

PRISMA

PRECISION MEDICINE IN AUTISM



EXCITING NEW FACES
JOIN OUR TEAM

WHAT'S YOUR THING?
EXPANDING IN ALBERTA

A NEW PRISMA STUDY:
AUTISM AND GENETIC
TESTING

IMPLEMENTATION
SCIENCE UPDATED

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ME!

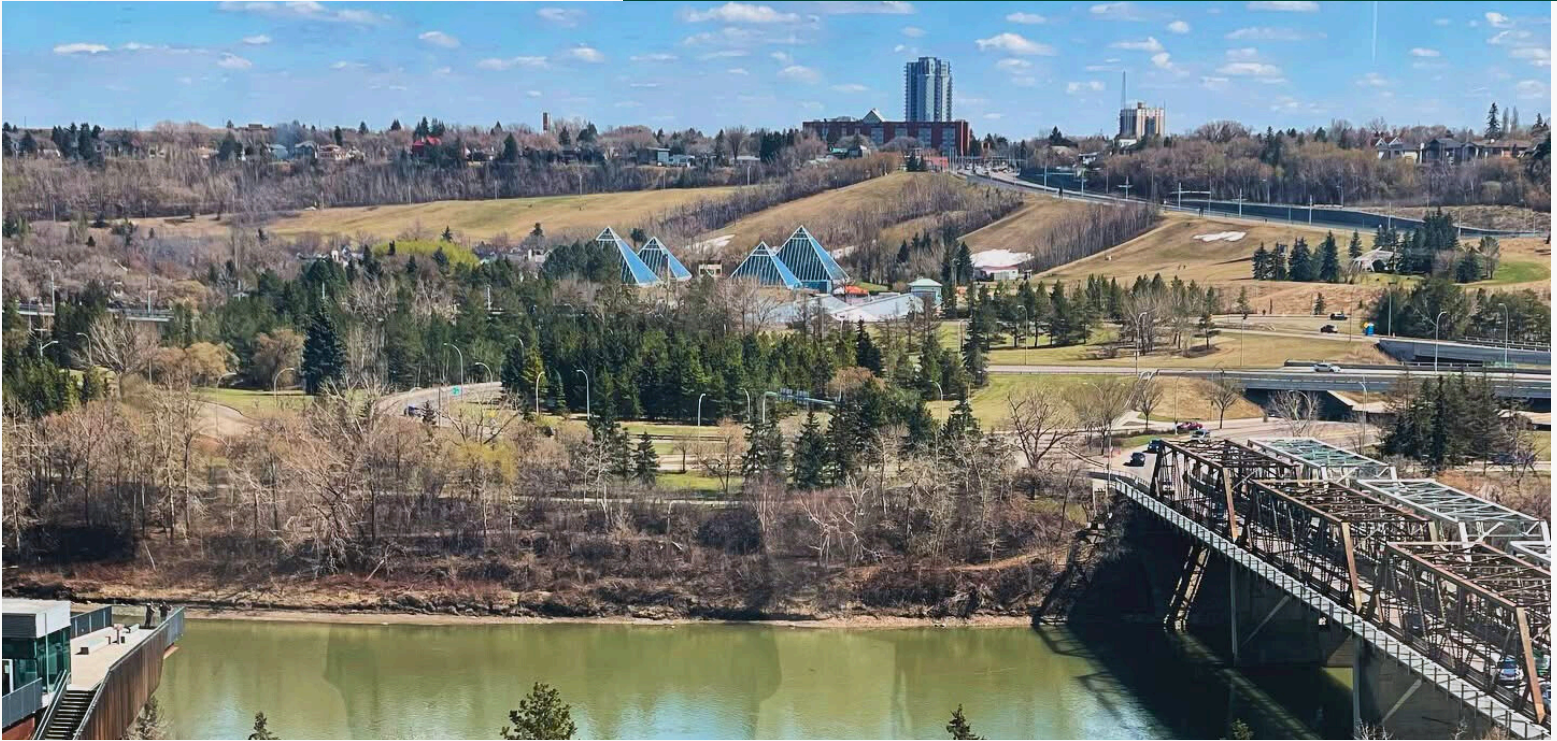


To get a copy online



Welcome

TO PRISMA RESEARCH GROUP



It has been a year of exciting changes and *new beginnings*, and I'm happy to share some highlights from the past months and reflect on the incredible progress we've made together as a group and community.

One of our key milestones this past year has been welcoming new team members: **Dr. Dener**, our postdoctoral fellow, and **Keely**, our research assistant, who have joined us and quickly become an integral part of **PRISMA** as we bid farewell to Jack, our research assistant for the past couple of years.

This growth also allowed us to launch a new research study focused on **Identifying the challenges and opportunities of implementing genetic testing after an autism diagnosis**, with the goal of making this key part of healthcare widely available as the standard of care.

At the same time, our **17q12 study** continues to move forward, with more families joining from around the world. Even more exciting, we can now offer it fully in **Spanish!** We were also delighted to host the inaugural **17q12 CNV Virtual Family Forum**, which brought together families, researchers, and clinicians to review the latest findings and set priorities for the future.

The past year also gave us a wonderful opportunity to connect with the autism community in Alberta. In Calgary, we hosted

the second photoshoot for the Canadian edition of our **"What's Your Thing?"** campaign and recently celebrated its opening, building on the success of our launch in Edmonton. We are especially grateful to **Autism Edmonton and The Sinneave Family Foundation** for partnering with us on these events, serve as a bridge between science and community, fostering meaningful new connections.

We have seen encouraging growth in our new clinical services: **Genomic Psychiatry and Psychiatric Genetic Counselling Consults**, which continue to address the needs of individuals with neurodevelopmental conditions through a genetics-informed approach.

None of these achievements would have been possible without the dedication of our team and the incredible support of families like yours. Thank you for being part of **PRISMA's** journey, we could not do this without you. **Here's to another year of discovery, collaboration, and connection!**

A handwritten signature in black ink, appearing to read "Daniel M.", written in a cursive style.

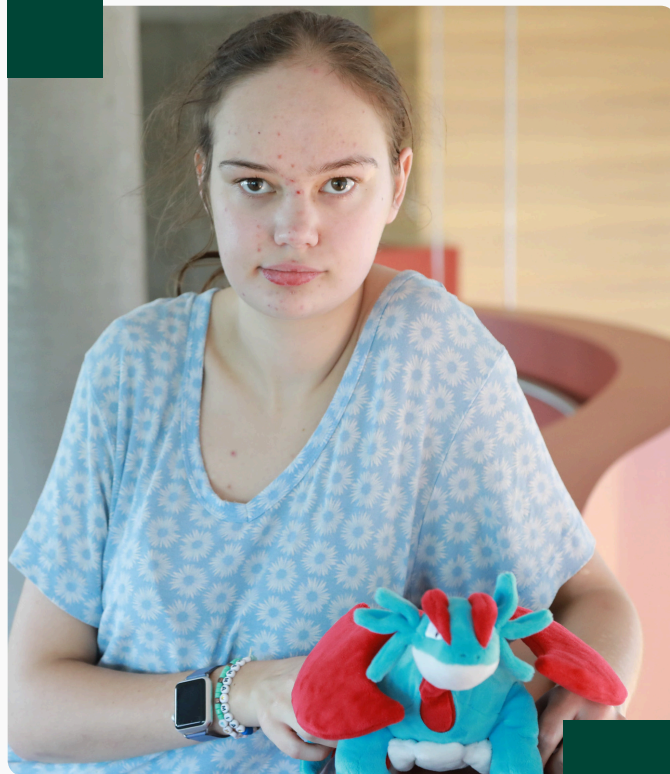
Daniel Moreno De Luca, MD MSc
Director & Principal Investigator PRISMA

CALGARY EDITION

What's your thing?

Last year, we were thrilled to expand our “*What's Your Thing?*” exhibit to Calgary, creating an opportunity to celebrate each person's unique passions and interests. This was made possible through the generous support of **CASA Mental Health, Autism Edmonton, and the Sinneave Family Foundation.**

The photoshoot was a special opportunity to connect through participants' stories and interests. We're grateful to our photographer and partner, **Rick Guidotti** of **Positive Exposure**, whose warmth and care made the experience truly memorable.



Hayley and Mohib's Stories

Among the 16 people in the project, **Hayley**, along with her dragon—likely her spirit animal—brought a unique and inspiring story to the project. Hayley shared her incredible experience as an actress in the film ***The Maiden*** which was featured at the **Venice Film Festival**. She offered fascinating insights into the movie and her role, showcasing her remarkable creativity and talent.

Among the many beautiful moments, **Mohib's session** was especially meaningful. His mom shared, “*It was a wonderful experience working with Rick. He knew how to get Mohib's attention and focus—something we've rarely seen in photos. It became a beautiful memory for our family.*” She added, “*Mohib would try to run off but kept coming back to Rick with a smile, ready for the next shot.*” For her, it was more than a photoshoot—it was a moment of pride. “*We're gifted and we shouldn't hide—in fact, we should celebrate the little things our kids do that we cherish.*”

Thank You to Our Community

We're deeply grateful to all the participants and families who shared their time and stories. Your contributions have brought so much meaning to this project and will continue to inspire others. The exhibit opened at the **Central Library in Calgary on September 12, 2025**, and was on display for a month. It then moved temporarily to the **University of Calgary Downtown Campus until January 2026**, and will remain available in the “*What's Your Thing?*” section on our website.



EDMONTON EDITION

What's your thing?

In the summer of 2024, the Art Gallery of St. Albert, **Autism Edmonton**, and **CASA Mental Health** came together for a special event: the opening of the Edmonton Edition of our exhibit "*What's Your Thing?*". Through this collaboration, we honoured and celebrated each person's uniqueness by highlighting their specific interests and passions. Partnering with **Positive Exposure**, we used photography to raise awareness of the value of diverse perspectives and their collective power to strengthen both our *shared humanity* and our community.

During the event, participants were able to see their photographs displayed as part of the exhibit. They also shared their personal experiences and stories with visitors, creating a space filled with connection and understanding. A standout moment of the evening occurred when **Catherine**, one of the exhibit's participants, shared a touching poem she had written, titled *Your Worth*. In her words: "*When you're feeling weak, when you struggle to feel strong, your worth belongs to you; in worthiness, you belong.*" Alongside her, her friend **Jessica** thanked the attendees for their support and encouraged them to take that appreciation into their daily lives, workplaces, and communities. She called for meaningful action *to create a more inclusive and accessible world for the Autistic community* while emphasizing the need for support and understanding within society.

Adding to these personal reflections, **Melinda Noyes from Autism Edmonton**, **Rick Guidotti from Positive Exposure**, and **Dr. Daniel Moreno De Luca, Director of PRISMA**, spoke to the audience about the story of this collaboration. They also shared how the project has now expanded to two different countries, anchored in the collective goal of **celebrating our shared humanity**.

To explore the online version of the exhibit, please visit: precisionmedicineinautism.org/positive-exposure

Thank you once again to all the visitors and supporters who helped create this incredible event!



PRISMA AROUND THE GLOBE

17q12 CNV Virtual Forum

On March 11, 2025, we hosted the *17q12 CNV Virtual Forum*, a global online event that brought together clinicians, researchers, and families to share knowledge and experiences related to *17q12 deletions and duplications*. The forum featured presentations by experts, community discussions, and meaningful opportunities for collaboration, helping to advance research and improve care for individuals with *17q12 CNVs*. We are deeply grateful to everyone who participated and helped make the event a success.



This time, from Canada, we were thrilled to reunite with the **17q12 community** and catch up on the latest developments. The forum, hosted by **PRISMA** at the **University of Alberta** in collaboration with the **17q12 Foundation**, brought together participants from around the world, spanning numerous time zones for a truly global conversation.

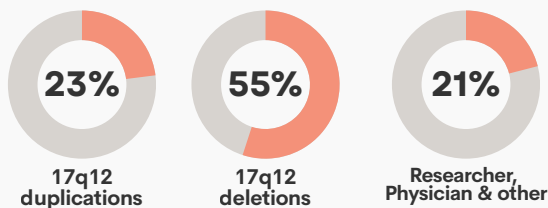


panel. **Christina, Craig, Sheralyn and Allaina** shared their personal experiences with *17q12 CNVs*. They spoke openly about the challenges, the uncertainty, and the strength it takes to navigate this path, along with the success stories and wisdom they've gained. Afterwards, the forum highlighted exciting new research, with presentations from scientists at **Yale University, Brown University, Baylor College of Medicine**, and the **PRISMA group at the University of Alberta**, all of whom presented developments in areas such as precision medicine, diagnostics, and immune studies, also inviting participation in ongoing research.

We are honoured to have hosted another successful conference and look forward to continued collaboration with this wonderful community.

Dr. Daniel Moreno De Luca, the Principal Investigator of the **PRISMA group**, officially opened the event, followed by **Allaina Wellman**, President of the *17q12 Foundation*, who shared updates about the community, information about the patient registry, and the many ways families can stay involved and find support. This was followed by a series of expert talks on neurology, psychiatry, kidney health, genetics, and behaviour. Doctors and researchers presented complex information in clear and thoughtful ways, and also made time to answer important questions from families.

Later in the day, the focus shifted to the community with a heartfelt family -



It was incredibly meaningful to reconnect, albeit virtually, since our last in-person gathering in 2019, held in **Providence, Rhode Island, USA**. At that meeting, we shared valuable face-to-face moments, deepened connections, and learned together through research, community, and clinical updates.

Global Participation at a Glance





17q12 Virtual Forum
**MISSED THE
LIVE EVENT?**

You can now watch all the
forum presentations
anytime and anywhere.
Check the link below!

bit.ly/17q12virtualforum



Through the prism: 17q12 Stories

From the moment you meet him, Alex shines with joy and energy. “He’s crazy about anything with wheels, especially dump trucks as big as he is! He loves dancing, bowling, swimming, and (dangerously for the wallet) sushi. We’re so proud and thankful to watch him grow and discover all the things he loves.”



“For us, there was a lot of empowerment in receiving a diagnosis.”

Alex is three years old, the younger son of Christina and Craig, who run the **Brentwood Chiropractic Clinic**, a wellness centre in Sherwood Park, Alberta. Along with his big brother Teddy (5), Alex fills their days with excitement, play, and curiosity. Christina and Craig kindly shared some of what they’ve experienced and learned as a family.

Looking back, Christina and Craig recall how their journey began. As they explain, “We first realized something was going on with Alex in utero. He had echogenic kidneys, so we did follow-up testing and ultimately received a diagnosis of ARPKD (*Autosomal Recessive Polycystic Kidney Disease*). We wanted to know if we were carriers, so we advocated for a genetic test and discovered that Alex had a 17q12 deletion, which was not something we had ever considered. It doesn’t change who he is—it’s just information about how we can best support him.”

However, one of their biggest challenges was the absence of follow-up care, something common for many families facing rare diagnoses. As they recall, “We were like, what do we do now? We went to Facebook groups, and through the 17q12 community, we connected with others. It was a great way to learn the spectrum of the diagnosis. There’s a lot of worry, but also a lot of hope. The community provided support, information, and helped us become better advocates for our son. We think there are real opportunities to collaborate across advocacy groups working on 17q12 symptoms.”

A turning point came when they connected with *the Robin Hood Association*. “They provided early monitoring and encouraged us to apply for **PALS (Playing and Learning at School)**. We knew Alex would likely face developmental challenges. When he started, he was nearly three and non-verbal. Now, he can speak in short sentences, ask for what he wants, advocate for himself, and follow instructions.”



Thanks to PALS, they saw an absolute explosion in Alex’s vocabulary, his receptiveness to speech, and his ability to express his needs. “The play between himself and his brother has changed exponentially for the better because he’s able to communicate. Alex wakes up every day calling for “Teddy Bear” which is his name for his brother, and they play beautifully together.”

Building on their clinic work and inspired by the *Nephrotic Syndrome Foundation* welcome packet, Cristina and Craig want to help create a more formal, proactive welcome for Canadian families receiving a 17q12 diagnosis.

“There’s still so much stigma against kids who are different, not just for the children, but for parents and siblings, too. It’s important to know you’re not alone and to have a proactive circle of care. When we talk to Teddy, we explain Alex’s kidneys are sick, using words he understands. We’d love to see more support on how to explain diagnoses to siblings and family, and more formal resources for people diagnosed later in life, so they know, *you’re part of this community now, and we’re here to help you.*”

Their advocacy expanded further when Christina and Craig participated as panellists and attendees at this year’s 17q12 Virtual Forum. “It gave us incredible insights into the disease, symptoms, and research. We connected with **Dr. Alexander**, who is studying magnesium exchange and its role in kidney function. Knowing more has helped us advocate, especially when daycare policies don’t account for medical potty-training challenges. This knowledge opens doors to inclusion through awareness.”

As they reflect on their journey, Christina and Craig remain eager to connect with others. “We always say that if anyone in the community wants to reach out, we’re here. We’re passionate about accessible health care, good treatment, diagnosis, and supporting families on this journey.”

We thank the Breckenridge family for opening their hearts and sharing their journey with us.

17q12

RESEARCH STUDY

A GENOMIC APPROACH TO
PRECISION MEDICINE IN AUTISM AND
NEURODEVELOPMENTAL CONDITIONS

Now enrolling
children and adults
who have had diagnostic
genetic testing that
showed a deletion or a
duplication in
chromosome 17q12.

We want to see

how genetic testing results could be used in the future to inform medication choices, behavioural interventions, educational interventions, and identification and treatment of other medical conditions that may impact mental health.



The study will take

approximately six hours and involves collecting health information and completing questionnaires and assessments online.



Who can enrol?

People with **deletions or duplications in chromosome 17q12** identified through diagnostic genetic testing who:

- Are **children or adults**
- **May or may not** have any neuropsychiatric conditions
- Speak **English or Spanish**
- Are passionate about advancing our understanding of 17q12 to enhance healthcare



To participate

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



**SCAN
ME!**



Learn more about 17q12 CNVs and get more information about our research study.



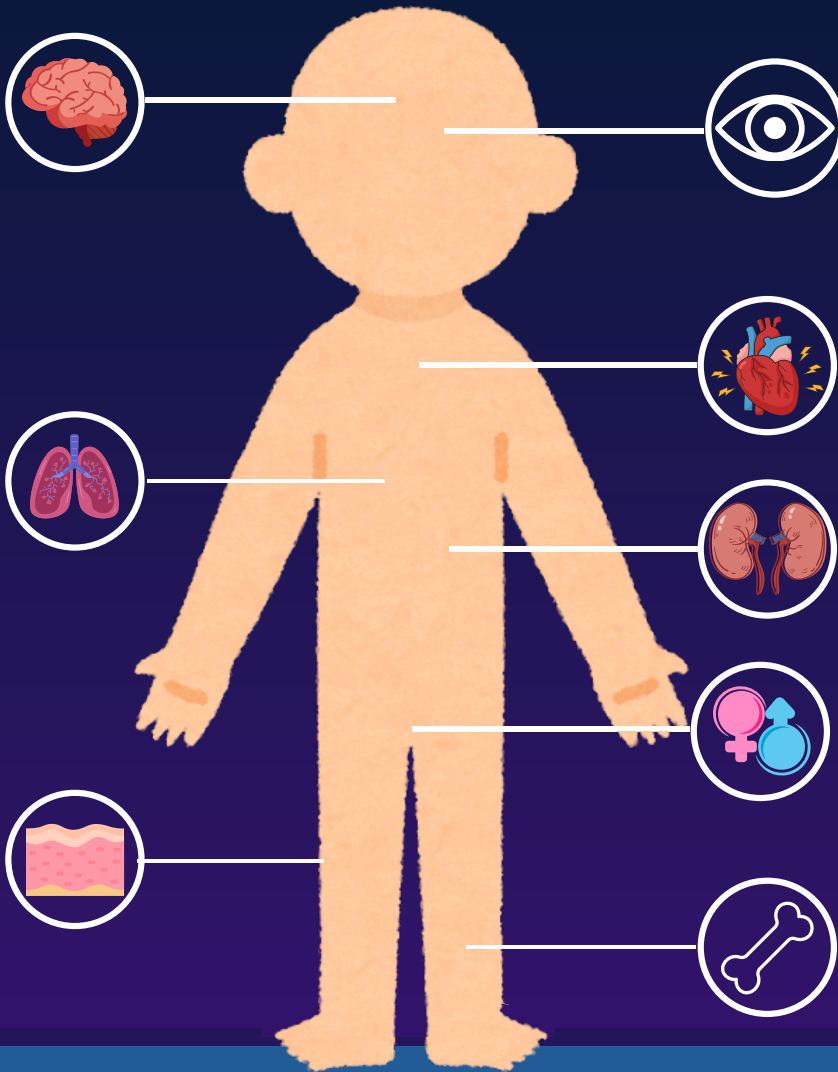


GENE GENIE CHRONICLES

PLEIOTROPISM

A pleiotropic gene influences multiple, seemingly unrelated traits in an organism. A single gene produces a product (such as a protein) that affects multiple body systems, tissues, or functions.

Pleiotropy is important because it helps us:



Explain Complex Symptoms

A single gene mutation can lead to symptoms across multiple body systems.

Inform Health Care

Pleiotropic effects show why some genetic diseases are hard to treat and why targeting one gene can cause side effects elsewhere.

Explain How Genes Work

Pleiotropy means one gene affects many traits, helping scientists link genes to different body functions.

Here are some examples of medical conditions caused by changes in pleiotropic genes



Gene: FBN1
Marfan Syndrome
Tall stature, long limbs, lens dislocation, aortic enlargement



Gene: PAH
Phenylketonuria (PKU)
Intellectual disability, lighter skin pigmentation, metabolic issues



Gene: NF1
Neurofibromatosis Type 1
Skin spots, nervous system tumors, learning disabilities



Gene: CFTR
Cystic Fibrosis
Lung infections, digestive problems, infertility



Gene: HBB
Sickle Cell Disease
Anemia, joint pain, malaria resistance



Gene: MECP2
Rett Syndrome
Neurodevelopmental regression, motor and language impairments



Have you noticed that when there's a power outage in your house—maybe during the summer—not only do the lights go out, but the air gets warmer and you can't heat food in the microwave?

A single element, electricity, affects the lights, the air temperature, and even your food! In the same way, there are genes in our DNA that have effects on many seemingly unrelated areas of the body, such as the brain, skin, and heart. When there's a problem in one of these genes, you might receive a diagnosis such as autism, skin spots, and heart malformations—all caused by the same underlying issue. This characteristic is called pleiotropism, and it's very important in genetics and medicine.



Exciting New Faces Join Our Team

PRISMA IS GROWING



Dener
Cardoso Melo
Postdoc Fellow



Keely
Vachon
Research Assistant

We're so excited to welcome two amazing new members to the PRISMA team! They bring unique experiences, fresh ideas, and a shared passion for understanding autism, which will have a significant impact on our work.

Meet Dener

Originally from **Brazil**, Dener trained as a speech-language pathologist and audiologist at the *State University of Health Sciences of Alagoas (UNCISAL)*. During an exchange year at the *University of Arizona*, he took a course on autism that immediately fascinated him and completely changed his life. It was through this course that he discovered he was Autistic and received a late diagnosis, which deepened his interest in psychiatry. Dener then moved to the Netherlands, where he earned a *Master's degree and a PhD in Psychiatry at the University of Groningen*.

He made the leap to Canada to join our team and help us explore the diverse experiences of people on the autism spectrum. When he's not working, Dener loves discovering new music and diving into linguistics to find connections between different languages. After years of grey, rainy weather in the Netherlands, he has been pleasantly surprised by how green and sunny Edmonton is. His favourite spot so far is the **river valley**—offering the perfect mix of nature and city in every season.

Keely Joins the Team

Joining Dener is **Keely**, who also calls Edmonton home. She completed a *Bachelor of Science in Psychology at MacEwan University*, where she was a student-athlete and played for the *MacEwan Griffins Women's Hockey Team* from 2017 to 2021. She went on to earn a *Master's in Pediatric Medical Sciences at the University of Alberta*, focusing on neurodevelopmental disorders and global developmental delay. Now, as the newest member of our team, Keely is excited to contribute to our research.

In her free time, Keely spends much of her time at the hockey rink, playing competitively in Edmonton and assisting as a coach for the *Sherwood Park U18AA Fury*. She also loves the variety of outdoor activities Alberta offers. In the spring and summer, she frequently visits the Rocky Mountains to explore the fantastic trails in the national and provincial parks. Her favourite spot in Edmonton is the **Strathcona Farmer's Market**, where creative small businesses offer delicious and unique goods. For Keely, it's the perfect way to support local businesses while catching up with friends.

*With Dener and Keely on board, we're excited about the **new perspectives and energy** they bring to PRISMA. Their **diverse backgrounds** and shared enthusiasm for understanding autism will help shape the **future of our research**. We look forward to seeing the contributions they'll make and can't wait for you to get to know them better!*

Where Psychiatry Meets Genetics



PRISMA AT HOME

Genomic Psychiatry and Genetic Counseling Services

As part of the Precision Medicine in Autism initiative, our clinical services focus on genomics to better support individuals with autism and other neurodevelopmental conditions.

Autism spectrum & other neurodevelopmental conditions have a strong genetic basis, and we can identify a genetic cause for these diagnoses in up to 1 out of every 3 people. Because of this, genetic counselling and testing is recommended as the standard of care after receiving one of these diagnoses. However, it only happens in a small proportion of patients, and when a genetic result is identified, there can be challenges with knowing how to use these results to enhance healthcare. We created two clinical services to address these challenges, and are excited to bring this expertise to clinical care in our community.



- **PSYCHIATRIC GENETIC COUNSELLING CONSULTATION SERVICE**

Here, we see kids or adults with a diagnosis of autism or other neurodevelopmental conditions who have incomplete, outdated (older than five years), or no prior genetic testing to provide genetic counselling and discuss professional recommendations, carry out genetic testing for people who agree to move forward with this, and deliver the results.

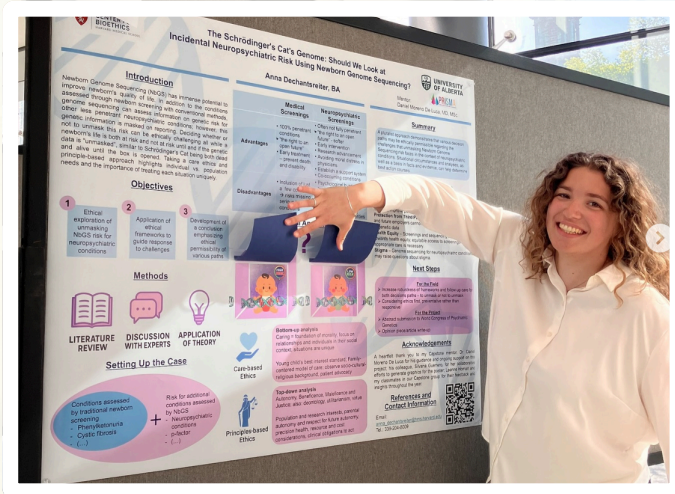


- **GENOMIC PSYCHIATRY CONSULTATION SERVICE**

Through this service, we see kids or adults with autism or other neurodevelopmental or psychiatric conditions who already had genetic testing and in whom a pathogenic (positive, or abnormal) genetic result was identified, to provide psychiatric management recommendations based on those genetic results. In doing so, we help families and clinicians understand what the findings mean in practical terms and offer guidance that can shape treatment, long-term care, and support.

Advancing Psychiatry through Education

At PRISMA, we believe that education is at the heart of everything we do. Over time, we've created opportunities for learning through the creation and delivery of genetics courses, dedicated clinical fellowship & subspecialty tracks focused on neurodevelopment and autism, and leadership in educational efforts as part of national and international organizations. This time, we're especially happy to share two milestones that reflect how our group is growing, both in supporting future professionals and in connecting with our community.

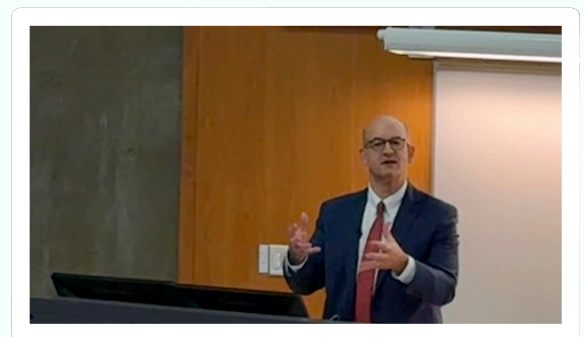


Anna Dechantsreiter - MSc in Bioethics from Harvard Medical School

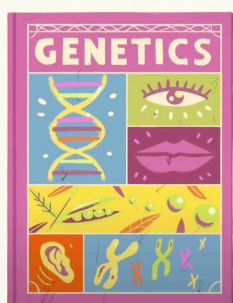
We are proud to highlight the work of **Anna Dechantsreiter, MSc** in Bioethics from Harvard Medical School, who carried out her research under the mentorship of **Dr. Moreno De Luca**. Her project explored the ethical challenges of genome sequencing as a tool for newborn screening, and how to deal with genomic results that may increase chances for neuropsychiatric conditions. With interests in global health ethics, health and human rights, and genomic medicine, her work illustrates how bioethics continues to address pressing questions at the intersection of science and society.

CASA Research Chair Ground Rounds - Dr. Matthew Siegel

As part of our **CASA Research Ground Rounds** series, we had the pleasure of welcoming **Dr. Matthew Siegel**, who spoke on "**Intensive Assessment & Treatment of Challenging Behavior in Youth with Autism and Intellectual Disability.**" His talk offered valuable insights that focused on the importance of creating strategies that truly meet individuals on the spectrum where they are, providing the right level of care to best support their needs.



Implementation of psychiatric genetics educational recommendations



In our leadership role in the Education Committee of the International Society of Psychiatric Genetics, and in close partnership with the NNCI, co-lead by **Dr. David Ross**, we are working on the development of educational modules in rare genetics, pharmacogenetics, common genetics, epigenetic, and gene-environment interactions. This initiative aims to strengthen the training of clinicians in psychiatry and beyond by bridging genetics and healthcare, providing tools to make more personalized and informed healthcare interventions. It reflects PRISMA's ongoing commitment to bring the latest advances in science directly into medical education, and healthcare delivery.

Supporting Clinicians in Genetic Testing:

A New *Interactive* Resource



Our previous work showed that only a small minority of patients on the autism spectrum or with other neurodevelopmental conditions received genetic testing, despite this being the standard of care and recommended by multiple medical professional societies.


Those studies also let us know that the majority of patients with these diagnoses and their families would be interested in moving forward with genetic testing if it were available to them. These findings pointed us towards a key strategy to ensure that genetic testing is appropriately available, supporting the physicians who care for them.

To achieve this goal, we created an **Interactive Genetic Testing Handbook**, a comprehensive step-by-step guide designed specifically for clinical teams. Inside, clinicians can find ways to start conversations about genetic testing with their patients, explainer videos covering key concepts, and even support on how to place an order in their electronic health record, along with referral forms for our clinical services.


We also wanted to make sure physicians felt comfortable delivering the results of such tests, and receive extra help in this process when needed. To support this, we created a template for the delivery of negative (normal) results, and a streamlined referral process to our **Psychiatric Genetic Counselling Consult Service**, so that we can help deliver positive (abnormal) results when needed.

These resources help ensure clinicians and caregivers alike can fully understand the process and make informed decisions at every stage.

Meet the Team

 **Dr. Daniel Moreno De Luca** - Director & Principal Investigator, Adult-Child Psychiatry

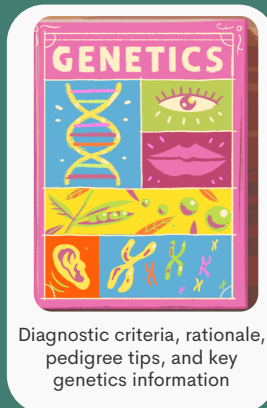
 **Julia Heaton** - Clinical Genetic Counsellor

 **Molly Goldman** - Research Genetic Counsellor

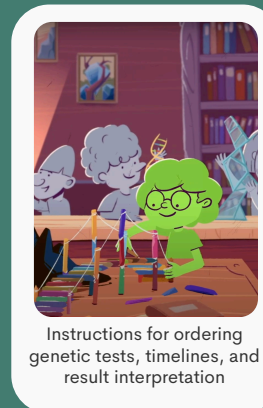


What's Inside the Handbook?

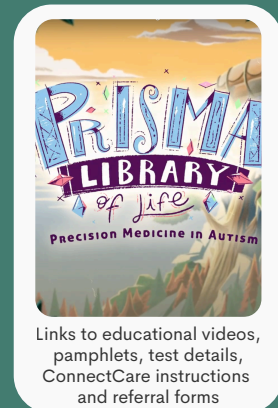
Our handbook is more than just a document—it's a dynamic resource packed with essential tools and information, including:



Diagnostic criteria, rationale, pedigree tips, and key genetics information



Instructions for ordering genetic tests, timelines, and result interpretation



Links to educational videos, pamphlets, test details, ConnectCare instructions and referral forms

We hope this comprehensive guide supports clinicians in delivering effective and compassionate care. Our team can also assist with test ordering and provide post-test counselling through the *Psychiatric Genetic Counselling Consultation Service*.

Explore and Use the Resources

We encourage all clinicians to explore the handbook and use its tools to support high-quality, patient-centred care. Together, we can make genetic testing more accessible, understandable, and impactful for every individual and family.

How to Use This Resource

Scan the QR code with your camera or visit www.precisionmedicineinautism.org/asd-genetic-testing-qi-project to access the material!





Meet our friend Lucas

Lucas is a 7-year-old boy on the autism spectrum. Since early childhood, his parents noticed he had trouble falling asleep and often woke up several times during the night. These restless nights left him irritable during the day, with more repetitive behaviours and difficulty paying attention in school.

His paediatrician asked the family to keep a sleep diary and later referred him to a specialist. In the meantime, he ordered genetic testing in light of his autism diagnosis, and found that Lucas had Smith-Magenis syndrome.

This result alerted him to the abnormal sleep pattern that many people with this genetic condition have, and allowed him to start melatonin in the evening to help with sleep consolidation, and a beta blocker medication in the morning to regulate the production of melatonin throughout the day. He also identified that his ferritin was low, indicating low iron, which is often a cause of restless legs syndrome and sleep problems; he provided oral iron supplements to correct this. To complement these interventions, the care team started a structured bedtime routine, and adjusted his sleep environment to be dark and cool.

Within a few weeks, Lucas was falling asleep faster, waking less often, and showing better focus and behaviour during the day. For his parents, the combination of medical evaluation and genetic insight provided both answers and help.

BETTER SLEEP TIPS



SLEEP ENVIRONMENT

Keeping the bedroom calm: dark, quiet, and cool helps with restful sleep.

BEDTIME ROUTINE

Predictable, short (20–30 minutes), with relaxing activities like reading or soft music.

SLEEP/WAKE SCHEDULE

Maintain consistent sleep and wake times across weekdays and weekends.

EXERCISE

Engage in exercise during the day, but avoid vigorous activity near bedtime.

AVOID CAFFEINE

Limit intake, especially in the afternoon and evening; remember it is present in coffee, tea, chocolate, and sodas.

NAPS

Appropriate for preschoolers, but in older individuals late naps may interfere with nighttime sleep.



Ask a Doctor

Is there a genetic link between autism and sleep problems, and how can medicine help?

Sleep problems are among the most common challenges in autism. According to the article “Sleep problems in autism, explained” from Spectrum News, between 44 and 83 percent of individuals on the spectrum experience difficulties such as insomnia, frequent awakenings, or sleep apnea. In contrast, only about 10 to 16 percent of children in the general population have similar problems. The same article reports that people with autism take, on average, about 11 minutes longer to fall asleep and spend around 15 percent of their total sleep in REM — the stage that supports memory and learning. Neurotypical individuals, in comparison, spend closer to 23 percent of their nightly rest in REM. This helps explain why sleep in autism often feels less restorative. Poor sleep has consequences beyond nighttime. As Spectrum notes, insufficient or disrupted sleep can increase repetitive behaviours, make social interactions harder, and lower performance on cognitive tasks. Families often notice that when sleep improves, so does daytime behaviour and attention.

When doctors evaluate sleep, we start with interviews and sleep diaries. We may also check for medical conditions such as sleep apnea or restless legs syndrome, order blood tests, or consider genetic testing. Genetic factors can play an important role, since certain genes that regulate melatonin — or syndromes like Smith-Magenis — are directly linked to abnormal sleep cycles. As a reminder, up to **one of every three people on the autism spectrum can have a genetic cause for their autism**, and many of these individually rare but collectively common causes, like the ones above, can be associated with sleep problems as well. Treatment can range from simple changes to more specialized care. A predictable bedtime routine, a quiet and cool bedroom, and regular sleep and wake times are usually the first steps. If problems continue, melatonin or other medical treatments may be recommended.

What is encouraging is that ongoing research, including genetic studies, is opening the door to precision medicine approaches. By tailoring care to each individual’s biology, we hope to help people with autism achieve more restorative sleep, and better days that follow.

Have a question about autism, genetics, or precision medicine? Send us an email at prisma@ualberta.ca. In our next edition, we’ll select one to answer!

NEW STUDY

Help Us Shape the Future of Genetic Testing for Autism

Bringing precision medicine to psychiatry starts with understanding what makes genetic testing work—or not work—in real-world settings. We're especially interested in identifying the barriers that make it harder for **genetic testing to be used in the autism community**.

That's why we launched our study called **Identifying Challenges and Opportunities for Implementing Standard of Care Testing for Autism & Neurodevelopmental Conditions**. We are inviting *autistic individuals, their families, and physicians* who provide care for autistic individuals to share their experiences and perspectives on genetic testing for autism. At this time, we are only enrolling individuals who **live or work in Alberta**.



WHAT'S involved?

Taking part in this study will take about two hours and will include:



- **An online survey (20–30 minutes)**. You'll be asked about your experiences and views on genetic testing for autism.
- **A one-on-one interview (up to 60 minutes)**. Some participants may be invited to share more detailed thoughts and experiences in a follow-up conversation.

WHY participate?

Your input can help us understand how to make **genetic testing more accessible and useful for autistic people**. By identifying the *challenges and opportunities*, we aim to bridge the gap between what's recommended by professionals and what's actually happening in clinical practice. This is an important step toward creating more personalized care and bringing us closer to precision medicine.

HOW does it work?

If you're interested, **please contact us to set up a call**. During the call, we'll explain the study, answer your questions, and help you decide whether you'd like to participate. If you choose to join, you'll complete a consent form to **enrol in the study**. If you're an *autistic adult or a parent/guardian enrolling on behalf of your child or family member*, we'll also ask you to sign a medical release form. This will allow us to access medical records to confirm an autism diagnosis.

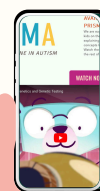
Languages



You can use whatever language works best for you.

Want to get involved?

SCAN ME !



Or contact **Dr. Daniel Moreno De Luca** or the PRISMA research team at prisma@ualberta.ca or +1 780 492 4467