PRISMA

PRECISION MEDICINE IN AUTISM

We are delighted to be starting this new chapter in our new Canadian home!

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

We are delighted to welcome you to this next chapter of our PRISMA Group! In addition to the changing seasons, we are also embracing a dramatic change in scenery as our PRISMA Group has traded the ocean in Rhode Island for the Rocky Mountains in Alberta! Under the framework of my new position as the CASA Research Chair at the University of Alberta, Alberta Health Services, and CASA Mental Health in Edmonton, Canada, we are doubling down on efforts to develop cutting edge research, clinical care, and educational initiatives tailored towards the neurodevelopmental and Autism community, maintaining our strong focus on genomics and precision medicine. As a testament to our commitment to our shared vision, our entire group has moved here, either virtually or in person, bringing with us the spirit that we have created together. Stay tuned for the wider reach of our initiatives and exciting updates in our new surroundings. As always, thank you for your support!

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

OUR NEW HOME WELCOME TO EDMONTON, ALBERTA

Edmonton, Alberta, is a city brimming with vibrant culture, stunning landscapes, and a unique blend of urban and natural attractions. Nestled in the heart of Canada's western province, Edmonton offers an abundance of experiences that will leave visitors in awe. Here are some fun and interesting facts about this captivating city:

- Edmonton is renowned as the "Festival City" due to its abundance of exciting festivals throughout the year. From the famous Fringe Festival to the multicultural Heritage Festival, there's always something happening in Edmonton.
- West Edmonton Mall is not only the largest shopping mall in North America but was also the largest mall in the world until 2004. It features an indoor amusement park, water park, ice rink, and much more, making it a must-visit destination for shopping and entertainment.
- Edmonton earned the nickname "City of Champions" due to the multiple sports championships won by its professional teams, such as the Edmonton Oilers (NHL) and the Edmonton Eskimos (CFL).
- River Valley Parks: Edmonton boasts the largest urban park system in North America, offering over 7,400 hectares of green space. Visitors can enjoy activities like hiking, biking, and picnicking while immersing themselves in the city's stunning natural beauty.
- Edmonton is ideally located for viewing the mesmerizing Northern Lights (Aurora Borealis) during the winter months. The city's proximity to the magnetic North Pole enhances the chances of witnessing this breathtaking natural phenomenon.
- Sunshine City: Edmonton shines with an average of 2,299 hours of sunshine annually, with summer days stretching up to 17 bright hours.

The Edmonton region is home to more than 1.4 million people who make up a diverse and vibrant community. It's a place where we recognize and celebrate our differences, are ready to pitch in and help each other out, and welcome you with open arms.

These fun facts highlight the diverse and vibrant aspects of Edmonton, showcasing its love for festivals, natural beauty, unique attractions, and the city's distinct charm. Edmonton truly offers something for everyone, making it a fantastic destination to visit or call home.

TERRITORIAL ACKNOWLEDGEMENT

PRISMA respectfully acknowledges that Alberta is located on Treaty 4, 6, 7, and 8 Territory traditional lands - a traditional gathering place for diverse Indigenous peoples, including the Cree, Blackfoot, Métis, Dene, Stoney-Nakoda Sioux, Saulteaux, Siksika, the Pikuni, the Kainai, the Tsuut'ina, the Stoney-Nakoda First Nations and many others whose histories, languages and cultures continue to influence our vibrant community.

A JOURNEY *Through Time*

Research: Understanding psychiatric diagnoses through the lens of rare genetics.

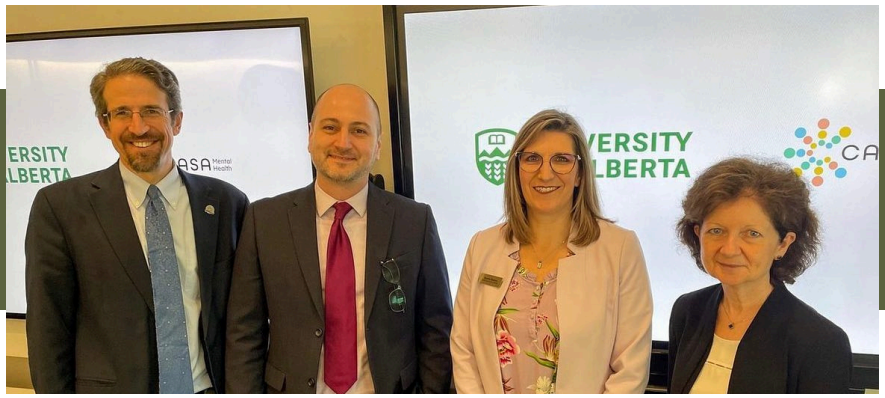
No two children with autism, or other neurodevelopmental conditions, are alike, despite sharing the same clinical categorical diagnosis. This clinical heterogeneity also extends to their underlying biology, with many individually rare genetic causes leading to these clinical presentations. By shifting the starting point of our studies from people who share a diagnosis like autism (a "phenotype-first" approach), to people who share a rare genetic variant that increases risk for those mental health conditions (a "genotype-first" approach), we will have a clearer link between biology and diagnoses, and understand why some people with a high risk genetic variant develop clinical symptoms while others don't, allowing us to identify opportunities for precision medicine interventions from early on.

For this purpose, we'll study people who have an extra (duplication) or missing (deletion) piece of their chromosome 17, at location 17q12. These genetic changes are collectively known as 17q12 copy number variants (CNVs), and are strongly associated with multiple mental health conditions, like autism spectrum disorders and schizophrenia, and medical conditions, like renal cysts and diabetes. Although the association of 17q12 CNVs with categorical psychiatric diagnosis has been established, we do not yet know its impact on dimensional neurobehavioral traits, how diverse medical comorbidities correlate with the expression of psychiatric phenotypes, how background common genetic variation may affect the expression of associated medical and behavioral phenotypes, or how these change over time.

As the inaugural CASA Research Chair in Child and Adolescent Mental Health, I'm eager to work together with our community to create state of the art programs that leverage cutting edge scientific advances to support research, clinical care, and education for the benefit of children in Alberta and beyond. Aligned with CASA's goal of providing services for all children, and simultaneously aware of the need of a focused approach to gain traction and develop infrastructure that can then be expanded, our work will initially center on one of the populations that has some of the most acute and significant mental health needs: children and adolescents with autism spectrum disorder, and those with intellectual and developmental challenges.

Clinical Care: Bringing genomics and precision medicine to children's mental health

Focusing on autism and neurodevelopmental conditions, studies from our PRISMA group and collaborators have been providing the evidence base and reasoning to develop targeted clinical services in precision medicine for children's mental health, where genomic information will be a key factor in the development of treatment recommendations when needed. As part of the CASA Research Chair, we will focus on developing unique clinical avenues to bridge the gap between professional recommendations around genetic testing and their clinical adoption. We'll do so through the creation of two new clinical services in a consultation model, working closely with referring clinicians - Psychiatric Genetic Counseling Service and Genomic Psychiatry Consultation Service.

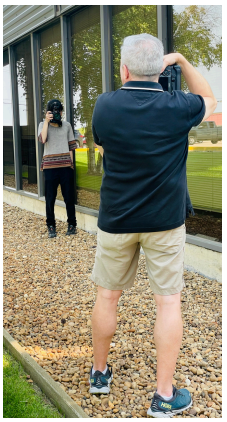


Education: Enhancing knowledge and clinical expertise around genomics and autism spectrum and other neurodevelopmental conditions

We will work closely with leadership at the Department of Psychiatry, the Division of Child and Adolescent Psychiatry, and the Child and Adolescent Psychiatry Training Program to develop and implement a curriculum that focuses on gaining knowledge and expertise in working with children with autism and other neurodevelopmental conditions. In addition to our work through the formal fellowship track above, we will also develop tailored educational interventions around autism spectrum and neurodevelopmental conditions, and genetics, for the community of practicing physicians and patients alike. Lastly, as part of the autism and intellectual developmental disabilities committee of the American Academy of Child and Adolescent Psychiatry, we have assembled a network of specialized training sites in autism spectrum disorders across North America, and provide career and networking opportunities for medical students, residents, and fellows who elect to follow this path.

Community: Celebrating our shared humanity

Through our community efforts, we will continue to ensure that the voices of the community we serve are heard loud and clear when defining research and clinical priorities, and that we have a close working relationship as we pursue our shared goals. We will achieve this in an engaging and exciting way by bringing science and art together and ensuring that our communications and media are easily accessible and clear. We are excited to bring the next chapter of our **"¿What's your Thing?"** campaign with Positive Exposure to Edmonton, aiming to recast the way that people with neurodevelopmental and genetic conditions are portrayed in general media and in medical education as we celebrate our shared humanity through photography and video exhibits that highlight the unique interests, strengths, and humanity of participants.



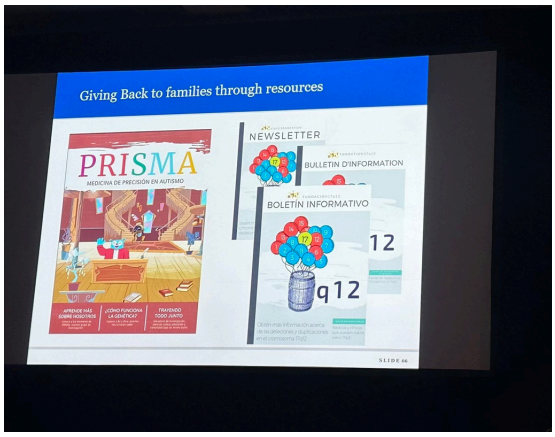
PRISMA AT HOME

"My first trip to Alberta was an extraordinary treat. I will hold onto memories of the great open highways, the underground maze of Rat's Nest Cave, the wild elk, black bear, and mountain goat sightings, the dramatic peaks and enchanting glacier lakes of Banff and Jasper Parks, and the unexpected hailstorm! We were well taken care of in Edmonton, a vibrant, trendy, yummy, and incredibly welcoming city. Thank you to everybody we met who made it so. We will be back soon" **Molly Goldman**

"I had such an incredible trip to Alberta! I was in complete awe of the raw, rugged beauty of the Rockies - the mountains, aquamarine lakes, glaciers, and the wildlife. I loved my time in Edmonton, what a great city! We were welcomed so warmly, and it was truly a pleasure to meet with so many people and to learn about the amazing work happening at the University of Alberta and in the community. I can't wait to visit again" **Carrie Best**

PRISMA AROUND THE GLOBE

We've been busy working with the International Society of Psychiatric Genetics since our last newsletter! We had the wonderful opportunity of going to the **World Congress of Psychiatric Genetics in Florence, Italy, to share our work on 15q13.3** deletions and galantamine, and then, Dr.Daniel Moreno De Luca was invited as the keynote closing speaker for the congress in Montréal! We had the amazing opportunity to boost collaborations and learn about cutting-edge research, all while enjoying these very different but unique venues!



In addition to traveling to Florence and Montréal for the World Congress of Psychiatric Genetics, we also had the opportunity to visit Toronto and New York for the 2022 and 2023 American Academy of Child and Adolescent Psychiatry. Here, we had the privilege of sharing our work on the implementation of autism genetic testing in an inpatient autism unit, and to showcase our "¿What's your Thing?" collaboration with Rick Guidotti and Positive Exposure in the framework of the Harris Forum, one of the most special events of the meeting. This also has allowed us to start bringing our two homes, the US and Canada, together under the autism umbrella as we foster collaborations between the American and Canadian Academies of Child and Adolescent Psychiatry. Plus New York and Toronto in the fall are quite a beautiful scene!

We met with the Latin American Genomics Consortium to advance psychiatric genetics research in Latin American populations and facilitate collaborations globally. This reflects PRISMA's commitment to the Latin American community, and we prioritize Spanish materials for better communication and accessibility. Learn more at **www.latinamericangenomicsconsortium.org**

Autism Track

The prevalence of autism spectrum disorder (ASD) and intellectual developmental disabilities (IDD) has been gradually rising along with the use of mental health services in this population due to higher psychiatric co-occurring conditions. However, access to appropriate and timely mental health care has been limited. While challenges in access to mental health care are multifactorial, one factor includes clinicians' lack of comfort and familiarity with treating individuals with ASD or IDD, impacting their self-perceived competency. This limited exposure during training can negatively impact provider competency and subsequently result in hesitation to treat this population, which may result in delay of care or incorrect treatment selection. To address the needs described above and train future child psychiatrists to develop deeper skills in assessing, understanding, and treating patients with ASD and IDD, we created an ASD/IDD track. We are excited to introduce our first graduate, Julia Katz.

"Lean in to learning experiences, be curious about your patients, their families and their environment and most importantly allow your patients to be your teachers"

Could you share a short bio of how your career has gotten you to where you are now?

I started volunteering with youth with neurodevelopmental disabilities when I was in high school and was immediately struck and captivated by their unique ways of making connections, as well as their strengths and challenges. As I learned more about autism during my undergraduate education, I decided to pursue a career in mental health and attend medical school to become a psychiatrist. Throughout every step of my training, my encounters with individuals with neurodevelopmental disorders (NDDs) remained incredibly rewarding and challenging. I was drawn to understanding the complexity of their lived experience and ways to support them and their families as they navigate through life. During my adult psychiatry residency, I've become interested in therapy and continued working on these skills during my child and adolescent fellowship with the hope to apply them during my practice with individuals with NDDs when I graduate.

What made you interested in the autism track and the child and adolescent psychiatry fellowship at Brown University?

Knowing I wanted to work with kids with ASD and NDDs, I was looking for a fellowship that would allow me to broaden my knowledge in the field and learn about the various factors that contribute to kids with NDDs to seek mental health care, as well as the various levels of care and interventions we have available to help support them. I was lucky to be accepted to the fellowship at Brown, which has robust services for kids with ASD including outpatient pharmacotherapy and therapy, partial hospital program, after school program, inpatient unit as well as several group homes and school placements. Being able to see such a large variety of presentations and treatment modalities allowed me to better understand the challenges that children with ASD and IDD and their families face and ways to support them and help them succeed in accomplishing their goals. The additional benefit of having so many services is the availability of many mental health providers starting from psychiatrists to psychologists and support staff that had years of experience working with this population. All had their unique knowledge base experience and approach and were passionate about imparting their knowledge and expertise.

How do you hope to leverage your growing expertise in autism moving forward?

I'm hoping to continue working with kids and adults with ASD and IDD and their families in a multidisciplinary approach that would allow to address the challenges this population faces in a more comprehensive manner that would consider environmental factors that could contribute to mental health struggles. In addition, I hope to continue learning and adding to the literature about both pharmacologic and other therapeutic interventions that are helpful and well-studied for this population. Finally, one aspect of working with this population that was evident throughout my training is the limited access to timely mental health care, and I hope to continue working on ways to make care more accessible both by thinking creatively and using technology, as well as focusing on educating and training more mental health providers on how to provide care for this population.

On the background of our diverse and multilingual patient population, could you tell us a bit about the several languages you speak?

I speak Hebrew, Russian and English mostly thanks to my family's background. One of the initial aspects that drew me to working with individuals with American Sign Language was learning about how they relate and connect with others and language in its different forms plays a role in that as well. I'm interested in learning other languages and thus far enjoyed learning ASL and practicing it as well as exploring other forms of communication with my ASD patients.

If you want to know more about our autism track within child and adolescent psychiatry fellowship at Brown University, as well as our brand new compact clinical fellowship in autism for child and adolescent psychiatrists here at the University of Alberta, check out our web at www.precisionmedicineinautism.org/education

ASK A DOCTOR

I HAVE HEARD ABOUT GENETIC TESTING FOR AUTISM, BUT IT SEEMS UNCLEAR WHAT THE BENEFITS ARE. HOW WOULD THAT INFORMATION BE USEFUL FOR MY CHILD AND THE DOCTORS? IS THIS A DIFFERENT WAY TO DIAGNOSE AUTISM?

This is an excellent question. We know that autism has a strong genetic component, and in fact, a genetic cause for autism can be detected in up to 40% of people; that percentage can be even higher if there are other accompanying diagnoses such as intellectual disability or seizures. There are many reasons why genetic testing is useful for autism, but before we discuss them, we should make to important points:

- Autism is a clinical diagnosis, meaning that it is given based on the observations by the doctors during an office visit, or with additional psychological testing, such as the Autism Diagnostic Observation Scale (ADOS). That means that genetic testing is not used to diagnose autism; it is recommended after the diagnosis of autism has already been made to uncover potential causes.
- The decision about carrying out genetic testing should be a joint process with your medical team, and the decision to proceed is entirely in the hands of each family.

There are multiple different genetic tests, and for autism, the ones recommended are chromosomal microarray testing and Fragile X testing. These two tests are considered the standard of care for autism spectrum disorders and are recommended by multiple medical professional societies, including the American Academy of Child and Adolescent Psychiatry, the American Academy of Pediatrics, and The American College of Medical Genetics and the American Society of Human Genetics jointly, with growing support for exome sequencies as a first-tier test.

Chromosomal microarray testing looks for missing or extra pieces of genetic material across the genome, called deletions and duplications, or also known collectively as copy number variants (CNVs); With that information in mind, here are a few examples of the potential benefits that genetic testing may bring:

- Finding an explanation and underlying cause for the autism in a given family and putting an end to the diagnostic odyssey.
- Identifying risk for other behavioral and medical conditions associated with a given genetic change, such as cardiac or renal abnormalities, which may in turn impact clinical management, including additional workup and medication choice.
- Obtaining genetic counseling and risk assessment for family planning.
- Having a clearer picture of areas of strengths and vulnerabilities based on information from other families with the same genetic abnormality and accessing specific medical resources.
- Connecting with support groups of other families with the same genetic variant.
- Being eligible for clinical trials targeting people with a specific genetic variant. All the examples above encompass what we now call precision medicine - the ability to use precise individualized information for tailored clinical management.

After highlighting the benefits of genetic testing above, it is also worth mentioning that some families elect not to have genetic testing. Some of the reasons they cite include feeling guilty about potentially having passed on a genetic variant to their child, feeling that there is no clinical use for this information and that this test would not change their child's (or their own) clinical management, religious reasons, ethical concerns, or privacy concerns. While many of these reasons are important, some of them are rooted in misinformation or misunderstanding of the process. We would recommend that the best way to move forward, whether you would like to move forward with genetic testing or hold off, is to have an open discussion with your doctor first to make a well-informed decision.

Remember, the decision of getting genetic testing is entirely up to you and your family and should be a joint process with your doctor. Hopefully, the information above can help you and your family decide on what is right for you!

We're happy to open up this section for all the questions you have about autism, genetics, genetic testing, and precision medicine. We have an outstanding network of clinicians available to answer them! Please send an email to prisma@ualberta.ca, and in the next edition, we will choose one question to answer.

Gene Genie Chronicles

22q11.2 Deletion Syndrome

Is a disorder caused when a small part of chromosome 22 is missing. The term ***22q11.2*** deletion syndrome covers terms once thought to be separate conditions including:

- **DIGEORGE SYNDROME**
- **VELOCARDIOFACIAL SYNDROME**
- **CONOTRUNCAL ANOMALY FACE SYNDROME**

THEY WERE ALL PART OF A SINGLE SYNDROME WITH MANY POSSIBLE SIGNS AND SYMPTOMS.

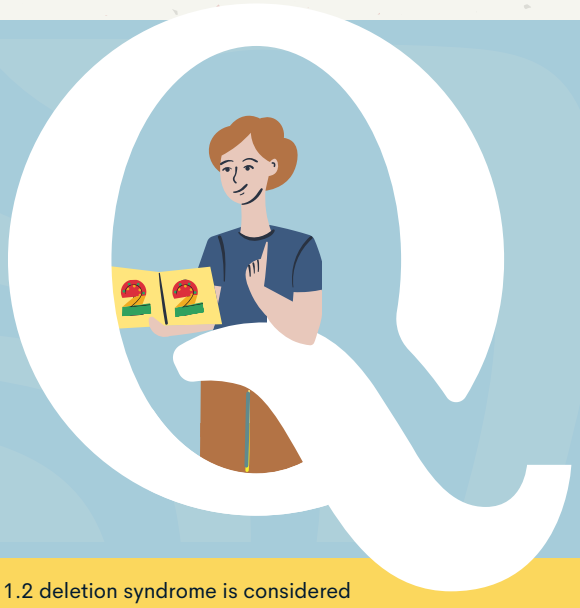
22q11.2 DELETION SYNDROME AFFECTS AN ESTIMATED:

1 *in* 4000 PEOPLE

SIGNS AND SYMPTOMS

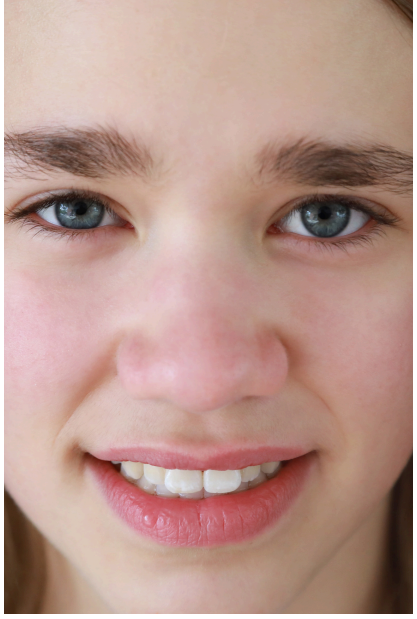
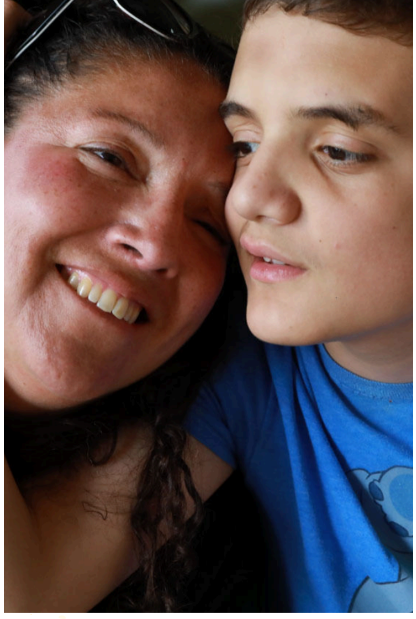
can vary in type and severity, depending on what body systems are affected and how severe the defects are. Some signs and symptoms may be apparent at birth, but others may not appear until later in infancy or early childhood.

- Heart Defects
- Frequent infections
- Certain facial features
- Difficulty Feeding
- Problems with the palate
- Delayed speech nasal-sounding speech
- Delayed grow
- Poor Muscle Tone
- Delayed development
- Learning delays or disabilities
- Mental Health conditions like autism and schizophrenia
- Breathing problems



Each person has two copies of chromosome 22, one inherited from each parent.

- The inheritance of 22q11.2 deletion syndrome is considered **autosomal dominant** because a deletion in one copy of chromosome 22 in each cell is sufficient to cause the condition. One copy of chromosome 22 is missing a segment that includes an estimated **30 to 40 genes.** Many of these genes haven't been clearly identified and aren't well understood.
- The deletion occurs most often as a random event during the formation of reproductive cells (eggs or sperm) or in early fetal development.
- Affected people typically have no history of the disorder in their family, though they can pass the condition to their children. In about 10 % of cases, a person with this condition inherits the deletion in chromosome 22 from a parent. In inherited cases, other family members may be affected as well.


What's your thing?

COMMUNITY EVENTS

Our team genuinely believes that by immersing ourselves in the community and forging deeper connections, we can nurture stronger communication, genuine care for one another, and a profound appreciation for the beautiful tapestry of diverse perspectives. This collective effort will undoubtedly contribute to improved overall health outcomes for each and every one of us. We can't wait to embark on this exciting journey together!

In collaboration with photographer Rick Guidotti and the team at Positive Exposure, we recently organized an inspiring exhibit at the Brown Medical School. The exhibit, titled "Celebrating our shared Humanity," aimed to shine a spotlight on the unique interests, strengths, and inherent humanity of local participants. It was a true celebration of diversity and inclusion.

The opening reception of the art exhibit was a heartwarming event that brought families together. They gathered to honor the momentous display and share in the joy of recognizing the beauty in each other. In addition to the physical exhibit, we also offered digital exhibits that provided further insights into the lives of the individuals featured in the show.

Building on the success of this initiative, we embarked on another exciting endeavor in partnership with Positive Exposure. As part of their "Faces Redefining the Art of Medical Education" (FRAME) video series, we conducted interviews with several young adults on the autism spectrum and their families. The aim was to educate healthcare professionals about mental health matters and reshape the portrayal of individuals with neurodevelopmental and genetic conditions in medical education. We believe that by promoting a more inclusive and empathetic approach, we can improve the overall well-being of our community.

We would like to extend our heartfelt gratitude to all of you who participated in these events and supported our mission. Your involvement has been instrumental in creating positive change and promoting understanding. Now, as we embark on a new adventure in Canada, specifically in Alberta, we are excited to connect with the local communities there.

As we settle into our new surroundings, we kindly invite you to share with us all the exciting activities happening in and around Edmonton. We are eager to participate and contribute to these events, as we firmly believe in the power of collaboration and community engagement.

Thank you once again for your unwavering support. Together, we can continue to make a difference and build a more inclusive and compassionate society.

***"If you happened to miss the event, don't worry! You can still be a part of it by visiting* www.precisionmedicineinautism.org/positiveexposure."**

ELSEWHERE IN THE GENOME

FRAGILE X

Karen is a mother of three children who have Fragile X syndrome, a rare genetic disorder that affects learning and development. She is also a researcher who recently completed her master's degree in science in pediatrics, focusing on the cognitive strategies that parents use to achieve successful parenting moments with their children.

Karen's eldest son, Noah, is 21 years old and has the full mutation of Fragile X. He has a passion for sports and a collection of jerseys that he proudly displays on his wall. Karen says that these jerseys are more than just clothing items, "They are a form of communication, and it's his way to engage with the community around him," she says.

Noah's love for sports has led him to join a hockey program in his hometown of Calgary, Alberta, where he has made a difference and found a sense of belonging. Karen says that Noah's involvement in hockey has improved his social skills, self-esteem and well-being.

"He feels like he belongs somewhere, that sense of belonging has led to unimaginable growth and development. This is similar to the strategies I identified in my thesis through the Physician Learning Program (PLP). I have been given a platform to translate these parenting strategies into tools of engagement to really identify those strengths and assets that are available with all of the champions around you, and really enhancing care while we wait for targeted treatments to come along," she says.

Karen's other two children, Sydney and Ty, also have Fragile X, but with different degrees of severity. Sydney, 17, has the full mutation, while Ty, 16, is mosaic, meaning he has a mix of cells with and without the genetic change. Karen says that each of her children has their own unique challenges and successes, and that she tries to focus on the positive aspects of their development.

She says that her family's journey with Fragile X has shaped their lives in many ways, and that she hopes to share her insights and experiences with other parents and professionals who work with rare diseases.

"Personally, it's engaging in the community that Noah and my other kids are involved with and really enhancing that ability to belong somewhere," she says.

Karen's research project was part of the Physician Learning Program, a collaboration between the University of Alberta and the Alberta Medical Association that aims to improve healthcare quality and outcomes. Her thesis was titled "Insights into successful parenting moments: Capturing expertise in Fragile X syndrome to achieve best possible health outcomes in rare disorders". She says that she plans to continue her work in this field and to advocate for more support and awareness for families affected by Fragile X.

Karen says that her family's journey with Fragile X began when Noah was three and a half years old and they received the diagnosis, which was unexpected and unfamiliar to them. She says that they had to deal with a lot of negative emotions and uncertainty about the future, but they also learned to appreciate their children for who they are and not what they are supposed to be.

"Over so many years it's helped me realize that it isn't all about creating a child who I'm assuming is going to be typical, but really identifying who these kids are and, you know, what makes them tick and really drawing upon that to see where things go," she says.

Karen says that one of the most important factors in their lives has been the role of community, which has provided them with support, opportunities and acceptance.

She says that they have met many people along the way who have helped them in different ways, from teachers who noticed Noah's potential and challenges, to coaches who welcomed him into their teams, to employers who offered him a job at the Saddledome, a sports arena in Calgary.

"They've given us space where he feels like he belongs and he's just become the person he wants to be, and he was offered a job at the Saddledome and it wasn't because a mom calls up and says "Hi, I have a kid with special needs that loves hockey, can you please give him a job? it was Calgary Sports

ELSEWHERE IN THE GENOME

and Entertainment who approached SuperHEROS hockey wanting to offer employment opportunities to players who love hockey and want to be a part of their team." It gave him a place to be someone that impacts other people and not just other people impacting him," she says.

Karen says that she is grateful for the community that has embraced her family and that she hopes to give back by sharing her story and her research with others who may benefit from it. She says that she wants to raise awareness and understanding about Fragile X and the potential that people with this condition have.

"When you look beyond the diagnosis it's amazing what happens!" she says.

Upon reflecting on a unique bond between her children, who all have Fragile X but with different levels of expression, Karen says that her children love, support and encourage each other, and that they have a sense of what each other needs and how to offer it. She says that she does not think that their family is unique in this regard, but that they are very close and supportive of each other.

As president of the Fragile X Foundation of Canada, Karen says that she wants to raise awareness and knowledge about Fragile X throughout Canada, and to foster collaboration and trust between researchers, clinicians and families. The family conferences she has hosted in Alberta, with impressive turnouts and positive impact, brought the community together and highlighted beneficial reciprocal relationships between these groups. Karen believes they can learn a lot from each other. She says that she hopes to change the narrative from focusing on the challenges and deficits of Fragile X to highlighting the strengths and possibilities of people with this condition.

Karen's research, highlights the expertise and the burden that parents develop in caring for their children with rare diseases, and that it is important for them to take care of themselves and to find champions who can support them.

"A champion is someone who's not expecting something in return... The champions we've had in our life don't expect anything in return and they're so significant in the lives of our kids, in my life and in my husband's lives, and in the relationship between my husband and us, that if it wasn't for them that we wouldn't be where we are because we can't do it alone and it's so important to be someone to inspire that meaningful and active participation in life because in the end, it's what everyone wants. They just want to feel like they belong somewhere,"

Karen's story is an inspiring example of how a mother's love, curiosity and resilience can lead to positive outcomes for her family and her community. She hopes that by sharing her experiences and her research, she can help other families living with Fragile X and other rare diseases to find their own strengths and champions. She also hopes that by raising awareness and understanding about Fragile X, she can contribute to the advancement of research and care for this condition.

Karen's children share what they would like people to know about Fragile X:

"You need to help people, like kids and doctors, help them like my mom does, and I do. You need to be happy and enjoy your life" **Noah (age 21)**

"I don't really focus on Fragile X. My arthritis is more important to me and is more about what I think about. But if I had to share, I would say to help people with Fragile X you need to know they learn differently. You need to listen and be patient...I would say don't be afraid to ask for help, get help, find what works for you, don't give up and believe in yourself" **Sydney (age 17)**

"When you have Fragile x and you are at school or in class learning and focus is hard but you have to have a good attitude in class " **Ty (age 16)**

FRAME

FACES REDEFINING THE ART OF MEDICAL EDUCATION

We have the privilege of hearing from Pedro and Julian's mom about their story as a family, what motivates them, and their participation in the FRAMES Film.

Can you share some reflections on your families' participation in making the FRAMES film Autism: The Life Behind the Word? The boys were really excited. Julián being himself, I don't think he understands that much about what is happening and what is the purpose. At the same time, being around people that are supportive and are doing something with a purpose, I think the kids can feel it. Not all things need to be understood but some things just need to be feeling good. I think Julián was comfortable. He looked happy doing it.

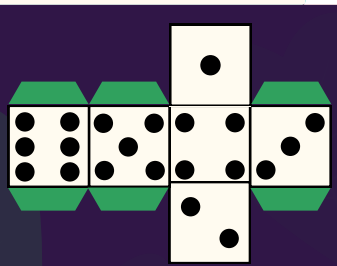
What was the experience like for Pedro as a sibling to someone with an ASD diagnosis? I think for Pedro to be part of it, because he is a key component of the whole process of being in Julián's life, was amazing. I also appreciate that because sometimes one of the things we forget about is the siblings and how it can be equally hard for the siblings. Making sure that Pedro felt appreciated by everybody and seeing him being so proud of saying how much he feels like he helps Julián (and how much he is there for him). There's a phrase that Pedro says that stuck to me forever. "When he calls my name, I'm sure to be there for him" and every time I see them together, I think "Wow, that's true". I appreciate them being together all the time but how much this means for Pedro is amazing.

Does Pedro have ways to connect with other siblings? I'd like to see in the future a way for Pedro to connect with other siblings that have brothers and sisters on the spectrum. We go to a lot of things with families that have special needs but nothing specific to siblings of kids on the spectrum.

What are your thoughts on additional ways we can support families? I think these videos are [intended] more for medical professionals. Making it available for the public in general and for communities to understand what it's like to be on the spectrum and what its like for family members...I think it can create awareness and create more kindness around different disabilities. The types of people I have in my mind are teachers and the school community. I'm [specifically] thinking about Pedro. Julian gets all the help he needs at school, and I make sure to advocate for his needs. [However], can you imagine what it's like to leave home after you see your brother being dysregulated and be expected to have the same type of accomplishments as a kid who left home in a nice, calm environment? I imagine that it is a little bit harder for those who experience these types of things when the leave home and they still need to accomplish and follow the same expectations as other kids. The amount of regulation that Pedro needs to go through throughout the day or even at night is a lot and I always try to tell Pedro that it is ok for him to be supportive of Julian and for him to have space for himself and to have a safe space to talk about his feeling and feeling overwhelmed without feeling guilty for feeling that way.

"YOU CAN CHECK OUT THE VIDEO AT WWW.VIMEO.COM/836557272/EC5A200D8A

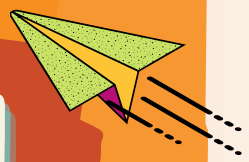
SNAKES AND GENOMES



DiNA needs your help in finding Bo! Roll the die to help her move forward in this path - she'll get there quicker by climbing up the genomes but watch out as she may slide back if she lands on a snake!

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 Dé 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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