PRISMA

PRECISION MEDICINE IN AUTISM

LEARN MORE ABOUT US

Meet the team members of our PRISMA Research Group

HOW DOES GENETICS WORK?

Meet Bo & Dina, who will let us know

BRINGING IT ALL TOGETHER

Research, Clinical Care, Education, and Community efforts under one roof

WHY PRISMA

Prisms are wonderful objects; a simple but elegant piece of glass that can take a single stream of light and transform it into a beautiful spectrum of colors, which can be used to shine light in dark places so that we can better see the way.

That description recapitulates the mission and vision of our Precision Medicine in Autism (PRISMA) group; we aim to understand the unique strengths and challenges of people with autism or other neurodevelopmental conditions in the context of the diverse rare genetic changes that can be found in up to 1 in 3 people with these diagnoses. By bringing together research, clinical care, education, and community engagement, we focus on creating a comprehensive strategy to improve the healthcare of people with psychiatric conditions stemming from rare genomic disorders. Plus, it highlights our focus on diversity, as "prisma" is the Spanish word for prism!

Welcome

to PRISMA Research Group

"Dr. Moreno De Luca is also a musician and he recently released a song inspired by his experience getting to know individuals with Autism! The song is called "Outer Space"- Click here to listen the song"

It's a pleasure for us to share with you our inaugural PRISMA newsletter! Along with its challenges, this past year has brought the opportunity to grow several of our efforts in PRISMA, which you will learn about here.

Among the good news, we received a large grant from the National Institute of Mental Health (NIMH) to study the mental and physical health of people with a specific rare genetic change, 17q12 copy number variants (CNVs), and understand how additional genetic factors may influence any clinical features. On the clinical side, we have grown our genomic psychiatry consultation service, where we provide clinical care to people with mental health conditions stemming from rare genetic changes, and created a new genetic counseling service to help make clinical genetic testing available for families who need this. We have also launched a clinical project where we have been offering standard of care genetic testing (chromosomal microarray and Fragile X testing) to all patients currently hospitalized in the autism unit at Bradley Hospital. We have continued to develop educational material for physicians in training and families about the relevance of genetics in psychiatry, and launched a new Autism Track as part of the Child and Adolescent Psychiatry Fellowship training program at Brown University.

Most importantly, we have been able to continue fostering our relationship with the families we serve in research and clinical care, gathering key insights into their priorities and working together to make sure their interests and needs are well represented in all our efforts.

I hope you enjoy reading this newsletter as much as we enjoyed creating it. As the director of the PRISMA group, thank you for working with us along this path.

Daniel Moreno De Luca, MD MSc
Director & Principal Investigator PRISMA

Jack Biedermann
Research Assistant

Molly Goldman
Genetic Counselor

Carrie Best
Project Manager

Bo & Dina
Teachers and Owners
Prisma Library of Life

Julia Katz
Child and Adolescent Psychiatry
Fellow, Autism Track

Daniel Moreno De Luca
Director & Principal Investigator
Child and Adolescent Psychiatry

Discover our Team

We are very happy to introduce the team members of our PRISMA Research Group, including the new additions to our team!

Jack Biedermann is our research assistant, whose primary role is to support families through recruitment, enrollment, and assessments for our research studies, and to help create and maintain the PRISMA database. He graduated from Tufts University where he earned a BA in Psychology.

Carrie Best is our research coordinator. She holds a Master of Public Health degree in Health Behavior and Health Education from the University of Michigan School of Public Health, as well as undergraduate degrees in Sociology and Music Performance.

Julia Katz is currently a Child and Adolescent Psychiatry Fellow at Brown and is our inaugural Autism Track Fellow. She received her medical degree from the University of North Carolina at Chapel Hill School of Medicine, and completed her Psychiatry Residency at the Icahn School of Medicine at Mount Sinai in NYC. Her clinical and research interests include developmental disabilities and ASD Julia speaks 3 languages (English, Hebrew, Russian) and a little bit of ASL.

Molly Goldman is our certified genetic counselor with experience in a variety of clinical and research environments. By managing the Genetic Counseling Clinic and providing pre and post test counseling to families with autism & neurodevelopmental diseases, Molly hopes to expand the availability of, and access to, medically recommended genetic tests. Her role extends as part of the PRISMA research team to assure that the very best care is delivered to each of our participating families.

Daniel Moreno De Luca is a physician and scientist specializing in child, adolescent, and adult psychiatry with advanced training in neurogenetics. He splits his time between clinical care, where he see people with autism spectrum disorder, bipolar disorder, schizophrenia, or other mental health needs who also have an underlying genetic cause for these, and research on the rare genetic causes of mental health conditions and how we can use this information to provide more specific clinical recommendations.

Last but not least, **Bo & Dina** run the PRISMA Library of Life, which you can visit on our website below. Bo loves fixing things and collecting old clocks, and Dina likes learning everything about dinosaurs and loves discovering new things in books.

The role of these two new members is to help us learn more about genetics and genetic testing in the context of autism spectrum disorders in a fun way. Their participation in our group is part of an important implementation strategy using media to create a bridge between community and science so that families have readily available information delivered to children and adults alike, in a fun and accessible way. We hope that these materials will be used to start a conversation with clinicians and empower families to be active participants in their healthcare as it relates to genomics.

We are also lucky to count on guest appearances by key teammates who make all of this possible, including **Silvana Guerrero**, our marketing expert who is in charge of developing this newsletter, our web and our flyers, and **Manuel García**, **Wilson Cáceres** and **Juan Fernando Arango**, who have helped us create the video where you will meet Bo and Dina.

Thanks to the **PRISMA** team and especially to the families who make these projects possible and help us advance Precision Medicine strategies for autism spectrum and other neuropsychiatric conditions.

PRISMA SPOTLIGHT

In our first edition of the PRISMA newsletter, Daniel Moreno De Luca, Director & Principal Investigator at PRISMA, shares more about research at PRISMA. He's sharing with us his vision and work for the 17q12 community throughout the last seven years and into the future. To learn more about 17q12 and see additional resources, visit our website: precisionmedicineinautism.org/17q12-cnvs

How did you become interested in 17q12 syndromes?

I first became interested in 17q12 CNVs (copy number variants) when I was a postdoctoral fellow in neurogenetics at Emory University, when we started seeing that some people with autism or developmental disabilities that were sent for clinical genetic testing were being diagnosed with a 17q12 deletion or duplication. Up until then, these mental health diagnoses had not been previously described in people with 17q12 CNVs, and by launching a large international study that included over 70,000 people worldwide, we were able to show that 17q12 deletions increased risk for autism and schizophrenia. I had the pleasure of meeting several families with 17q12 CNVs, and this was the most rewarding aspect of this research and what got me most interested in choosing to work on understanding the clinical consequences of this rare genetic change; I very much treasure the life stories that they have allowed me to learn about and the long-lasting bonds we have established with a group of families that have now blossomed and grown into the 17q12 Foundation.

17q12 FOUNDATION

The 17q12 Foundation was born from the group that was initially organized through Geisinger Health System for their research study. At the family conference in Chicago in 2017, a group of parents and a genetic counselor decided it was time for us to embark on the journey toward becoming a free-standing non-profit organization dedicated wholly to increasing awareness and driving research forward for both the deletions and duplications. For more information visit their website www.chromo17q12.org/

RESEARCH

What does your research project focus on specifically for 17q12?

At PRISMA, our research group at Bradley Hospital and Brown University, we are focused on understanding the mental health and medical "blueprint" that is associated with 17q12 CNVs, as well as other background genetic factors that may influence whether someone with a 17q12 CNV develops any of the clinical features that have been previously described in this population. We particularly want to focus on moving to "higher resolution" behavioral and medical diagnoses - for example, learning not only how common is a diagnosis like autism spectrum disorder in people with 17q12 deletions, but more importantly, how do people with 17q12 CNVs do on the social functioning scale regardless of whether they meet criteria for an autism diagnosis. Likewise, we want to understand how common a diagnosis like diabetes is, but also more importantly, where do the blood sugar values lie over time for people with 17q12 CNVs irrespective of whether they have a diagnosis of diabetes. This will ultimately allow us to move from a yes/no diagnosis, where a clinical condition is either present or absent, to a nuanced understanding of these clinical areas that tell us about people's areas of strength and challenges. A simple but useful example would be to compare this to boiling water for a nice cup of tea; moving from asking whether water is boiling at any given time (like a yes/no clinical diagnosis), to knowing the actual temperature of the water ("high resolution" quantitative, continuous traits like sociability or blood sugar levels). Lastly, we want to know the genetic factors in the rest of the genome that may contribute to someone developing these diagnoses, even if they all have a 17q12 CNV. Going back to our boiling water example, we want to understand why water boils more easily in some conditions like high altitude above sea level (background genetic factors, in our analogy), even if we have the same temperature for all (17q12 CNVs in this e.g.)

COMMUNITY

How will your research project benefit the 17q12 community?

Our hope is that by understanding the areas of clinical strengths and challenges regarding mental health and other medical diagnoses and their additional genetic influences in people with 17q12 CNVs, we will be better able to individualize treatment for those who need it, a prime example of what we call Precision Medicine. In fact, we have created the Genomic Psychiatry Consultation Service at Bradley Hospital and Brown University where we see people with mental health conditions who have had genetic testing that revealed a genetic cause for this (including 17q12 CNVs, and other genetic changes) to provide actionable clinical recommendations based on this genetic information. The more we learn with studies like the one we are carrying out, the more specific the knowledge and medical help we can then transmit back to families in the future! If you or anyone in your family has a 17q12 CNV and is interested in learning more, please dont hesitate to contact us our email prisma@lifespan.org

17q12 Conference 2019

We cant believe it's already been 3 years since we had the pleasure of hosting the last International 17q12 CNV Family Meeting! It was a treat getting to chat directly with families, share results from our ongoing studies, bring expert clinicians, researchers, and community advocates together, and hear about priorities directly from the 17q12 families, all while enjoying the New England summer. We're looking forward to continuing these connections and celebrating difference as a team. In the words of one of the participants, "it was all a reminder that it is cool to be unique!".

Experts presented clinical updates in different areas related to 17q12 deletions and duplication

We had a special visit from our friends at the Big Nazo Lab

The families enjoyed a RI classic, Del's lemonade

The Kids enjoyed music therapy with the Hands in Harmony group

Families had the chance to visit the Roger Williams Zoo and its wildlife during the second day of the conference

Conference Data:

The third conference was a success
Here are some facts :

- 20 families
- 42 Adults
- 25 Kids
- 184 Remote Participants

Positive Exposure

With the common goal of celebrating difference, we've partnered with Rick Guidotti and Positive Exposure to recast the way that people with genetic conditions are portrayed and to bring back their stories. We asked him about what keeps him going:

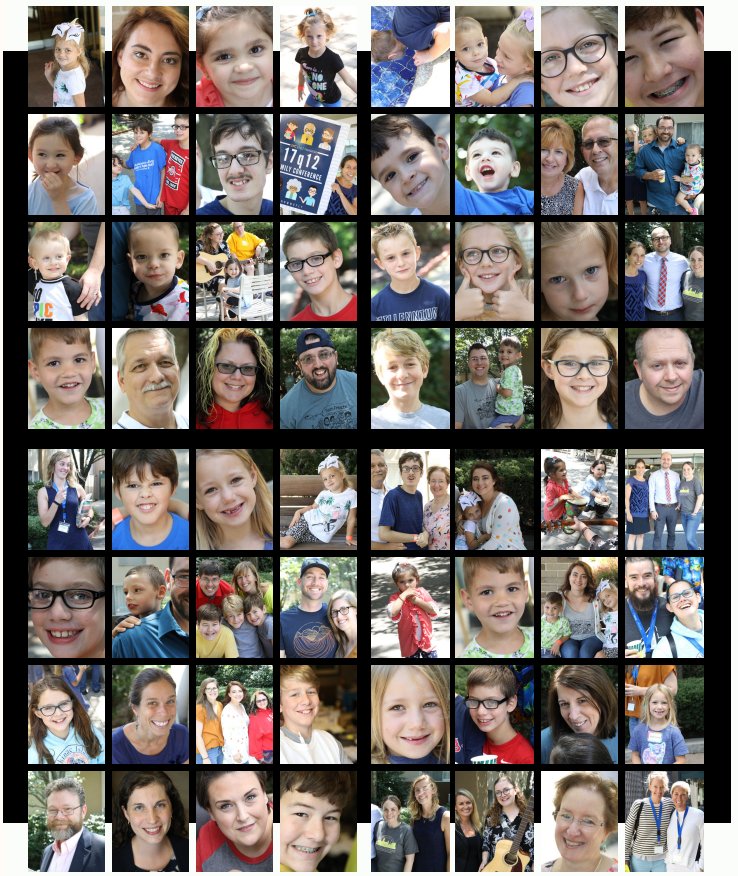
"Since Positive Exposure's inception 25 years ago it has always been our goal to collaborate with individuals and communities that are at risk of stigma and exclusion. We have worked with so many different communities: genetic, physical, behavior and intellectual difference, of course.

We're always about celebrating the beauty and richness of our shared humanity, seeing humanity first, seeing a person first, helping a healthcare provider in training understanding that it's never what you see always who you are treating, and steadying the public's glance so that they don't look away when they see someone that is different because of fear or stigma, etc.

We're so aware after working with many of our communities, that metal health challenges carry the greatest stigma but at the same time are invisible. So many important conversations needed to be had. We feel we create a vehicle using the arts photography and film. It really creates an opportunity to have these sometimes difficult conversations but nonetheless, have those conversations about stigma associated with mental health challenges. We create a space to feel safe and to share and to be proud of our individuality and uniqueness. It's our uniqueness that unites all of us. This isn't a story about mental health victims this is the story of individuals living with mental health challenges.This is a story about us, all of us.

It's creating an opportunity for us all to feel safe in that space, so that we can all address our own specific challenges and feel comfortable and not attacked for them and not blamed for them, but to realize that we are all living in that space and can have those conversations we need, both in the medical environment and broader public arena as well. We need to have conversations about mental health and take a look at stigma.

Our goal is always to eradicate stigma. I don't know if that will happen in my lifetime, but we'll work as hard as we possibly can, and do as best we can, to at least bring that down"

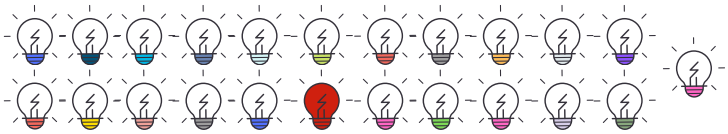


Check all the awesome jobs that they are doing for the medical and families communities at **www.positiveexposure.org**

What are 17q12 CNVs?

The 17q12 Foundation represents two separate syndromes:

17q12 deletion syndrome



is caused by a missing piece of chromosome 17 (deletion) that is present from the moment the child is conceived

17q12 duplication syndrome

is caused by an extra piece of chromosome 17 (microduplication) that is present from the moment the child is conceived

1 in 14500 people

in the general population have this deletion syndrome. It's more common in populations with developmental disorders developmental delay, intellectual disability autism spectrum disorder) and schizophrenia

The deletion is most often a brand new (de novo), sporadic event in the person that is diagnosed

of the time a person with the deletion will have inherited it from a parent

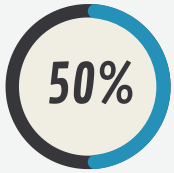
chance that each of that parent's children will also have the deletion

chances that a sibling has if it was de novo (both parents tested negative)

1 in 2500 individuals

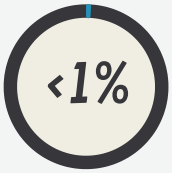
in the general population have this duplication syndrome. It's more common in populations with developmental disorders (developmental delay, intellectual disability autism spectrum disorder) and schizophrenia

The duplication is most often inherited from a parent. Oftentimes, the duplication is only identified in parents after the child is diagnosed and could have similar, milder or apparently no features



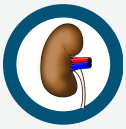
chances that each of that parent's children will also have the duplication

of people with the duplication will have a brand new (de novo) duplication that wasn't inherited from either parent



chances that a sibling has the duplication if it was de novo (both parents tested negative)

A syndrome is defined as a recognizable group of signs and symptoms that consistently occur together.



Kidney and urinary tract abnormalities

Macrocephaly (Large head size)

Maturity onset diabetes of the young type 5 (MODY-5)

Neurodevelopmental/Psychiatric: developmental delay, autism spectrum disorder, learning disabi-lity, intellectual disability, anxiety, bipolar disorder, schizophrenia

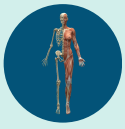
It's important to remember that no two people with the deletion will have the same combination and/or severity of symptoms, even people within the same family

The most common features of the duplication are related to neurodevelopment.

Skeletal differences

Microcephaly (small head size)

Seizures in up to 75% of individuals



Hypotonia (low muscle tone)

Neurodevelopmental/Psychiatric: intellectual abilities ranging from typical to severe disability, speech delay, motor delay, behavioral concerns (aggression, compulsive disorders), autism spectrum disorder

It is important to remember that no two people with the duplication will have the same combination and/or severity of symptoms, even people within the same family

For More Information
visit www.chromo17q12.org/

INFOGRAM MADE BY PRISMA RESEARCH

PRECISION MEDICINE IN AUTISM - RESEARCH PROGRAM

PRISMA STUDY

Our research study "A Genomic Approach to Precision Medicine for Autism and Neurodevelopmental Conditions" is enrolling children and adults who have had diagnostic genetic testing that showed a deletion or a duplication in chromosome 17q12 to understand how genetics can impact their healthcare. The study will take approximately 6 hours, and involves collecting health information and completing questionnaires and assessments online.

To participate, contact Dr. Daniel Moreno De Luca or the PRISMA research staff at +1 780 492 4467 or prisma@ualberta.ca

CASA Mental Health | UNIVERSITY OF ALBERTA | Alberta Health Services

REB ID (Pro00129967) - DATE: August 29, 2023

Elsewhere in the genome

15q13.3 deletion

Interview By Julia Katz

"Being a mom of a child with disability is the most special thing in the world. I was meant to be his mom, we have a special bond. He's my whole world and I feel so blessed. S is my best friend and I love him more than I can tell you."

We all have unique stories to tell. For the first edition of our newsletter, we wanted to share the story of one of the families that has partnered with us to understand better the impact of genetics on mental health. It's a story about kindness, challenges, and growth, and it underscores the importance of advocacy across research and clinical care, but most importantly, within communities that can work together towards a shared goal.

We spoke with Ms. G, who told us a bit about her experience of being the mother and daughter of someone with a 15q13.3 deletion, and finding that she had this rare CNV herself in the quest for answers. She has done a fantastic job advocating for the 15q13.3 deletion community, and she created a group called "Unique and Special 15q13.3 Microdeletion".

She learned that her son had the 15q13.3 microdeletion back in 2008, when he was 38, after his psychiatrist ordered a genetic test called chromosomal microarray. "I thought: he doesnt have anything like that. Nobody ever said he could have something like that or that we needed to do genetic testing. Nobody. Two weeks later, I got a phone call, and they said your son has a 15q13.3 microdeletion. I cried for two or three days because I knew that doctors had been missing something. I'm a carrier and I felt bad at first. I blamed myself, but my mom and sister have it too. It would've been helpful for the family to know about that earlier. Before genetics, he got a lot of different diagnoses."

"He has a heart of gold and a really good sense of humor", says Ms. G. "He is 52 years old now and he likes to watch movies and sports with me. He loves John Wayne and Chuck Norris movies, as well as old western movies. A few years ago he was in a group home where they wrote to Chuck Norris and Chuck Norris wrote back with a picture and a signature! He loves mom's homemade burritos and football and Tom Brady. He used to work at a grocery store, and a food court at a mall; all vendors loved him. He was also in special Olympics and ran track, bowling and golf."

Unique and special 15q13.3 microdeletion

Ms. G had to figure out a lot of things on her own while her son was growing up. They moved around a lot and found mental health services as a helpful first step that connected them to other resources like case managers, social security, and information about guardianship. "When I lived in Georgia I went to NAMI (National Alliance on Mental Illness) It helped being able to talk to other parents."

"I joined a group when I found out about the diagnosis. After that, I started reading about how to start a group and started my own group, which I named Unique and Special 15q13.3 Microdeletion. At least once a week a new parent joins the group; we have around 500 parents, including people from England and other countries. Somebody will say - I'm new to the group, help , I'm not sure what to do – we try to help them and give them advice, and support each other throughout. It is a good way to connect with others about research, resources, and emotional support. We have also had a positive experience with docs and wanted to have them involved in the group.

What advice would you offer to parents navigating the system new to the 15q13.3 community?

"Mental health is the place to start, get a good neurologist and good psychiatrist. It is hard to find good psychiatrists" says Ms.G, stating that they have been lucky with their current psychiatrist, of which they speak very highly. "Advocate for education, speak with genetic counselors, do your own research"

What has it been like to advocate for your son?

"I always advocated for S." says Ms. G, who then refers to experiences from other families, "Because some can't advocate for themselves, we have to do it for them. I've always gone to mental health services, which helped connect with other services. The genetic diagnosis allowed him to get more services".

15q13.3 Deletion

Is a chromosomal change in which a small piece of chromosome 15 is deleted in each cell. The deletion occurs on the long (q) arm of the chromosome at a position designated q13.3

THIS CHROMOSOMAL CHANGE INCREASES THE RISK OF

INTELLECTUAL DISABILITY

that is usually mild or moderate. Many of these individuals have delayed speech and language skills.

BEHAVIORAL PROBLEMS

including a short attention span, aggression, impulsive behavior, and hyperactivity

PSYCHIATRIC DISORDERS

particularly schizophrenia or bipolar disorder

SEIZURES (EPILEPSY)

in about one-third of people with this chromosomal change

DEVELOPMENTAL DISORDER

that affects communication and social interaction (autism spectrum disorders)

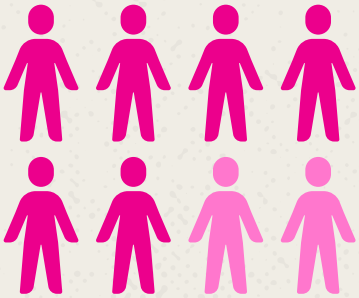
OTHER SIGNS AND SYMPTOMS

can include heart defects, minor abnormalities involving the hands and arms, and subtle differences in facial features

However, some people with a 15q13.3 deletion do not appear to have any associated features.

15q13.3 DELETION LIKELY OCCURS IN ABOUT

PEOPLE IN THE GENERAL POPULATION



75% individuals with 15q13.3 deletion inherit the chromosomal change from a parent

15q13.3 microdeletion is **inherited** in an autosomal dominant pattern, which means one copy of the deleted region on chromosome 15 in each cell is sufficient to increase the risk of intellectual disability and other characteristic features. However, not everyone with the deletion is going to have the same symptoms, or any symptoms at all.

In the remaining cases, 15q13.3 microdeletion occurs in people whose parents do not carry the chromosomal change, which we call **de novo**. In these individuals, the deletion occurs most often as a random event during the formation of reproductive cells (eggs and sperm) or in early fetal development.

IMPLEMENTATION SCIENCE

Our first step to identify opportunities and challenges for the implementation of genetic testing in autism and developmental disorders was to ask the ASD community directly about their perception and knowledge of genetics, as well as their experience accessing genetic testing within clinical care. Their answers were simultaneously enlightening and sobering, revealing the important work that lied ahead to address barriers for genetic testing at different levels. We were eager to put in place comprehensive strategies to address these challenges, and here we outline several of our efforts for this purpose within quality improvement, education and clinical care informed by the Consolidated Framework for Implementation Research. This has only been possible by working together with families, advocates, clinicians, educators, scientists, and hospital administration. Lastly, but importantly and in line with our strong commitment to equity, all clinical services and as well as our educational and outreach material for the community - including this newsletter - will available in English and Spanish).

QUALITY IMPROVEMENT

Now on its sixth year, the genomic psychiatry quality improvement project at Bradley Hospital, spearheaded by Dr. Moreno De Luca in close collaboration with the Verrecchia Clinic for Children with Autism and Developmental Disabilities, and the Child Psychiatry Fellowship program at Brown University and Bradley Hospital, has achieved important milestones in improving access to genomic information for ASD families.

Successes include:

- Creation of Genetic Counseling Service
- Creation of Genomic Psychiatry Consultation Service
- Procurement of administrative support to help with the preauthorization process for genetic testing
- Direct negotiation with multiple genetic testing companies to obtain the most competitive alternative
- Implementation of genetic testing as part of the inpatient hospitalization protocol for patients on the spectrum receiving their care at Bradley Hospital

We have been working hard on next steps, and can't wait to update you on what's to come!

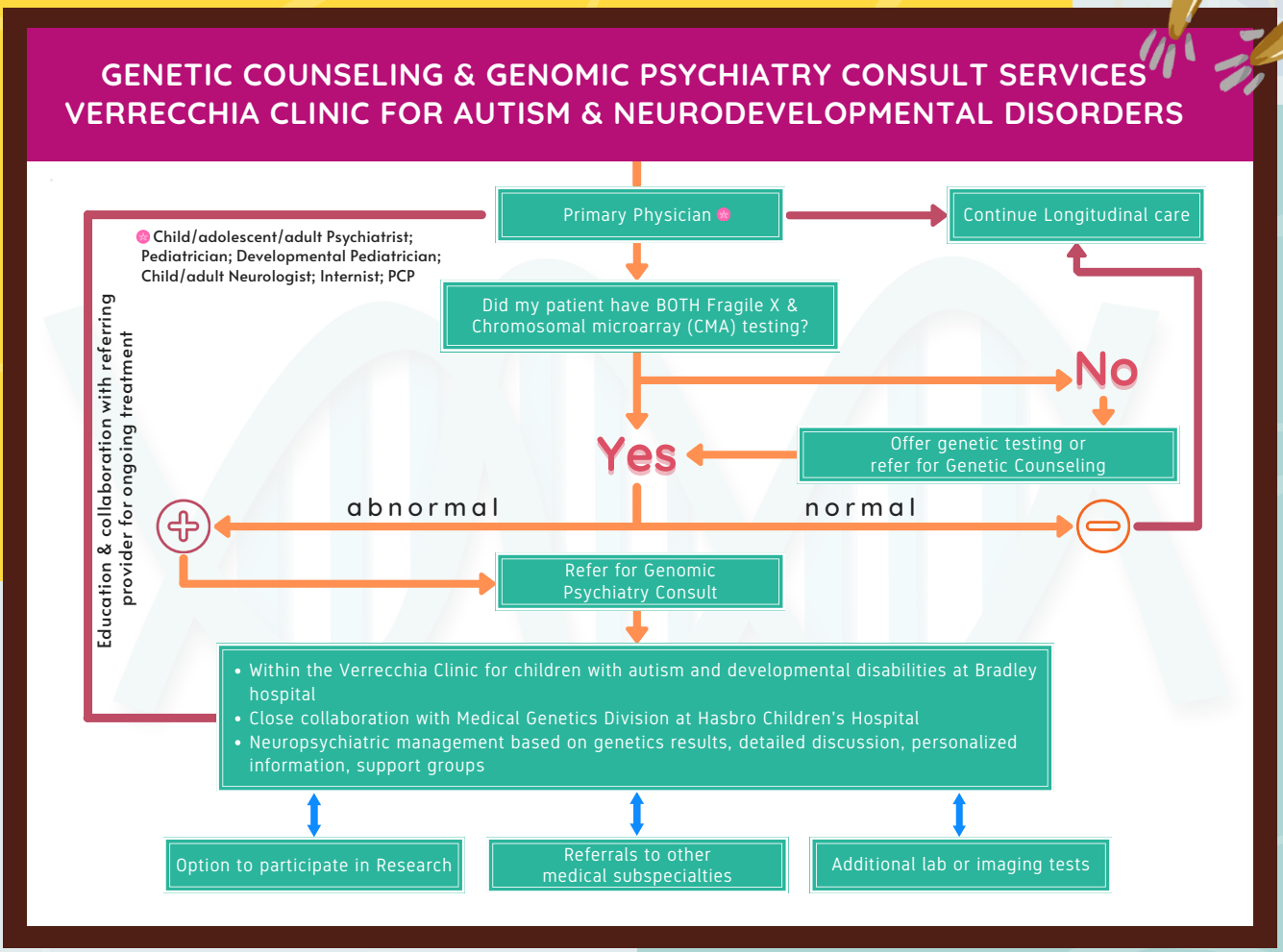
EDUCATION

We have recently launched a **new specialty track in autism spectrum disorders** within the child and adolescent psychiatry fellowship at Brown University and Bradley Hospital. Through this program, we aim to bolster expertise in autism spectrum disorders for clinicians and researchers, who will then serve as a resource for the ASD community nationwide, building on our previous education initiatives in ASD such as **coursework** for psychiatry residents, child psychiatry fellows, psychology interns and residents, social workers, and the general public. We had the privilege of receiving our inaugural **ASD track fellow** this year, Dr. Julia Katz, whose work you can see prominently featured in this newsletter! We have also launched a new **ASD journal club** with our physician colleagues across healthcare systems in RI, and continue to welcome child and adolescent psychiatry fellows in our ASD clinic - all of whom have continue to serve the ASD population in RI, reinforcing our ability to deliver excellent care in our own backyard. Lastly, we are eager to launch a new video aimed at kids on the autism spectrum and their families, explaining in a clear and engaging way key concepts like autism, genetics and genetic testing. **Watch the video in our website, or click here**, where you'll meet Bo, DiNA, and the rest of their friends!

CLINICAL CARE

Are you curious about the new clinical services we described within our quality improvement efforts earlier? Find out more about them below.

GENETIC COUNSELING & GENOMIC PSYCHIATRY CONSULT SERVICES
VERRECCHIA CLINIC FOR AUTISM & NEURODEVELOPMENTAL DISORDERS



GENETIC COUNSELING SERVICE

Led by our genetic counselor, Molly Goldman MS CGC, and in collaboration with Daniel Moreno De Luca, MD MS, we have created this service to address the large gap between professional clinical recommendations for genetic testing in ASD and actual clinical practice. Here, we see kids and adults with a clinical diagnosis of autism spectrum or other neurodevelopmental disorders to discuss genetic testing, carry it out if they agree, and deliver the results, in close partnership with their current clinical team.

GENOMIC PSYCHIATRY CONSULTATION SERVICE

Aware of the growing need for expertise on how to translate results of clinical genetic testing into actionable recommendations within psychiatry, we created the Genomic Psychiatry Consultation Service. Through this process, we evaluate people with autism or developmental disorders who have previously had genetic testing that revealed a pathogenic, or abnormal, genetic result, and use that genetic information to tailor their medical management. We have expertise in the breadth and depth of rare genetic changes, which allows us to understand their impact on psychiatric and medical features, and put recommendations in place that account for this.

Genetic Testing

The information in our genome can be compared to an encyclopedia organized into books (chromosomes) that spell out all the words (genes) containing the instructions for making us who we are. Looking closely at this material can indicate genetic changes, akin to changes in spelling or extra or missing paragraphs or whole books, that might explain why someone has a given medical or psychiatric condition. Although autism cannot be diagnosed with a genetic test, it is very important to look at the genome of people on the autism spectrum after a diagnosis is made, as finding a genetic change can help people understand better the reason for autism in their case, and give doctors important information to help when needed

CHROMOSOMAL MICROARRAY

This test looks for any missing or extra pieces of DNA throughout the entire genome, and tells us what specific part of the encyclopedia is involved. Because it looks at every single book, it can uncover big and small changes alike, and tell us whether a genetic change is likely causing clinical symptoms or not.

FRAGILE X TESTING

It looks at one single word, or gene, on the X chromosome, as genetic changes in this gene are one of the most frequent causes of autism and cause Fragile X syndrome.

EXOME AND GENOME SEQUENCING

These tests too look at our entire genome, but they are also able to read each of the words in our encyclopedia, our genes, to detect any changes in their spelling. Some of these changes don't lead to clinical symptoms, while others change the function of our genes; in those cases, we call them mutations.

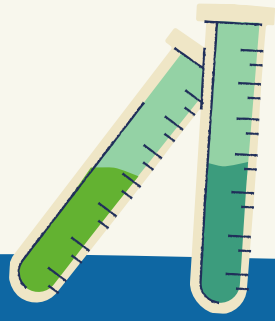
It is important to remember that the decision about testing is entirely up to families, just as any other medical test. Be sure to ask your doctor or team any questions you have as you make this important decision! Check the complete video: https://youtu.be/iXvd_GEdNCE

Genetic Testing

STEPS:

It's as easy as your ABC's

Cheek swab "Buccal" kit sent directly to your home or go the lab for blood sample

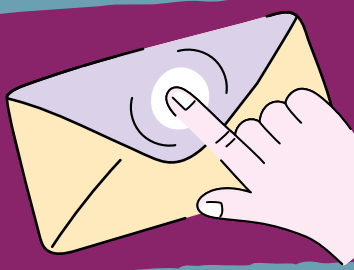


Collect BUCCAL/BLOOD sample:

- Dont eat, drink or chew gum 30 min prior
- Label with DOB & collection date

Sign CONSENT

Send SAMPLE to lab



I told my genetic counselor that I APPROVED, want genetic testing!

If you have question contact us at prisma@ualberta.ca

WHERE IS BO?

FIND OUR PRISMA BEAR BO, He's hiding in 7 different places throughout the PRISMA village!

PRECISION MEDICINE IN AUTISM - RESEARCH PROGRAM

PRISMA STUDY

Our research study "A Genomic Approach to Precision Medicine for Autism and Neurodevelopmental Conditions" is enrolling children and adults who have had diagnostic genetic testing that showed an abnormal (also called pathogenic) result into a research study to understand how genetics can impact their healthcare. The study will take approximately 6 hours, and involves collecting health information and completing questionnaires and assessments online.

To participate, contact Dr. Daniel Moreno De Luca or the PRISMA research staff at +1 780 492 4467 or prisma@ualberta.ca

CASA Mental Health | UNIVERSITY OF ALBERTA | Alberta Health Services

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