22q & Friends

2018 Newsletter

Inside you will also find...

- Family Background Study
- Recent publications
- Inside the Clinic
- International 22q Conference
- Our website
- Healthy Drinks
- CGRP team
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- Clinic events
- Online resources

And more...

Hello and happy reading from all of us!

Hello again from the The Dalglish Family 22q Clinic and the Clinical Genetics Research Program (CGRP)! We hope you, your family and friends had a wonderful 2018. As we progress with our Clinic and 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 2019! 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 newcomers for our clinical research studies that help us understand more about 22q. We have seen about 360 adults so far with 22q from across Canada and have helped others from around the world. We have seen 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 ambition is to continue to learn more about 22q and similar genetic syndromes. Most importantly, we want to find out how to best manage the associated illnesses and conditions to help improve the long-term outcomes for people with 22q.

Happy reading!

Productive and fun: International 22q11.2 Deletion Syndrome Conference in Whistler

The highlight of 2018 has to be our trip to Whistler, British Columbia, for the 11th Biennial International 22q11.2 Conference!

Dr. Anne Bassett, our Clinic Director, is a Trustee and a Founding Board Member of the International 22q11.2 Society, which consists of researchers who learn about 22q and doctors who take care of patients with 22q. The Society organizes an international conference for professionals every two years and often a family meeting. This year, these meetings were held in picturesque Whistler from July 11th to 15th, 2018. We learned from other international participants from 16 countries through informative presentations, in-depth discussions, and valuable networking opportunities. We also presented our clinical research results. In fact, our Toronto Dalglish Family 22q team gave a full 11% of the presentations at the professional meeting and 10% of talks at the family meeting on topics from molecular genetics to IQ scores and cognitive strengths to medical information cards for patients. We congratulate Tracy Heung, Research Analyst affiliated with our Clinic, who received the Junior Investigator Award.

While the presentations and discussions

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.

Productive and fun: International 22q11.2 Deletion Syndrome Conference in Whistler continued

satisfied our quest for knowledge at the main conference, the many surrounding meetings (there were 6!) gave us a chance to meet with colleagues and present to families all about 22q.

Being in Whistler was more than just about attending the conference, though. We were also treated to a performance of First Nation Dance, where the dancers explained to us the symbolism of each move. Some of us took the opportunity to plan a holiday around the meeting and enjoy the great outdoors. Overall, the trip was a very productive experience for our Clinic team and colleagues.

Conference participants and First Nation Dancers

Clinic awards 2017 - 2018

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

UHN Centre for Mental Health Fellow Research Award - 2017-2018

Dr. Elemi Breetvelt received this award in 2017 for his work in schizophrenia research, with the generous support of the Mukhurjee family. This acknowledges Fellows at UHN who have made outstanding contributions to the field of Mental Health. Dr. Breetvelt won this award again in 2018. Our former fellow, Dr. Erik Boot, was a previous recipient of this award.

Left to right: Dr. Anne Bassett, Dr. Elemi Breetvelt, Dr. Donna Stewart

Junior Investigator Award – July 2018

Our research analyst, Tracy Heung, was one of two recipients of the Junior Investigator Award for the most outstanding presentation by a junior faculty member at The 11th Biennial International 22q11.2 Conference in Whistler, BC. Tracy's presentation was titled "Predictors of All-Cause Mortality in Adults with 22q11.2DS". Ania Fiksinski, graduate student with Dr. Bassett, and a familiar face in Toronto, was the 2016 recipient of this international award.

Left to right: Dr. Anne Bassett, Tracy Heung

Advancing our knowledge of the heart: a study about Tetralogy of Fallot

We recently had the chance to interview Dr. Miriam Reuter, who worked with Dr. Anne Bassett on a new study about a congenital heart condition called tetralogy of Fallot (TOF) that is common in individuals with 22q. She told us about some of the exciting findings and what this means for our 22q community.

What was your role in the study?

I'm a genome analyst from the Ted Rogers Cardiac Genome Clinic at SickKids. We are analyzing whole genome sequencing data to identify molecular causes of different congenital cardiac disorders. One of our group's collaborations is with Dr. Anne Bassett, who has – over the course of many years – established a unique research study on the genetics of TOF.

Tell me a bit about your background.

I studied medicine in Germany. I received training in clinical genetics there, and did research on the genetics of, mostly, neurodevelopmental disorders. I came to Canada to work in research to analyze whole genome sequencing data, mostly regarding genetic causes of rare developmental disorders.

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

We studied genetic causes of one of the most common heart malformations in humans, TOF. We found that disruptions of a well-known pathway, called "VEGF", a very important cellular pathway, can be found at a high frequency in patients with TOF, in over one in every 10 patients who do not have 22q. Our aim is to understand why such congenital heart diseases such as TOF develop, and what molecular mechanisms are involved.

Why study tetralogy of Fallot, and what does this mean for patients with 22q?

The 22q11.2 deletion is the most common known cause for TOF. It is still uncertain why some individuals with the deletion have TOF, and what predicts the severity of this and other congenital heart defects. The genes and pathways we recently identified could also modify the risk for having a heart defect in individuals with a 22q11.2 deletion. This will be the subject of future research projects.

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

We hope to better understand why heart malformations such as TOF develop. We hope that if such heart defects occur, that we are better prepared to predict outcomes and to counsel families. Ultimately, of course, the aim is to find ways to prevent such malformations from occurring or to identify new and improved strategies.

We'd like to thank Dr. Reuter and the team of researchers for doing this great work and making an impact in the lives of people living with TOF. See page 11 for more information about this research.

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 20 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.

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.

Dr. Elemi Breetvelt: Clinic Fellow

Dr. Breetvelt has been our Clinical Fellow at the Dalglish Family 22q Clinic for 2017-2018. His fellowship was supported by a generous gift of the Mukherjee family.

Dr. Breetvelt trained as a child psychiatrist at UMC Utrecht in the Netherlands, where he followed children, adolescents and young adults with 22q. The main focus of his research is on the early factors that may indicate an increased risk of developing psychiatric illness. Dr. Breetvelt will be leaving to take a child psychiatrist position at SickKids Hospital in 2019. We wish him well!

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.

Research and Other Staff

Gladys Wong: Coordinator/research analyst

Joanne Ha, Tracy Heung, Kelly Kranics: Research analysts

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

Andrea Tyrer: Research Fellow

22q The Dalglish Family 22q Clinic

We also support and work with a number of outstanding students and clinicians, including:

Greg Costain: Senior Medical Genetics resident

Lily Van: Resident in Psychiatry, Clinician Scientist Program

Sarah Voll Malecki: Resident in Internal Medicine

Spencer Van Mil, Christian Fenn, Judy Truong: Medical students

Tommaso Accinni: Psychiatric Resident from Rome

Steven Reuver: PhD candidate, International Visiting Graduate Student from Utrecht

Cameron Watson: Senior Medical Student visiting from Cardiff, Wales

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!

Ongoing research at a glance: The Family Background Study

Many of our patients and families have had the pleasure of meeting Ania Fiksinski to participate in our family background study. We recently interviewed her, and she explained what this study was about and what's next for her exciting research.

Tell me about yourself: what are your educational background and work interests?

My name is Ania Fiksinski and I'm from the Netherlands, which is where I studied. I did a Bachelor's degree in Psychology and Linguistics. Then I did my Master's degree in Clinical Psychology, so I'm a psychologist by training, and I've worked as a psychologist in the Netherlands for about 6 years now. My current research is to try and understand the early course of 22q in adults. And this family background study is part of that.

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

The family background study is about the fact that we have very little understanding of why there are such big differences between different people with 22q, in terms of their symptoms. Some people develop treatable illnesses like anxiety or schizophrenia, but others do not. Some people function at an average level but some people have mild to more severe intellectual disabilities. We really don't have a clear understanding of why there are these big differences between different people who are missing the same piece of their chromosome 22. So with this family background study, we are trying to understand, if by looking at family factors, both genetic and those that are observable or measurable, we can identify factors that can help explain these big differences between people with 22q.

Can you give me an example of one of the tests people complete in this study? What are they called, and what's involved?

These tests are often a lot of fun. One of the things people do when they help us in this study is a brief cognitive assessment. This assesses different parts of the thinking processes. It gives us results about how peoples' verbal skills are developed, and how their more visual-spatial skills are developed. What the person would actually do for that part, for example, is make puzzles with blocks.

What's the most exciting part about working on this research for you?

It's very nice to be able to meet the parents and see how they are dealing with being a caregiver of someone with 22q, what they struggle with, what they find strength in. And also to see the differences or the resemblances between the parents and their children. I sometimes say that this study is an example of how different people are needed, and different professionals are needed, to get to a successful clinical evaluation of the patient, but also for a successful research study. It's not only me as a psychologist seeing the patient, but also the dietitian or the social worker or the doctor. It's really us as a team pooling our expertise together both for research and for clinical care. So I like that too.

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

Two things: one is more from a clinical perspective. I hope this study will give us some clues as to be better able to predict how things will be as an adult for someone who is born with 22q. So that we will be better able to say, "it's likely that your child will be able to live independently", or not, based on what we find from this study. The other thing,

Ongoing research at a glance: The Family Background Study

from a research perspective, is that I really hope it will give us some insight into understanding the huge differences between people with 22q. Some of this may be related in part to the variability in their parents. And by that I mean both in their genetic makeup and in their observable characteristics. Because if that would be the case, for example, for learning capabilities, this would be an important step forward.

Do you have any other projects in mind that you are planning on studying next?

It's not a specific project yet, but, I do hope that at some point we will be able to use this new knowledge to take the next step. In other words, investigate what we can actually do in order for us to prevent schizophrenia from developing, or to prevent any worsening of learning ability or functioning from occurring. That would mean, for example, a study where we would look at treatments and see if they are a success for people with 22q. This is my goal, and I think, a way that we should go forward.

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.

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

Greg Costain,
Senior Medical Genetics resident

Tracy Heung,
Research assistant

Lily Van,
Resident in Psychiatry, Clinician Scientist Program

Sarah Voll Malecki,
Resident in Internal Medicine

Spencer Van Mil,
Medical student

Judy Truong,
Medical student

Christian Fenn,
Medical student

Andrea Tyrer,
Research Fellow

Clinic events

22q at the Zoo

On June 10th, 2018, the 22q Deletion Syndrome Clinic at SickKids Hospital and the Dalglish Family 22q Clinic at Toronto General Hospital co-hosted the 7th annual 22q at the Zoo event. 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, which each run for about 90 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 our patients to meet each other, participate in some fun activities and to discuss topical subjects with staff facilitation.

For those parents and caregivers unable to attend in person, an on-line weblink is offered to give access to the group from home. Our most recent peer support group was held on November 1st, 2018. Please stay tuned for information about the next session!

22q Family Conference

On November 3rd, 2018, the 5th annual 22q11.2 Family Conference was held at SickKids Hospital in Toronto. This family run and family organized conference gave individuals with 22q and their families the chance to learn more about 22q and to meet other families sharing their same experiences.

The focus of the conference was education. Information sessions were primarily aimed at parents of young children, but also some for teens and young adults were offered on various topics. Our Clinic Social worker, Lisa Palmer, played a key role in presentations during the conference.

Transition Program

“How will 22q symptoms affect my life as I become an adult?” is a common question for many of our patients and their families.

The Dalglish Family 22q Clinic, in collaboration with the SickKids Hospital 22q Deletion Syndrome Clinic, offers a 22q Transition Clinic several times per year. This full day program gives teens with 22q and their caregivers information about adult care and enables a smooth transition from the 22q Clinic at SickKids to our Clinic at the Toronto General Hospital.

This program is one of the first in the world to focus on transition to adult care for patients with 22q. We look forward to holding our next 22q Transition Clinic. Please stay tuned for more information!

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 several other 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, 2018-2019

Canadian Institutes of Health Research (CIHR)
Five-year operating grant, 2014-2019

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.

Canadian Institutes of Health Research (CIHR)
Five-year project grant, 2016-2021

Canadian Institutes of Health Research (CIHR)
Planning and Dissemination grant, 2018, for our 22q meeting at Whistler

2018 CAP Research Internal Grant Program (UHN)
Clinic social worker Lisa Palmer, with help from Clinic dietitian Samantha D'Arcy, received a grant to fund a clinical research study entitled, "Complex Care Patients and Sex: Identifying and Addressing Issues Related to Sexual Health".

We are very grateful to these agencies for their research support!

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. Hanna Faghfoury & Dr. 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

Dr. Susan George,
Endocrinologist,
Toronto General Hospital

Dr. Danielle Andrade,
Neurologist,
Toronto Western Hospital

Dr. Connie Marras,
Neurologist,
Toronto Western Hospital

Dr. Anthony Lang,
Neurologist,
Toronto Western Hospital

Atlantic Canada

Dr. Sarah Dyack,
Medical Geneticist,
Halifax, NS

Dr. Pamela Forsythe,
Psychiatrist,
Saint John, NB

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

Asia

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

Europe

Ann Swillen, PhD
Child psychologist,
Leuven, Belgium

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

South America

Dr. Gabriela Repetto
Geneticist,
Santiago, Chile

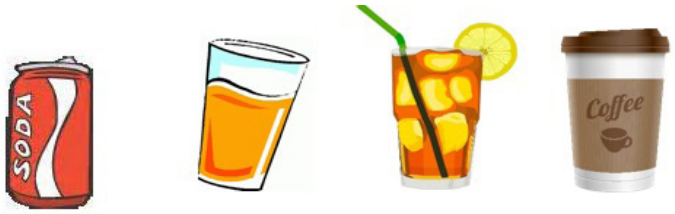
Australia

Linda Campbell, PhD,
Psychologist,
Newcastle, Australia

Healthy Drinks

Part of healthier eating is eating less added sugar. Many drinks have too much added sugar in them – look at how many teaspoons of sugar are in some common drinks:

Pop: 10 teaspoons in 1 can (350 ml) **Juice:** 6 teaspoons in 1 juice box (250 ml)
Iced tea: 7.5 teaspoons in 1 can (350 ml) **Coffee "double double":** 10 teaspoons in 1 large (550 ml)



10 tsp 6 tsp 7.5 tsp 10 tsp

Try to drink healthier choices such as water or milk more often.

Help with finding a job

A job is a great way for you to have structure in your life and can help you to feel good. The government of Ontario has supports available to help you find a job that may be a good match for you. Check out some resources below:

Youth Job Link

You have to be 15 - 29 years old.

You have to live in Ontario.

This program can help with resume writing, preparing for interviews, job searching and matching you up with employers, and learning skills needed for a job.

Youth Job Connection

You have to be 15 - 29 years old.

You have to live in Ontario.

You can't be in regular school or training.

This program can help with at least 60 hours of paid job training, up to 6 months of job placements, mentorship and coaching.

Youth Job Connection Summer

You have to be 15 – 18 years old.

You have to live in Ontario.

You have to be in high school (or just completed your last year).

This program can help with at least 20 hours of paid job training, a summer job for up to eight weeks, mentorship and coaching.

Ontario Disability Support Program (ODSP) – Employment Supports

You must be receiving ODSP.

This program can help with community supports for getting and keeping a job.

Get in touch with our Clinic by calling (416) 340-5145 to ask more questions about job supports that might be good for you.

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!

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.

New 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 yourselves and other people with 22q worldwide!

Elucidating the diagnostic odyssey of 22q11.2 deletion syndrome

Palmer LD, Butcher NJ, Boot E, Hodgkinson KA, Heung T, Chow EWC, Guna A, Crowley TB, Zackai E, McDonald-McGinn DM, Bassett AS

Individuals with 22q are often undiagnosed for years because this condition is not easily recognizable. This study analyzed the time it took for a diagnosis of 22q and reasons for delays across age and groups in Toronto and Philadelphia. Problems with the palate and heart were associated with shorter times to a 22q diagnosis. Non-European ancestry lead to longer times. There is a great need for education about 22q so that the condition is more widely known.

Neuropsychiatric expression and catatonia in 22q11.2 deletion syndrome: an overview and case series

Butcher NJ, Boot E, Lang AE, Andrade D, Vorstman JA, Bassett AS

Catatonia is a set of symptoms that include abnormal involuntary movements and behaviours that sometimes occur in individuals with psychiatric conditions like schizophrenia and depression and in some neurological diseases. In this study, the authors provide an overview of the psychiatric and neurological symptoms and conditions associated with catatonia in adults with 22q. The results may help with the diagnosis of catatonia so that effective treatment can be provided as early as possible.

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.

In this study we investigated specific areas of neurocognition in 99 adults with 22q, identified areas of relative strengths and weaknesses, and found that the better one's functioning in certain skills (called Executive Performance), which could potentially be improved through cognitive interventions, the better one's daily-life functioning as an adult. These results may help caregivers and clinicians to recognize the relative strengths and weaknesses in learning of people with 22q.

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, Mil SV, Wald RM, Walker S, Marshall CR, Silversides CK, Scherer SW, Kim RH, Bassett AS.

The authors discovered that some patients with tetralogy of Fallot carry variants in the VEGF pathway genes, which code for proteins important in blood vessel and heart development. The variants include ones that change the protein sequence and are expected to cause the protein to malfunction, as well as those that lead to partly-deleted protein. This research supports the idea that changes in the VEGF pathway genes are involved in the formation of tetralogy of Fallot.

Presentations

Each year we participate in several presentations on 22q and other related conditions. Some of our recent presentations relating to 22q include:

Advanced Issues in Dual Diagnosis

Centre for Addiction and Mental Health Dual Diagnosis Specialty Service

Toronto, Ontario - March 21st, 2018

In this meeting for professionals working with patients with complex developmental and mental health issues, our Clinic fellow, Dr. Elemi Breetvelt, spoke about "22q11 Deletion Syndrome and the Criminal Justice System". He provided insight on issues that may arise when law enforcement officers interact with adults with 22q.

Phoenix Children's Hospital 4th Annual 22q Family Symposium

Phoenix, Arizona - May 19th, 2018

Dr. Anne Bassett was the keynote speaker at this family event, where the theme was "Transition to Success". She spoke about health issues associated with 22q over the lifetime, especially in teenagers and adults.

5th Annual 22q Family Conference

Toronto, Ontario - November 3rd, 2018

At this conference geared towards young patients and their families at SickKids Hospital, Clinic social worker Lisa Palmer facilitated workshops about advocacy and disclosing a diagnosis of 22q.

In July 2018, we presented our research projects at the International 22q11.2 Conference in Whistler. Here is a small sample of some of the presentations given by our Clinic team:

Dr. Anne Bassett: Medical Multimorbidity in Adults with 22q11.2DS

Dr. Elemi Breetvelt: A Normative Chart for Cognitive Development in 22q11DS: Implications for 22q11DS and Beyond

Ania Fiksinski: The Impact of Parental IQ on the Variable Penetrance of Intellectual Impairment in 22q11DS

Dr. Erik Boot: Parkinsonian Motor Features in Adults with 22q11.2DS

Tracy Heung: Predictors of All-Cause Mortality in Adults with 22q11.2DS

Spencer van Mil: Late Mortality in a Genetic Subtype of Tetralogy of Fallot

Joanne Loo: Personalized Medical Information Cards for Adults with 22q11.2DS

Lisa Palmer: Identifying Issues Related to Sexual Health in Adults with 22q11.2DS

Lily Van: Treatment of Schizophrenia in 22q11.2DS

Samantha D'Arcy: Characterizing Cooking Habits and Confidence in Food Skills in Adults with 22q11.2DS

Our extended Toronto team!

Further Reading

Ever wondered how you can make the most out of your medical appointments? In October, we put together tips for "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 recently 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/

Online Resources: Job Supports

Here are some websites about finding a job:

Ontario Government – Employment supports for people under 30

www.ontario.ca/page/employment-programs-people-under-30

For individuals younger than 30, information about paid opportunities, summer jobs, skills training, mentoring and more.

Ontario Disability Support Program – Employment supports

www.mcss.gov.on.ca/en/mcss/programs/social/odsp/employment_support

For individuals receiving ODSP, and would like to access support in your community for getting and keeping a job.

Cannabis Legalization in Canada

On October 17, 2018, using cannabis ("marijuana" or "weed"), was legalized in Canada. Here is some important information you should know.

Our Clinic team continues to advise our patients not to smoke weed. Why?

1. Your health:

Many people with 22q have heart conditions and may struggle with learning difficulties, anxiety or schizophrenia.

Using weed can be dangerous for people with heart conditions.

Using weed can cause psychotic episodes, for example, hallucinations and paranoia.

Marijuana tends to worsen memory, concentration and the ability to think and make decisions.

2. Problems with the law:

It is still illegal to cross the Canadian border to the United States with weed.

Using weed and driving is illegal. Driving high increases your chance of being in a crash, and immediate effects of smoking weed can last for more than 24 hours.

It is illegal to use weed at work.

We can help!

If you have questions about the safety of using marijuana in your own case, please contact our Clinic at 416-340-5145 to make an appointment with one of our doctors.

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

International 22q11.2 Deletion Syndrome Foundation:

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.

In Memoriam

The Dalglish Family 22q Clinic

Sadly, over the decades and including very recently, we have lost members of our 22q family. They will never be forgotten and remain close to our hearts. Their research contributions live on, and through this knowledge 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 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

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