22q & Friends

2019 Newsletter

Inside you will also find...

And more...

Hello and happy reading from all of us!

Hello again from The Dalglish Family 22q Clinic and the Clinical Genetics Research Program (CGRP)! We hope you, your family and friends had a wonderful 2019. As we progress with our Clinic and our research specializing in adults with 22q11.2 deletion syndrome (22q) and related conditions, we like to keep you up-to-date. As always, our goal is to continue to personally contact and hopefully see each of you in 2020! This newsletter is intended to share news and updates as well as help to answer some important questions about 22q.

Our program continues to see old friends and new patients and families for our clinical research studies that help us understand more about 22q. We have seen about 400 adults so far with 22q from across Canada and we have also helped many others from around the world. We see and follow more adults with 22q than any other single centre worldwide. Our collaborative research program works in conjunction with the University Health Network (UHN) (Toronto General Hospital, Toronto Western Hospital, Princess Margaret Hospital, Toronto Rehab), the Centre for Addiction and Mental Health (CAMH) and the Hospital for Sick Children, as well as hospitals and clinics around the world. Our goal is to continue to learn more about 22q and similar genetic syndromes. Most importantly, we want to learn how to best manage the associated illnesses and conditions to improve the long-term outcomes for people with 22q and their family members.

Happy reading!

Studying movement disorders in 22q – how we can better serve our patients

We recently had the chance to interview Dr. Erik Boot, who has been working on a new study about movement disorders and 22q. He told us about some of his exciting findings and what this means for our 22q community.

What was your role in the study?

In fact, the study is still going on, although we are not collecting any new data at this time. From the start, I have led the study, and to a large extent also conducted it. In the period that I did my fellowship at the Dalglish Family 22q Clinic (from 2014-2016), I investigated movement issues in adults with 22q. In the past months, I have analyzed the results and written up part of the results with help from colleagues. I just submitted an article about the study findings for publication.

Dr. Anne Bassett,

Director, The Dalglish Family 22q Clinic & CGRP

Did you know...

- Most people with 22q do not inherit this condition from a parent. The genetic change usually happens as a new event in a family. We do not yet understand what may make this genetic change more likely to happen in any particular family.
- Every person (man or woman) who has 22q has a 50% ("50-50") chance of passing this deletion on at every pregnancy.
- The word "syndrome" means a collection of clinical features each of which can be recognizable but that can vary from person to person.

Studying movement disorders in 22q - how we can better serve our patients continued

Tell me a bit about your background?

I am a Dutch physician specialized in intellectual disability medicine. This is a distinct medical specialty that is only found in the Netherlands, addressing both physical and mental health problems of people with intellectual disabilities. I've had a special interest in people with 22q for over 15 years now. I remember well the moment that I met with a woman with 22q for the first time, when I did my internship in psychiatry. She was a nice young woman who suffered from recurrent psychoses. I started to read about 22q and wanted to learn more about it. That interest has only grown.

Can you tell me in a few sentences, what is this exciting new study about?

The study focuses on movement abnormalities that we see sometimes in adults with 22q, such as slowness of movements, stiffness, and shakiness. To get a better understanding of these issues, I have used different approaches including chart reviews, by doing standardized physical (motor) examinations, and by using electronic assessments. To get an idea to what extent movement issues are related to the genetic condition, I also studied adults without 22q.

Why study movement disorders, and what does this mean for our patients with 22q?

In recent years, we have learned that a minority of people with 22q develop Parkinson's disease during their lives. Parkinson's disease is a chronic disease of the brain that results in a gradual loss of brain cells, causing problems with movement and mental functioning. Also, we've had the idea that symptoms of Parkinson's are more common in people with 22q than in the general population. Indeed, our study and others have now shown that these symptoms are relatively common in 22q. This means that we will need to educate our patients and their families, and health care providers about this.

What do you hope this research can lead to in the future?

I see this study as a first step towards more knowledge and understanding of the motor issues that we may see in 22q when people age. We will also want to know who is at risk of Parkinson's disease or its symptoms, when and how symptoms start, and how they evolve. Ultimately, we will want to know how we can best treat, or even prevent the symptoms. For this, we will need to follow-up with patients and repeat assessments.

What other projects do you plan on studying next?

My new projects will focus on late medical outcomes in 22q, including movement disorders. I am interested to learn more about health and health problems in people with 22q at middle and old age. This is an important area that has been relatively neglected in research thus far. We really want, and need, to learn more about aging in 22q.

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We'd like to thank Dr. Boot and the team of researchers for doing this great work and making an impact in the lives of people living with movement disorders.
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Clinic status change and fundraising

The Dalglish Family 22q Clinic was established in late 2012, thanks in large part to generous donations from the Dalglish Family and The W. Garfield Weston Foundation. We are truly grateful to them and to the private donors who have helped to financially support the operations of our Clinic over the years.

Our team not only provides specialized, patient-centred care to adults and their families, but we also educate patients, families, trainees and health care providers. In addition, we perform world class research to improve the quality of life for individuals and families affected by the syndrome. As we are one of the only clinics in the world to specialize in adults with 22q, patients and families from different countries ask for our help, and medical professionals worldwide seek guidance about caring for patients and on setting up their own 22q clinics. In short, we strive to continually make significant contributions to the well-being of the 22q community worldwide.

Recent changes to our Clinic mean that we are no longer a privately-funded clinic. The Government of Ontario through the Ministry of Health provides important basic funding for our Clinic operations. This, however, does not support many aspects of our ongoing work. Our efforts to improve public education about 22q, our website, and much of our clinical research rely on private donations and other support. As many of you already know, 22q is not a well-known condition—not even among medical professionals. If we, as the leader in the 22q medical community, want to continue to advocate for our patients and to raise public awareness for the syndrome, we need financial help. Every amount, no matter how big or how small, will make a difference. For example, $26.38 a month could help us host our 22q.ca website, a comprehensive resource that both Canadians and people worldwide can access. A clinical research fellow (like Dr. Boot was!) is about $90,000 per year. A research fellow is about $55,000 per year. A summer research medical student is $4,000.

How can you and others you know help?

Donations to the Dalglish Family 22q Clinic support patient care, education, and research benefiting adults with 22q and their families. Please help us continue this important work. If you or someone you know (relatives, friends, coworkers) would like to make a donation, please visit our website at www.22q.ca.

Your support is greatly appreciated!

Back row (left to right): Dr. Maria Corral, Dr. Anne Bassett, Lisa Palmer, Radhika Sivanandan, Joanne Loo, Joanne Ha.
Front row (left to right): Tracy Heung, Samantha D'Arcy

Did you know...

22q actually had a lot of different names before:

- DiGeorge syndrome
- Velocardiofacial syndrome (VCFS)
- Conotruncal anomaly face syndrome
- Opitz G/BBB syndrome
- Cayler cardiofacial syndrome

These names were given by different doctors who worked in specialized areas before we knew they were all tied together by the piece missing on chromosome 22.

For more information, please visit our website: **www.22q.ca** and click on "Same Name Campaign".

Donations

The most important donation is your generous participation in our research studies. Without your support, we could not hope to make advances in treatment or achieve medical breakthroughs for patients with 22q.

Donations from individuals or groups may be made to the Dalglish Family 22q Clinic. The donations go directly towards helping support our research initiatives and Clinic.

Your gifts are greatly appreciated and ensure we can keep going with our studies!

To donate online, visit our website: **www.22q.ca** and click “Donate”

All donors receive a taxable receipt.

An inside look into The Dalglish 22q Clinic : staff members

Dr. Anne Bassett: Director

Dr. Anne Bassett is the Director of the Dalglish Family 22q Clinic and the Dalglish Chair in 22q Deletion Syndrome. She is a world renowned expert in the genetics of complex conditions - especially 22q. Dr. Bassett is also the Director of the Clinical Genetics Research Program at CAMH and a Professor of Psychiatry at the University of Toronto. She is a leader in international studies of 22q, and is the Medical Director for the International 22q Foundation. Dr. Bassett has been providing exemplary care to adults with 22q, their families and caregivers for over 25 years.

Dr. Maria Corral: Psychiatrist

Dr. Maria Corral completed her medical studies at the University of British Columbia. She worked for 30 years as a psychiatrist, educator and administrator until her retirement as Head of the Department of Psychiatry and Director of Medical Education at St. Paul’s Hospital, Providence Health Care, in June of 2014. She was also Vice-Chair of the Psychiatry Examination Board for the Royal College of Physicians and Surgeons of Canada from 2011-2014.

Now living in Toronto, Dr. Corral is a key member of the Dalglish Family 22q Clinic and its multidisciplinary team of clinicians.

Lisa Palmer: Social Worker

Lisa graduated with a Bachelor of Arts degree in Psychology in 2010 and a Bachelor of Education degree in 2012. She completed a Master of Social Work degree at the University of Toronto in 2015.

Working with the Clinic team, Lisa assists patients and their families in examining challenges and concerns and in helping to find the best available community resources. She also has her own clinical research studies to help us learn more about 22q and improve the services we provide.

Samantha D’Arcy: Registered Dietitian

As a Registered Dietitian at the Dalglish Family 22q Clinic, Samantha works collaboratively with patients and their families to create nutrition plans that fit their needs and lifestyle. In addition to one-to-one patient care, Samantha is involved with group facilitation and patient and caregiver education as requested. She has also led several 22q educational initiatives.

An inside look into The Dalglish 22q Clinic : staff members

Radhika Sivanandan: Patient Flow Coordinator

Radhika is likely the first person you will encounter when you come to the Clinic. She co-ordinates all appointments and ensures that all necessary documents are available. Please feel free to contact her at the Clinic's main number (416) 340-5145 if you have any questions or concerns regarding your appointments.

Ania Fiksinski: Graduate student

Ania is a PhD graduate student from the Netherlands who obtained her Master's degree in Clinical Psychology from the University of Utrecht in 2013. Since then she has been working as a psychologist in the Department

of Psychiatry at the University Medical Center in Utrecht, specializing in psychological assessment and treatment of children and adolescents with 22q. She has also been actively involved in academic research regarding the cognitive and psychiatric features of 22q. Ania is currently pursuing a PhD degree with Dr. Bassett in a collaboration between the University of Toronto and its international partner university, the University of Utrecht.

Clinical Research and Other Staff

Gladys Wong: Coordinator / research analyst

Joanne Ha, Tracy Heung: Clinical research analysts

Joanne Loo: Patient and family educational tool developer and website manager

Clinic by the numbers

Here is a quick look at how our Clinic is doing, by the numbers

Number of 22q educational resources developed by Clinic staff: **19**

Number of patients seen at the Clinic since opening: **353**

Number of family members supporting our patients and research: **522**

Number of patients who have personalized medical information cards made: **67**

Number of research papers published in 2019: **16**

Developmental History: A complete picture of 22q

As part of our routine clinical assessments, the clinicians and staff at the Dalglish Family 22q Clinic always ask patients with 22q and their families about family history, and about early childhood development and health conditions, also known as a developmental history.

To make sure we have the most complete and accurate information, we may call to clarify parts of this important history.

You may receive one of these calls from our Clinic. We look forward to talking to you to make sure we provide the best care possible!

The Clinical Genetics Research Program (CGRP) at the Centre for Addiction and Mental Health (CAMH)

Many of our Dalglish Family 22q Clinic team members also have a role at CGRP at CAMH. Together we are all working on research that will help us understand 22q and related conditions, like heart defects, schizophrenia, learning disabilities, multiple medical illnesses and autoimmune disorders.

Toronto, Ontario

Dr. Anne Bassett
Principal investigator & Director

Dr. Eva Chow
Research psychiatrist

Gladys Wong
Coordinator

Dr. Greg Costain
Senior Medical Genetics resident (soon to be a medical geneticist)

Tracy Heung
Research analyst

Joanne Ha
Research analyst

Dr. Spencer Van Mil
Resident in Pediatrics

Dr. Lily Van
Resident in Psychiatry

Dr. Sarah (Voll) Malecki
Resident in Internal Medicine

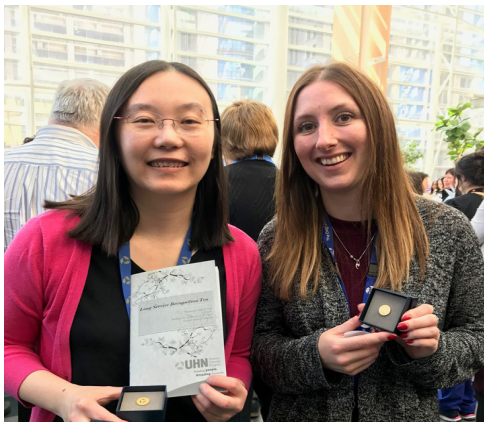
Christina Blagojevic
Medical student

Brigid Conroy
Medical student

Clinic awards 2019

Please join us in congratulating the following Clinic members and affiliates on their achievements:

Long Service Recognition Awards 2019
Lisa Palmer, RSW and Joanne Loo, PhD received this award in 2019 in recognition of reaching their service milestones of 5-years with The Dalglish Family 22q Clinic. We are thankful for the hard work and commitment that Lisa and Joanne bring to our 22q community!



Left to right: Joanne Loo, Lisa Palmer

Adjunct Lecturer Appointment February 2019
Our Clinic social worker, Lisa Palmer, RSW, was appointed as an Adjunct Lecturer in the Factor-Inwetash Faculty of Social Work at the University of Toronto. This appointment is in recognition of Lisa's support and training of social work students and her commitment to continuing education. Lisa has an important role teaching health care professionals and others about 22q.

UMC Utrecht Internationalization Committee Grant for Strategic Network Development award 2019
Ania Fiksinski received this €4,500 award to support her PhD research in intellectual functioning in individuals with 22q and visits to Toronto.

22q11.2DS International Consortium & more

The Dalglish Family 22q Clinic is part of an international group of scientists and clinicians working to better understand 22q. We are working with many other international leaders in the field, including those who are listed on the next page.

In 2015 we published the first international clinical guidelines for adult patients with 22q.

Through our major research efforts, we are interested in how 22q can affect all aspects of development and all important health systems, including the brain and the heart, lifelong.

Collaborating doctors & researchers

Along with the CGRP Team, there are many collaborating doctors and researchers who work with us locally and internationally on 22q and related research. They include:

Ontario

Dr. Candice Silversides
Cardiologist,
Toronto General Hospital

Dr. Erwin Oechslin
Cardiologist,
Toronto General Hospital

Dr. Danielle Andrade
Neurologist,
Toronto Western Hospital

Dr. Connie Marras
Neurologist,
Toronto Western Hospital

Dr. Anthony Lang
Neurologist,
Toronto Western Hospital

Dr. Susan George
Endocrinologist,
Toronto General Hospital

Drs. Hanna Faghfoury & Chantal Morel
Medical Geneticists,
Toronto General Hospital

Dr. Jacob Vorstman
Child Psychiatrist,
Hospital for Sick Children

Christian Marshall, PhD
Stephen Scherer, PhD
Geneticists,
Hospital for Sick Children

Atlantic Canada

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

Dr. Sarah Dyack
Medical Geneticist,
Halifax, NS

USA

Donna McDonald-McGinn
Director, 22q Clinic,
Children's Hospital of Philadelphia (CHOP)
Philadelphia, PA

Dr. Linda Brzustowicz
Geneticist,
New Brunswick, NJ

Bernice Morrow, PhD
Geneticist,
New York, NY

Asia

Dr. Brian Chung
Medical Geneticist,
Hong Kong SAR, China

Europe and UK

Ann Swillen, PhD
Child psychologist,
Leuven, Belgium

Dr. Therese van Amelsvoort
Psychiatrist,
Maastricht, Netherlands

Peter Scambler, PhD
Geneticist,
London, UK

South America

Dr. Gabriela Repetto
Medical Geneticist,
Santiago, Chile

Australia

Linda Campbell, PhD
Psychologist,
Newcastle, Australia

Research funding

We continue to submit several research grant applications every year. If and when these are funded, these grants support our research studies of 22q and of related conditions. Canadian funding tends to be modest so many grants are needed to support our advanced genetic studies and our research team. We have received funding from:

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)
Five-year sub-contract, 2019-2024

We are grateful to these agencies for their research support!

Clinic events

22q at the Zoo

On May 26th, 2019, our Dalglish Family 22q Clinic at Toronto General Hospital and the 22q Deletion Syndrome Clinic at the Hospital for Sick Children co-hosted the 8th annual 22q at the Zoo event, an international event with participants across the globe. The focus of the day was for individuals with 22q, their families and Clinic staff to socialize, network and raise awareness of 22q in the community. Participants also had the chance to join in fun activities with their friends and families and enjoy a day at the Toronto Zoo.

Peer Support Groups

The Dalglish Family 22q Clinic holds peer support groups periodically for individuals with 22q and their parents or caregivers. We facilitate two groups, each of which run for about 60 minutes:

A group for parents or caregivers – an opportunity to talk to other parents about concerns, challenges, and successes.

A group for our adult patients – a chance for patients with 22q to meet each other, participate in some fun exercises and to discuss topical subjects with staff facilitation.

Our most recent peer support group was held on December 10th, 2019. Please stay tuned for information about the next session!

22q Family Conference

On November 2nd, 2019, the 22q11.2 Parent Conference was held at the Hospital for Sick Children in Toronto. This conference gave parents and caregivers of individuals with 22q the chance to learn more about 22q and to meet other families sharing their same experiences.

Dr. Bassett's graduate student, Ania Fiksinski, was one of the speakers at this conference, and spoke about how we are now gaining a better understanding of intellectual functioning in individuals with 22q.

Transition Program

Becoming an adult is a process for everyone, including individuals with 22q11.2 Deletion Syndrome (22q). "How will 22q symptoms affect my life as I become an adult?" This is a common question for many of our patients and their families.

The Dalglish Family 22q Clinic, in collaboration with the SickKids 22q Deletion Syndrome Clinic, offers a structured 22q Transition Clinic several times per year. This full day program gives individuals with 22q information about adult care, along with lunch and snacks, and literally walks families over in order to make a smooth and pleasant transition from the 22q Clinic at SickKids to our 22q Clinic at the Toronto General Hospital.

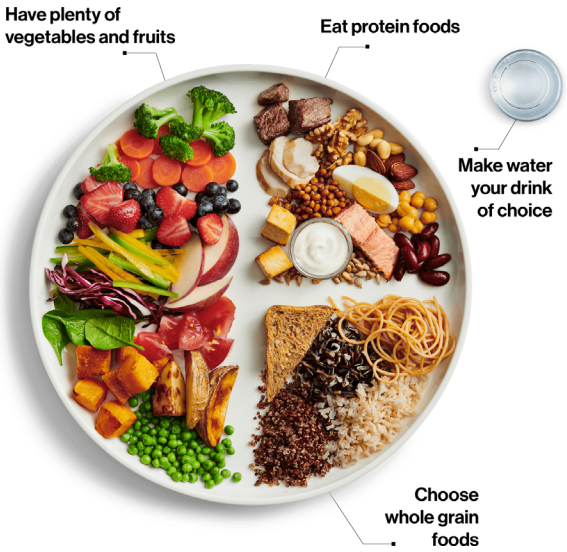
We look forward to holding our next 22q Transition Clinics, which will be Tuesday, April 14th, 2020 and Tuesday, May 12th, 2020.

Did you know...

- 22q may be more common than we once thought. An estimated 1 in every 2000-3000 babies are born with the deletion on chromosome 22.
- Treatable psychiatric illnesses are one of the most common features of 22q in adults. About 6 in every 10 adults with 22q have a treatable psychiatric illness.
- The characteristics of 22q occur along a spectrum. Some individuals have more distinct features that are more easily recognized. Others may not have the classic features. Genetic testing is the only way to confirm a 22q deletion.

The New Canada's Food Guide

Early in 2019, Health Canada published a new Canada's Food Guide. This helps teach Canadians how to eat healthy.



The main points in Canada's Food Guide are:

1. At every meal, eat half a plate of fruits and vegetables.

2. Eat a quarter plate of whole grains, like whole wheat bread, brown rice, or whole wheat pasta.

3. Eat a quarter plate of protein foods like fish and meat, and eat more plant proteins such as beans, tofu, nuts and seeds.

4. Drink water more, and drink fewer sugary drinks such as pop, energy drinks and juice.

How to sleep better

Having a better sleep will help you feel less sleepy, less stressed out, and less depressed. Here are some tips to help you sleep better.

1. Wake up and go to sleep at the same time every day.

2. Keep electronics out of the bedroom

3. Regular exercise is great for good sleep. You should aim for at least 30 minutes of exercise 5 days a week.

4. Eat a balanced diet (see the Canada's Food Guide above).

5. Don't take naps, especially after 4 pm.

6. Don't drink caffeine or alcohol, especially 4 to 6 hours before bed.

For more questions about healthy eating or sleeping, contact us at the Clinic (416-340-5145 or 22q@uhn.ca) to speak to one of the members of our team.

The importance of keeping up-to-date with vaccines

Vaccines help protect our bodies against infections, by "teaching" your body's immune system how to fight viruses and bacteria. It is important to ask your doctor questions you might have about vaccines, and be cautious of information you get from the Internet.

A yearly flu shot is a vaccine to help protect you against the flu that comes around every winter. Some other things you can do to help protect yourself includes:

- Ensure that people living with you are also vaccinated
- Clean hands frequently with alcohol-based hand rubs or soap and water
- Stay home if you are ill with new onset cough and fever
- Stay at least 2 meters (6 feet) away from people who are ill with the flu
- Avoid touching your face and eyes
- Frequently clean commonly touched surfaces
- Cough and sneeze into a tissue, dispose used tissues immediately and clean your hands. If no tissue is available, cough into your sleeve, not your hands.

If you have any questions about vaccines or flu shots, contact the Clinic (416-340-5145 or 22q@uhn.ca) to make an appointment with one of our Clinic doctors.

New and upcoming publications

We are excited to announce important publications on 22q since our last newsletter. These would not be possible without your generous contributions to our research. You are helping people with 22q worldwide!

Some of our recent and upcoming publications include:

A genetic model for multimorbidity in young adults.

Malecki SL, Van Mil S, Graffi J, Breetvelt E, Corral M, Boot E, Chow EWC, Sanches M, Verma AA, Bassett AS. Genet Med. 22(1):132-141, 2020. (doi: 10.1038/s41436-019-0603-1, PMID:31363180)

To study the burden of illness in 22q, we compared young to middle-aged adults with 22q to a large community-based Canadian general population sample of over 25,000 people. We defined burden of illness ("multi-morbidity") as using five or more prescription medications. In the 25-44 year age group the overall burden of illness was most similar to the burden in the general population at age 65. In the 45-64 year age group the burden of illness in 22q was about twice that of the general population. For younger adults, the pattern tended to be consistent with the conditions commonly associated with 22q, but in middle age in 22q the pattern looked more similar to older age groups of the general population. Our results highlight the importance of providing multidisciplinary and person-centred care for adults with 22q.

All-cause mortality and survival in adults with 22q11.2 deletion syndrome.

Van L, Heung T, Graffi J, Ng E, Malecki S, Van Mil S, Boot E, Corral M, Chow EWC, Hodgkinson KA, Silversides C, Bassett AS. Genet Med. 21(10):2328-2335, 2019. (doi: 10.1038/s41436-019-0509-y, PMID:30948858)

As information is limited on long term outcomes in 22q, we studied mortality and survival in 309 adults with 22q and their 1014 unaffected parents and siblings. The results showed that the probability of survival to age 45 years was approximately 95% for those with no major congenital heart defect, and 72% for those with a major heart defect. Although the 22q11.2 deletion and more severe forms of congenital heart defects contribute to a significantly lower life expectancy than family-based expectations, a substantial minority of individuals with 22q had outlived both parents. The average age at death was approximately 5 years older than the age we reported 10 years ago for the initial subgroup of 100 patients with 22q.

Neurocognition and adaptive functioning in a genetic high risk model of schizophrenia.

Fiksinski AM, Breetvelt EJ, Lee YJ, Boot E, Butcher N, Palmer L, Chow EWC, Kahn RS, Vorstman JAS, Bassett AS. Psychol Med. 49(6):1047-1054, 2019. (doi: 10.1017/S0033291718001824. PMID:30064532)

The results of this study showed the average relative cognitive strengths and weaknesses in 22q (e.g., relatively better on tasks related to visual than verbal memory, and better yet when given hints). The best overall performance for adults with 22q was in Daily Living Skills. Older age was significantly associated with better functional outcomes. Executive Performance (tasks requiring more abstract thinking and judgment) was significantly associated with functional outcome. The fact that there was substantial variability between individuals emphasized the need to recognize and balance individual capabilities and environmental demands in day-to-day situations.

We want your input!

We want to create newsletters that interest you. What information would you like to see in our next newsletter? Perhaps you have questions for us about 22q or another genetic syndrome. Maybe you have a comment or story to tell us. We want to hear from you so that we can make each and every newsletter more helpful and enjoyable to read.

We invite you to email us at 22q@uhn.ca or fax your comments to us at (416) 340-5145!

New and upcoming publications

Did you know...

- Most people with 22q are missing a piece of genetic material (DNA) that is about 3 million DNA building blocks long, on one copy of chromosome 22. This region contains about 46 genes that code for proteins. We do not yet know what all of these genes do.

- Individuals with 22q may have thyroid problems. The thyroid may be under or over active. Regular thyroid testing can help to identify a thyroid problem so that effective treatment can be started.

22q11.2 deletion syndrome-associated Parkinson's disease.

Boot E, Bassett AS, Marras C. Mov Disord Clin Pract. 6(1):11-16, 2018. (doi: 10.1002/mdc3.12687, PMID: 30746410)

This paper reviewed what is known so far about the hallmark motor symptoms and neuropathology of Parkinson's disease. Typical findings are present in individuals with 22q who develop Parkinson's disease, often at a young age (average 40 years). 22q11.2DS-associated Parkinson's disease accounts for about half of 1% of all individuals with early-onset Parkinson's disease. Studying Parkinson's disease in people with 22q could help us understand the mechanisms that cause this condition in the general population.

Low prevalence of substance use in people with 22q11.2 deletion syndrome.

Vingerhoets C, van Oudenaren MJF, Bloemen OJN, Boot E, van Duin EDA, Evers LJM, Fiksinski AM, Breetvelt EJ, Palmer LD, Vergaelen E, Vogels A, Meijer C, Booij J; Genetic Risk and Outcome of Psychosis (GROUP) investigators, de Haan L, Swillen A, Vorstman JAS, Bassett AS, van Amelsvoort TAMJ. Br J Psychiatry. 3:1-7, 2019. (doi: 10.1192/bjp.2018.25, PMID: 30604657)

The results of this study suggested that patients with 22q are at decreased risk for substance use and substance use disorders compared to individuals in the general population. Drinking, smoking, and drug use however are major health problems for some individuals with 22q, requiring active treatment and prevention measures.

Neurobiological perspective of 22q11.2 deletion syndrome.

Zinkstok JR, Boot E, Bassett AS, Hiroi N, Butcher NJ, Vingerhoets C, Vorstman JAS, van Amelsvoort TAMJ. Lancet Psychiatry. 6(11):951-960, 2019. (doi: 10.1016/S2215-0366(19)30076-8, PMID:31395526).

This review paper summarizes what we know about disorders common in 22q that involve the brain. These include intellectual disability, schizophrenia, attention-deficit disorder, anxiety disorders, seizures, and Parkinson's disease. Learning more about them in people with 22q may help scientists understand better these same conditions in the general population.

Haploinsufficiency of vascular endothelial growth factor related signaling genes is associated with tetralogy of Fallot.

Reuter MS, Jobling R, Chaturvedi RR, Manshaei R, Costain G, Heung T, Curtis M, Hosseini SM, Liston E, Lowther C, Oechslin E, Sticht H, Thiruvahindrapuram B, Van Mil S, Wald RM, Walker S, Marshall CR, Silversides CK, Scherer SW, Kim RH, Bassett AS. Genet Med. 21(4):1001-1007, 2019. (doi: 10.1038/s41436-018-0260-9, PMID:30232381)

Studying adults who were born with major heart defects ("blue babies"), but who did not have 22q, we discovered a new pathway to these important conditions. We used the most advanced genetic sequencing methods available and carefully examined for changes to genes that were likely to cause problems with the development of the heart. The findings focussed on a signalling mechanism that may be important for many individuals, including those with 22q.

Presentations

Each year we participate in several presentations on 22q and other related conditions. The following are some highlights from 2019:

European Human Genetics Conference
Gothenburg, Sweden
June 15-18, 2019

In this meeting for health professionals who work in human genetics, Dr. Anne Bassett presented on the burden of illness in patients with 22q, how this compares to the general public and how the patterns identified may in the future be used to identify genetic disorders within large populations.

22q11DS European Conference
Barcelona, Spain
November 16, 2019

At this annual congress about 22q for families and health professionals, Dr. Bassett presented a keynote presentation about optimizing outcomes for adults with 22q, met with families and participated in workshops.

Upcoming Presentations:

12th Biennial International 22q11.2 Conference
Split, Croatia
June 23-26, 2020

At this biennial conference for health professionals and family members interested in 22q, Dr. Anne Bassett and other members of our Clinic team and students will be presenting on the newest research and clinical practice about 22q.

Online Resources

Here are some websites that we hope you will find helpful about 22q and related issues:

www.22q.ca
The Dalglish Family 22q Clinic:
Provides up-to-date information about our Clinic, including specialized services to help patients and families.

www.22q.org
The International 22q11.2 Foundation Inc.:
Focuses on bringing awareness and support to individuals affected with 22q and their families.

www.schizophrenia.ca
Schizophrenia Society of Canada:
Information for patients and families affected by schizophrenia, including treatment, daily living and advocacy.

www.cachnet.org
Canadian Congenital Heart Network:
The learning centre contains information and resources for adults who were born with a heart defect.

Further Reading

Ever wondered how you can make the most out of your medical appointments? Check out "Preparing to see a doctor or other medical professionals". The information is available on our website. You can also download a PDF of this tip sheet.

Our Clinic has published a number of pamphlets that address common concerns for people with 22q. Some titles include:
"Alcohol"
"Mental Health"
"Sexual Health"
"Internet Safety"
"Healthy Eating"
"Emergency Preparedness"

To read these and other pamphlets, please visit our website: http://22q.ca/aboutus/our-pamphlets/

In Memoriam

The Dalglish Family 22q Clinic

Sadly, over the decades and including very recently (in December 2019), we have lost members of our 22q family from the age of 18 to 68 years. They will never be forgotten and remain close to our hearts. Their research contributions live on, and through the knowledge generated, they continue to help others with 22q. They are included in all of our studies and their generosity will further advance the care of individuals with 22q throughout the decades to come.

They are true angels!

Giving back to the 22q community

Over the years, with the help of our patients, families and researchers around the world, we have come far in learning about how to optimize care for individuals with 22q. Hundreds of patients and families have already participated by donating their time and a small blood sample. We also have very precious tissue donations from some of our patients who have passed away, that are helping us to understand more about all stages of the life cycle of 22q. All of our advances rely on the continued contributions of the amazing patients, families and caregivers who we see at our Clinic.

When our patients and families ask, “How can I help support other people with 22q?”, the answer is to get involved with research. We encourage patients and families to ask us what new research we are working on. We are happy to explain any of our ongoing and new studies and collaborations.

Every person with 22q can help! A special group are those who are doing especially well. These individuals can help us understand how well people with 22q can do, and can shed light on the secrets of good health in 22q.

We wholeheartedly appreciate everyone who has already given to the 22q community by participating in research. This is a gift that is improving healthcare for people with 22q – every day in our Clinic and around the world, and will do for many years to come.

If you or your friends wish to make a donation in memory of someone special, please visit our website at **www.22q.ca** and click “Donate” or there is always regular mail to:
The Dalglish Family 22q Clinic, Toronto General Hospital,
200 Elizabeth Street, Toronto, Ontario, M5G 2C4.

Contact us!

We are grateful to everyone who has donated their time for our 22q studies, and we hope that you will join us in the newest parts of our studies. All of the progress we are making is because of you!

As our studies progress, we need to maintain an up-to-date and accurate picture of your health status. We may contact you for updates on your family history and health status in the near future. Ideally, we like to see everyone with 22q once per year. This will also help you stay up-to-date on information about 22q.

The Dalglish Family 22q Clinic
Toronto General Hospital

8 NU (Norman Urquhart), Room 802
(8th Floor)
200 Elizabeth Street
Toronto, Ontario M5G 2C4
Phone: (416) 340-5145
Fax: (416) 340-5004
E-mail: radhika.sivanandan@uhn.ca

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

Clinical Genetics Research Program (CGRP)
Centre for Addiction and Mental Health

33 Russell Street, Main Building, 1st Floor
Toronto, ON M5S 2S1
Phone: (416) 535-8501 x 32734
Fax: (416) 535-7199
E-mail: gladys.wong@camh.ca

Please call Gladys for an appointment if we haven’t seen you in a while!