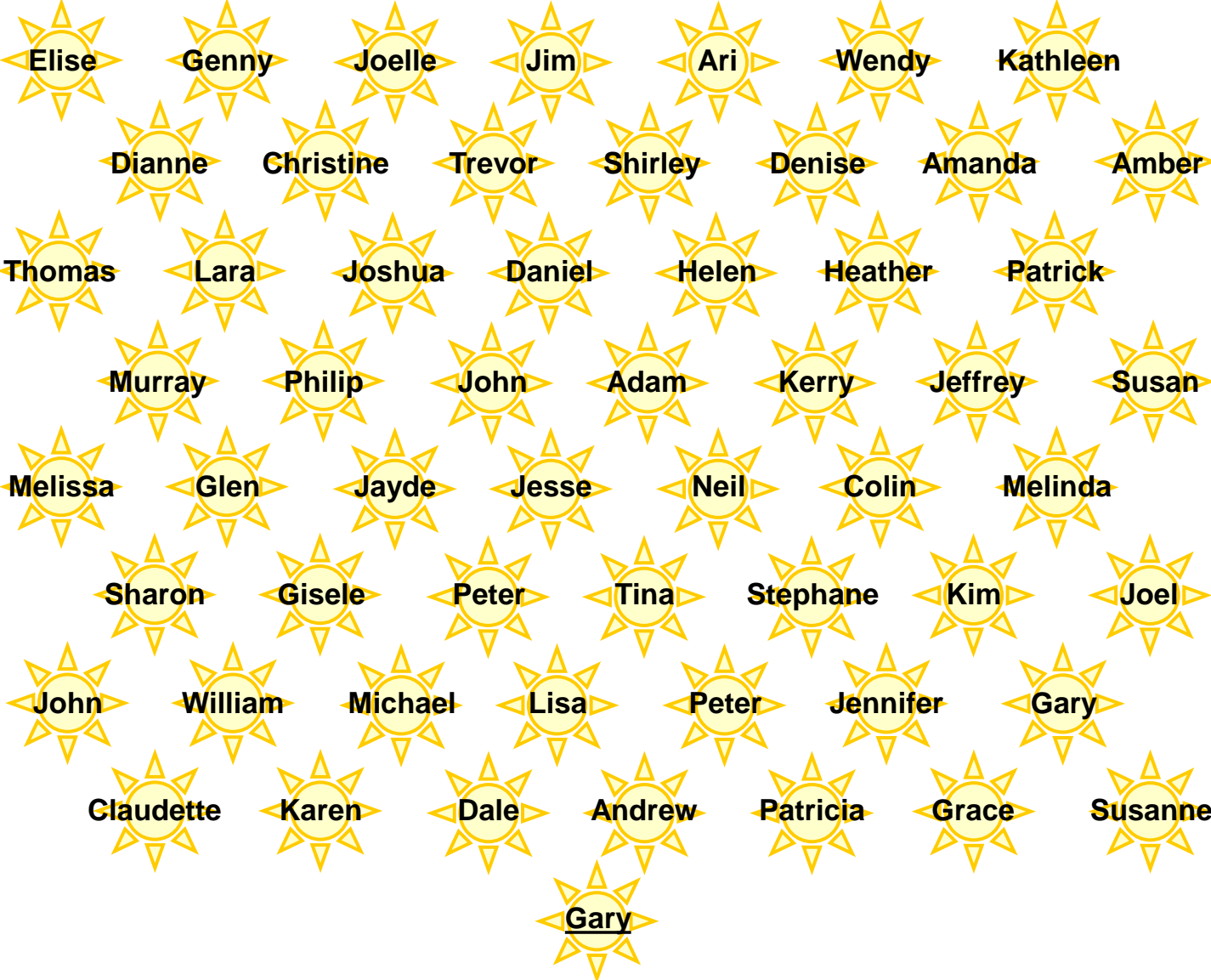
### 22q Nutrition Email Blast

Would you like to stay up-to-date on food and nutrition information? Then this weekly Email Blast is for you! Topics will include: short video recipes, food/product recommendations, nutrition news and more.

Stay tuned for more information in early January 2025.



# In Memoriam



These are the members of our 22q family we have lost over the past decades, including one individual in 2024.

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.

We are grateful to have known each and every one - and their loving families.

They are beacons of light that will forever shine!

## Join our research!

Over 95% of the individuals we follow at our Clinic are helping in our research about 22q. Without this help, we would know nothing about 22q11.2 deletion or related 22q conditions, how best to care for people, and continue to make progress.

We are one of the very few centres in the world to focus on **adults** with 22q – so every person who participates is precious. **We invite you, your parents, and your siblings to take part.**

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

## Contact us

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Please call or email Radhika at [22q@uhn.ca](mailto:22q@uhn.ca) for an appointment if we haven't seen you in a while!

Keep up with our news! Visit our website at [www.22q.ca](http://www.22q.ca)

## The future of our Clinic...

In June 2024, the support for Dr. Bassett's role as Dalglish Family Chair in 22q11.2 Deletion Syndrome ended and Dr. Corral retired, so you may be wondering about the future of our 22q Clinic. We are also wondering! The hospital and university process mandates that someone else will be recruited to a new Chair position supported by a generous donation from the Weston Family Foundation. No one connected with our Clinic will be involved in the search process for the new Chair. Dr. Bassett continues to provide clinical care, and we will hope to be able to keep you posted as we learn more about these changes.

## Why give? We can't do it alone...

The [UHN Foundation](#) manages all donations and you can direct your gift to our Clinic. Just a reminder that we receive no Ministry of Health or institutional funding support **for most of our educational, outreach and other activities (e.g., our website) for patients with 22q and their families.** Many families (and friends) have kindly made donations to help us. Please write **22q Deletion Syndrome Fund for Excellence** in the gift designation field online or the memo field of your cheque. **Thank you** for your support - every gift helps us help those with 22q!

Donate to our Clinic  
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using this [form](#)

And there is  
also the UHN  
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program



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