PRECISION MEDICINE IN AUTISM - RESEARCH PROGRAM



PRISMA GROUP

Community Booklet

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PRECISION MEDICINE IN AUTISM

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PRECISION MEDICINE IN AUTISM - RESEARCH PROGRAM

WELCOME TO OUR AUTISM & GENETICS COMMUNITY BOOKLET!

We're glad you're here! At the Precision Medicine in Autism (PRISMA) Research Group, we want to make information about autism and genetics easier to find, understand, and talk about. Whether you're autistic, supporting someone you care about, or simply curious to learn more, this booklet is for you.

Inside, you'll find educational materials, practical resources, answers to common questions, and ways to connect with our community. You don't need a science background to get started. Explore what interests you, take your time, and come back whenever a new question comes up.

We hope these pages help you feel more informed and comfortable starting conversations. The information provided is general and does not replace personalized medical advice. If you have questions about your health, genetic testing, or care, a qualified healthcare professional can help you explore your options.

Your privacy matters. Genetic information is personal, and understanding how it may be used, stored, and protected is an important part of making informed decisions about testing. You can discuss these questions with your healthcare team before deciding what feels right for you or your child.

Everyone brings different experiences, questions, and perspectives. We believe that listening with respect, recognizing diverse experiences, and learning from one another are essential to building a welcoming and supportive community.

Above all, we hope you find something here that is useful to you or someone you care about. Your questions and experiences matter, and we're always happy to connect. If you have questions, would like to request materials, or simply want to learn more, reach out to us at **prisma@ualberta.ca**.

Why PRISMA?

Connecting genetics, research, and diverse perspectives to better understand autism

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 colours, 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!**

Meet the PRISMA Team

The people behind PRISMA's research, care, and community connections

As a team, we bring together different backgrounds, experiences, and languages to support our work in autism, genetics, research, education, and community engagement. In the photo below, you can meet our team members, discover the languages they speak, and learn more about their roles at PRISMA.



We also bring science to life through Bo and DiNA, the characters from our PRISMA Library of Life. Bo loves fixing things and collecting old clocks, while Dina loves dinosaurs and discovering new ideas through books. Together, they help make genetics and genetic testing easier to explore. Their fun, multilingual resources connect science with community and encourage children, adults, and families to talk with clinicians and take an active role in their healthcare.



What We Focus On

Connecting research, clinical care, education, and community engagement

Now that you've met the PRISMA team, we'd love to introduce you to the different areas of our work and share some of the initiatives we have developed together with the community. Curious about the science? Looking for clinical or educational resources? Interested in discovering inspiring stories? Keep reading to learn more about what we do!

Research

Our studies aim to **understand how different rare genetic variants may lead to psychiatric and medical symptoms**, with the goal of identifying opportunities to improve health-care delivery. We also focus on understanding the barriers and opportunities around standard of care genetic testing after a neurodevelopmental diagnosis like autism is made.

Education

We're engaged in various initiatives, from crafting genetics courses tailored for child, adolescent, and adult psychiatry residents to developing resources supporting mainstreaming of genetics resources across diverse clinical specialties. Our goal is to **empower future professionals with the latest knowledge and skills to make a meaningful impact in the field.**

Clinical Care

The clinical arm of the Precision Medicine in Autism initiative includes services specifically tailored around genomics and psychiatry to ensure that individuals with diagnoses of autism spectrum or other neurodevelopmental conditions are **offered recommended genetic testing,** and that those for whom a genetic cause is **found receive care that takes into account this important information**.





Community

At our core, **we thrive on our connections with patients, families, and individuals in the autism community**. We prioritize and celebrate these relationships through family conferences, workshops, social media engagement, community collaborations, public art projects and exhibits, and educational materials. Our commitment lies in nurturing these bonds and fostering a supportive and inclusive community.

For more information, visit our website www.precisionmedicineinautism.org or send us an email to prisma@ualberta.ca

PRECISION MEDICINE IN AUTISM - RESEARCH PROGRAM

Understanding AUTISM & GENTICS



ABOUT THIS SECTION

Explore accessible information about autism, genetics, and genetic testing to better understand these topics, answer common questions, and support informed conversations.

After an Autism Diagnosis

A friendly guide for autistic people and their families

A clinical diagnosis of autism can bring many feelings and questions. This guide offers ideas for Autistic people and their families to discuss with healthcare providers and explore support. You can use the steps that feel helpful and go at your own pace. Services and referral processes vary by location, and support can begin while you are waiting for additional assessments or appointments.

1. Review the assessment and diagnosis

Talk with the professional who made the diagnosis. Ask them to explain the findings, answer your questions, and share a copy of the report. The assessment may include information about the person's development, communication, health, strengths, interests, and support needs.





2. Share what you notice in everyday life

Your observations can help the healthcare team understand what matters to the person. You may want to note changes or questions about communication, routines, sleep, eating, sensory experiences, transitions, health, or daily activities. Bring relevant reports or previous assessments to appointments.

3. Discuss health and other assessment needs

Talk with the healthcare provider about any physical, emotional, developmental, or social concerns. Depending on the person's needs, they may suggest additional assessments, such as hearing, vision, or speech and language evaluations, or referral to another provider. If there is a sudden loss of skills or an urgent health concern, contact a healthcare provider promptly.





4. Explore support while referrals are underway

Support does not have to wait until every assessment is complete. Ask about options that may help with communication, development, daily life, learning, school, work, health, or community participation. Depending on where you live, some services may allow families or individuals to contact them directly.

5. Talk with your doctor about genetic testing

Several medical societies recommend genetic testing after a clinical diagnosis of autism, whether or not other conditions are present. Testing is a personal choice: Autistic adults decide for themselves, and families can discuss testing for a child with their doctor. Clinicians do not need to specialize in genetics to start these discussions. This booklet can help you begin the conversation.





6. Make space for family and personal wellbeing

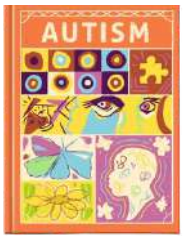
Ask who will coordinate care, which referrals have been made, and when to check in again. The people involved may include healthcare providers, therapists, educators, workplace supports, or community services, depending on the person's needs and preferences. Families and Autistic people can also ask what support is available for their own wellbeing.

THE STORY BEHIND THE DIAGNOSIS

Understanding autism through genetics, care, and connection



START WITH THE KEY POINT



- **Autism is diagnosed clinically**
- **Genetic testing does not diagnose autism**
- **Genetic testing should be part of the clinical pathway after a clinical diagnosis of autism.**

STANDARD OF CARE: Genetic testing is recommended as part of autism care

WHAT CAN GENETICS TELL US?

Genetics can help provide answers

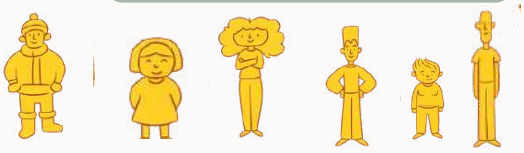


UP TO 30% OF AUTISTIC INDIVIDUALS MAY HAVE AN UNDERLYING PATHOGENIC FINDING

That information can help explain the diagnosis and support more personalized care

THE GAP IN ALBERTA

Many individuals still miss out



Only 10% of Albertans with autism have ever received standard of care genetic testing

WHY COULD GENETIC TESTING MATTER?

- Find an underlying explanation
- Connect families to resources and community
- Guide medical care and monitoring
- Identify associated health risks
- Support informed family planning and future decisions

HOW TO START THE CONVERSATION

A few simple steps can open the door

Talk to your:

- Pediatrician
- Family doctor
- Psychiatrist
- Developmental specialist
- Genetic counsellor
- Another clinician involved in autim care



You can ask:

- Should genetic testing be part of our care plan?
- Which tests are recommended?
- Would a genetics referral or genetic counselling be helpful?
- Genetic testing is a choice, but families should know it is part of recommended care
- Can you order this test for me directly?

THERE'S ALWAYS MORE TO DISCOVER



Explore autism, genetics, and genetic testing with Bo, DiNA, and their friends in the PRISMA Library of Life. Watch the video and continue exploring.
www.youtube.com/watch?v=iXvd_GEdNCE

Genetic Testing

How different genetic tests explore the genome



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.

ASD/ID Panel

This test takes a closer look at a large group of genes known to be linked to autism and intellectual disability, reading through their "words" to look for any changes in spelling. It uses samples from the child and their parents together, which helps us understand whether a change is new or inherited. By focusing on many relevant genes at once, this test can identify small changes that may affect how genes function—some of which may help explain a person's clinical symptoms, while others may not have an impact.

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!

Available English and Spanish.

Check the complete video: www.youtube.com/watch?v=iXvd_GEdNCE

SCAN ME!

Frequently Asked Questions

Clear, approachable answers to common questions about genetics and genetic testing in autism

Genetics and Autism

What is precision medicine?
Precision medicine focuses on using our unique genetic and biological characteristics so that healthcare can be personalized based on that information, helping tailor interventions, supports, and monitoring for those who need it.

Why is genetics relevant to autism?
Genetic information can help explain why autism occurs, especially when other medical or developmental conditions are present. It can guide care, support early identification of needs, and inform family planning.

Is autism genetic? Will my child be autistic if I am?
Autism is strongly influenced by genetics. In fact, if we bring together the many individually rare genetic causes of autism, they can collectively explain up to 30% of cases. In addition, genetic does not mean inherited, and many times, these genetic changes are not present in the biological parents but happen spontaneously, which we call de novo.

Do environmental factors contribute to autism?
Research suggests that some environmental factors (for instance, some infections during pregnancy) may slightly increase likelihood but do not determine outcomes on their own. Importantly, within environmental factors, it has been robustly proven that vaccines DO NOT cause autism.

Genetic Testing in Autism

Is genetic testing meant to cure autism?
No. It helps guide care and planning.

Is genetic risk inevitable?
No. It increases likelihood, not certainty.

What does genetic testing involve?
A blood or saliva sample is used to look for genetic changes. Genetic counselling helps to prepare for and interpret results.

What is the cost for genetic testing in Alberta?
It's overed by Alberta Health when eligibility criteria are met and testing is recommended by a physician.

Will my genetic information be safe?
There are laws such as the Genetic Non-Discrimination Act to help protect people.

Genetic Concepts and Interpretation

What does heritability mean?
A measure of how strongly genetic a medical or mental health condition is. Interestingly, it does not mean that this condition is inherited from parents to their kids.

Can environment help reduce genetic risk?
While environment cannot change someone's genetic makeup, it can influence how people adapt and cope, and their quality of life. Genetic influence does not remove the importance of support and context.

Does genetic risk mean something is inevitable?
No. Genetic changes may increase the chances of an outcome; they don't guarantee it. There is wide opportunity to help people achieve their maximum potential based on strengths and challenges in the context of genetic changes.

Rare Genetics

What is a copy number variant (CNV)? What is a deletion and duplication?
A copy number variant is when a small piece of a chromosome is missing (deletion) or duplicated (duplication). If they lead to clinical consequences, they are called pathogenic.

What can I do if my kid has/I have a pathogenic CNV?
Monitor development alongside your physician team, seek supports if needed (e.g., speech therapy, occupational therapy, physiotherapy), collaborate with schools and workplaces, and attend regular checkups.

How can I contribute to the greater understanding of rare genetic conditions?
Participating in research studies and patient registries helps advance our collective knowledge.

Want to Learn More?

Explore the full FAQ on our website or request a copy of our brochure.

If you have questions, contact us at prisma@ualberta.ca
Visit our web: www.precisionmedicineinautism.org/faq

Why Community Participation Matters

At PRISMA, we believe that research is about more than collecting information. It is also about people, their experiences, their questions, and the things that matter in their everyday lives. Every autistic person and family has a different story, and listening to these diverse perspectives can help us better understand autism, genetics, and the many ways they intersect with people's lives.

When people choose to participate in research, they contribute to something that goes beyond their individual experience. Their involvement can help researchers explore important questions, recognize patterns, and build knowledge that may inform future research, education, and healthcare. Not every study leads to immediate answers, but every contribution can be part of a larger effort to learn more.

We also recognize that not all communities have had the same opportunities to be represented in research. Different backgrounds, languages, cultures, and life experiences can bring perspectives that might otherwise be missing. By making research more accessible and welcoming, we hope to create opportunities for more people to share their experiences and contribute in ways that feel right for them.

Participation is always a personal choice. Whether someone decides to take part in a study, ask questions, share feedback, or simply learn more about our work, we value their interest and perspective. Research grows through curiosity, collaboration, and the willingness to keep learning from one another.

PRECISION MEDICINE IN AUTISM - RESEARCH PROGRAM

Explore Our RESEARCH



ABOUT THIS SECTION

Explore PRISMA's ongoing research studies and learn about opportunities to participate and contribute to a better understanding of autism and genetics.

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WHAT DO YOU THINK ABOUT GENETIC TESTING IN AUTISM?

Share your perspectives in a 20-minute online survey



- Autistic people in Alberta with a clinical diagnosis can participate
- Parents or guardians may participate on behalf of autistic people under their care
- Flexible communication options are available if needed
- Optional follow-up interview
- Receive a gift card for your time

SHARE YOUR PERSPECTIVE

Study title: Identifying Challenges and Opportunities for Implementing Standard of Care Genetic Testing for Autism and Neurodevelopmental Conditions

Contact: Dr. Daniel Moreno De Luca, prisma@ualberta.ca or (780) 492-8077

CASA Mental Health

UNIVERSITY OF ALBERTA

Recovery Alberta
MENTAL HEALTH AND ADDICTION SERVICES

REB ID (Pro00145198) - DATE: March 3, 2026

PRECISION MEDICINE IN AUTISM - RESEARCH PROGRAM

WHAT DO YOU THINK ABOUT GENETIC TESTING IN AUTISM?

Share your perspectives in a 20-minute online survey



- ☑ Physicians who care for autistic people in Alberta
- ☑ Flexible communication options are available if needed
- ☑ Optional follow-up interview
- ☑ Receive a gift card for your time

SHARE YOUR PERSPECTIVE

Study title: Identifying Challenges and Opportunities for Implementing Standard of Care Genetic Testing for Autism and Neurodevelopmental Conditions

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www.precisionmedicineinautism.org



PRECISION MEDICINE IN AUTISM - RESEARCH PROGRAM

DO YOU OR YOUR CHILD HAVE A 17q12 DELETION OR DUPLICATION?

Help advance research for the 17q12 community

- Open to children and adults with a confirmed 17q12 deletion or duplication, with or without an autism diagnosis
- This is a remote study, available in English, French, or Spanish
- Includes sharing health information, completing online questionnaires, and taking part in assessments
- Participation takes about 6 hours in total and can be completed in multiple sessions

Study title: A Genomic Approach to Precision Medicine for Autism and Neurodevelopmental Conditions

Contact: Dr. Daniel Moreno De Luca, prisma@ualberta.ca or (780) 492-8077 or visit our page www.precisionmedicineinautism.org/17q12cnvs

SCAN ME !

Take part in the study

CASA Mental Health

UNIVERSITY OF ALBERTA

Recovery Alberta
MENTAL HEALTH AND ADDICTION SERVICES

REB ID (Pro00145198)- DATE: September 19, 2024

PRECISION MEDICINE IN AUTISM - RESEARCH PROGRAM

From Community VOICES TO RESOURCES



ABOUT THIS SECTION

Discover PRISMA's educational materials, including practical resources, videos, and information about autism, genetics, and genetic testing, created to support learning and meaningful conversations.

Connecting Research and Community

Some of the most meaningful conversations begin with listening. Every person, family, and community brings different experiences, questions, and perspectives. These voices help us understand what matters to people, what information they are looking for, and how we can make autism and genetics easier to understand.

We also know that scientific information can sometimes feel complicated or difficult to access. That is why we care about creating educational materials that are not only accurate, but also welcoming, clear, and useful in everyday life. Whether it is a booklet, a video, a social media post, or a resource in another language, we want people to feel that the information was created with their needs in mind.

Our Community Liaisons, community conversations, and the feedback we receive help us see things from different perspectives. They remind us that there is no single experience of autism and that the questions people ask are just as important as the information we share. By listening and learning, we can continue improving the way we communicate and connect with others.

The materials in the following pages are part of this ongoing effort. Some are designed to answer questions, others to share knowledge, spark curiosity, or invite participation. We hope they offer something useful, encourage meaningful conversations, and remind you that there is always room for your voice in the PRISMA community.

Meet Our Community Liaisons

Every community brings its own experiences, questions, and perspectives. Listening to these voices helps us understand what matters most to individuals and families, and reminds us that meaningful research is shaped not only by scientific questions, but also by the people whose lives it hopes to improve.

Our Community Liaisons are an important part of the PRISMA team. They help us stay connected with two communities central to our work: the autism community and individuals and families affected by rare genetic conditions, including 17q12 deletion and duplication syndromes.

These communities bring different but equally valuable perspectives. Experiences within the autism community can help us better understand everyday needs, access to healthcare, and questions about genetics and genetic testing. Perspectives from the 17q12 community offer insight into living with a rare genetic condition, navigating healthcare, finding reliable information, and connecting with others who share similar experiences.

Meet Cristina and Catherine





Through their involvement, our Community Liaisons help strengthen relationships, share community perspectives, support conversations about research opportunities, and provide feedback on educational materials and community initiatives.

Their contributions remind us that while every experience is different, listening to one another can help build research and resources that are more inclusive, accessible, and meaningful.

We are grateful to have them as part of the PRISMA team and to continue learning from the communities they help us connect with. Their voices encourage us to ask better questions, consider different perspectives, and keep the experiences of individuals and families at the heart of our work. Together, we hope to create more opportunities for meaningful conversations, shared learning, and stronger connections between research and community.

PRINTED RESOURCES

Printed materials available upon request to support discussions on genetic testing.



Community Booklet

Our Community Booklet brings together information about autism, genetics, and genetic testing for Autistic people, families, and anyone curious to learn more. Inside, you'll find answers to common questions, educational materials, practical resources, and ways to connect with the PRISMA community.

Designed to explore at your own pace, this booklet offers a welcoming place to start, revisit a topic, or prepare for conversations with healthcare professionals. We hope it sparks curiosity, encourages questions, and helps you find information that is useful to you or someone you support.

Newsletters

Our PRISMA newsletters are also available as printed materials that can be requested for use in clinics and community settings.

These newsletters provide accessible information about genetics and autism, and can support conversations with patients and families.





Faq Brochure



Genetic Testing Fllyer



Research Study Flyer

If you would like to request any of these printed materials, please contact us.

LANGUAGE RESOURCES

At PRISMA, we believe that being able to access information in a language you understand matters. Clear information can help people and families explore questions about autism, genetics, and genetic testing, and find resources that may be useful to them.





• OUR WEBSITE

Our website is available in Spanish and French. Explore PRISMA's research, clinical work, and resources about autism, 17q12, and other genetic and neurodevelopmental conditions.

• SPANISH LANGUAGE BROCHURE

Our brochure brings together answers to common questions about autism, genetics, and genetic testing. It is designed to make this information clear and easy to explore for individuals and families.

• NEWSLETTER

Our newsletters share information about autism, genetics, community stories, resources, and PRISMA updates. We also have a newsletter about 17q12 available in Portuguese, making information about this genetic condition accessible to more families.



2026



2024



2020



2019

• LIBRARY OF LIFE VIDEO

Bo and DiNA make learning about genetics, autism, and genetic testing fun and approachable. The videos are available in English and Spanish, with a French version coming soon.

EXPLORE THE PRISMA LIBRARY OF LIFE

• PRISMA TEAM

Our team brings together different cultures, languages, and experiences. We speak Spanish and Portuguese, French and Italian, This diversity helps shape how we connect and work with our communities.

Where to Find Reliable Autism and Genetics Resources

To learn more about autism, genetics, or how the two can be connected, explore the trusted resources below.

How Genetics Works. University of Toronto
https://pie.med.utoronto.ca/htbw/module.html?module=genetics

An introduction to fundamental topics and basic explanations of genetics concepts. MedlinePlus
www.medlineplus.gov/genetics/understanding/

Glossary of genomic and genetic terms. National Human Genome Research Institute
www.genome.gov/genetics-glossary

How Can Genetics Help Us Understand Autism? Frontier's article
https://kids.frontiersin.org/articles/10.3389/frym.2025.1568090

Genetic Testing for Neurodevelopmental Conditions. GECKO-Genetics Education Canada
www.geneticseducation.ca/resources-for-clinicians/neurogenomics/ndd-main-landing/ndd-ind-families

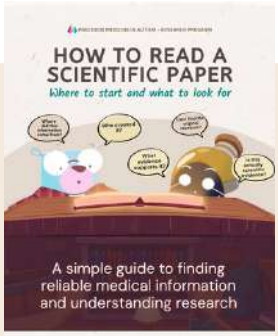
2014: Autism — what we know (and what we don't know yet) Ted Talk by Dr. Wendy Chung
www.ted.com/talks/wendy_chung_autism_what_we_know_and_what_we_don_t_know_yet

Canadian Genetic Non-Discrimination Act protecting genetic information
www.geneticseducation.ca/resources-for-clinicians/genetic-non-discrimination-act

Let's Learn Online Resources

Explore autism, genetics, and research through clear explanations and engaging visuals

Whether you're learning about these topics for the first time or looking to understand a little more, these carousels are a place to start. Each one breaks down a topic into simple steps you can explore at your own pace. You can also find them on PRISMA's **Instagram**, **Facebook**, and **LinkedIn**. We hope they spark your curiosity and help you feel more comfortable asking questions.



How to read a scientific paper



Autism+ Sleep



Myth or Fact



Fragile X Syndrome



Genetic Testing and Autism



A Brief History of Genetics



Autism



17q12 CNV's



17q12 TIPS

PRECISION MEDICINE IN AUTISM - RESEARCH PROGRAM

Beyond Research:
LEARNING AND CONNECTING



ABOUT THIS SECTION

Discover how PRISMA brings research, education, and community together through creative initiatives that encourage learning, meaningful conversations, and the sharing of diverse experiences.



WATCH NOW

SCAN ME!

Explore our Library of Life educational video, created to explain key concepts about autism, genetics, and genetic testing in a clear and engaging way.

The video is available in English and Spanish.Take a look at the links below

English: https://youtu.be/iXvd_GEdNCE

Spanish: https://youtu.be/mirvSuo3aS4

Library of Life

Our **body** is made of trillions of tiny cells, each with a **nucleus.**



The nucleus of each cell contains 23 sets of these chromosomes





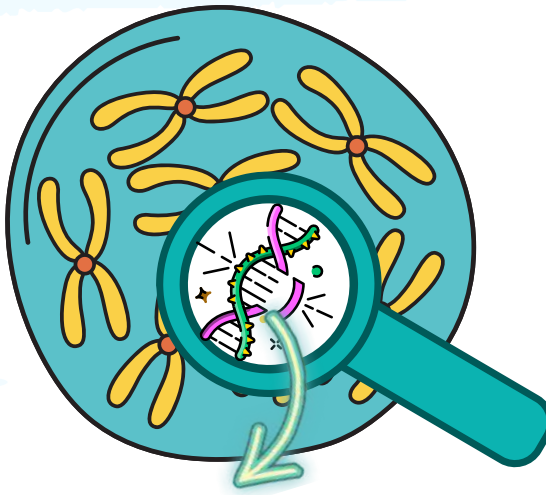
Chromosomes are like **books** in a library, holding instructions for how our body works.

Chromosomes are made of DNA strands





The instructions in your chromosomes are like words on a page.



A section of DNA is called a gene — like a specific word on a page.

A **gene mutation** is a small change in instructions that affects how the body works, like a **misspelled word** in a book.

MYTHS VS FACTS

Exploring 10 common myths about autism and genetic testing

1

MYTH: **Autism is unrelated to genetics.**

FACT: Autism is strongly influenced by genetics, but there is no single "autism gene." A specific genetic change may help explain the diagnosis in about 1 in 3 autistic people, particularly those with other medical or developmental conditions.

2

MYTH: **Autism is caused by parenting style or personal choices.**

FACT: Autism is not caused by parenting style or personal choices. It is a neurodevelopmental condition that arises from biological processes beginning early in development. Blame is not supported by scientific evidence and can increase stigma.

3

MYTH: **If autism is genetic, it must have been inherited from a parent.**

FACT: Some genetic changes associated with autism are inherited, while others occur spontaneously, called "de *novo*", and are not present in either parent's tested DNA. Identifying a genetic change does not automatically mean it was inherited from a parent.

4

MYTH: **Genetic testing can diagnose autism.**

FACT: Genetic testing cannot diagnose autism. Autism is diagnosed through developmental history, observation, and clinical assessments. Testing may identify an underlying genetic condition associated with autism, but it cannot confirm or rule out autism by itself.

5

MYTH: **Genetic testing is about trying to "cure" autism.**

FACT: Genetic testing is not about curing autism. Autism is a neurodevelopmental condition, not a disease to eliminate. Testing aims to better understand an individual's biology, identify associated medical conditions, and support care, planning, and decision making.

6

MYTH: **Genetic testing will not affect ASD care.**

FACT: Genetic information can help explain autism in some individuals, guide medical care, identify health risks and additional needs, inform family planning, and connect people with support groups. Genetic research also advances our understanding of neurodevelopment and helps improve supports and interventions.

7

MYTH: **All autistic people have an identifiable genetic change.**

FACT: Not all autistic people have an identifiable genetic change. Although genetics plays an important role in autism, testing cannot identify a specific genetic explanation for everyone. Both genetic and non-genetic factors may contribute to an individual's autism diagnosis.

8

MYTH: **Genetic testing is only useful for young children.**

FACT: Genetic testing can provide valuable information at any age. Results may guide medical care, identify health concerns that need monitoring, and connect people with specialized support and resources. They may also help professionals better understand a person's needs.

9

MYTH: **Genetic testing requires a blood draw**

FACT: Genetic testing may use blood or saliva to look for changes in chromosomes or genes. Genetic counseling helps individuals and families understand the results, their implications, and limitations.

10

MYTH: **If someone has already had genetic testing after an autism diagnosis, there is no reason to test again**

FACT: Genetic testing and knowledge continue to improve. People with previous negative or inconclusive results may benefit from discussing reanalysis of existing data or newer tests with a healthcare professional.

Curious to learn more?

Explore our **"Myth or Fact?"** series on PRISMA's social media channels (Instagram, Facebook, and LinkedIn) to learn more about common myths surrounding autism and genetics. Have a myth you'd like us to explore? Send it to prisma@ualberta.ca

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 **two 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 to pursue genetic testing is a personal choice. Autistic adults can make this decision for themselves, while families can discuss testing options for their children with their healthcare team.

There are multiple different genetic tests, and for autism, the ones recommended include: **chromosomal microarray testing.** This is considered the **standard of care for autism spectrum disorders** and is recommended by multiple medical professional societies, including the American Academy of Child and Adolescent Psychiatry, the American Academy of Paediatrics, and The American College of Medical Genetics and the American Society of Human Genetics jointly, with growing support for **exome sequencing 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 counselling 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 this section for your questions about autism, genetics, genetic testing, and precision medicine. We have an outstanding network of clinicians available to answer them. Submit your question at **prisma@ualberta.ca** and explore on **www.precisionmedicineinautism.org**



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 AND 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 night time 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.

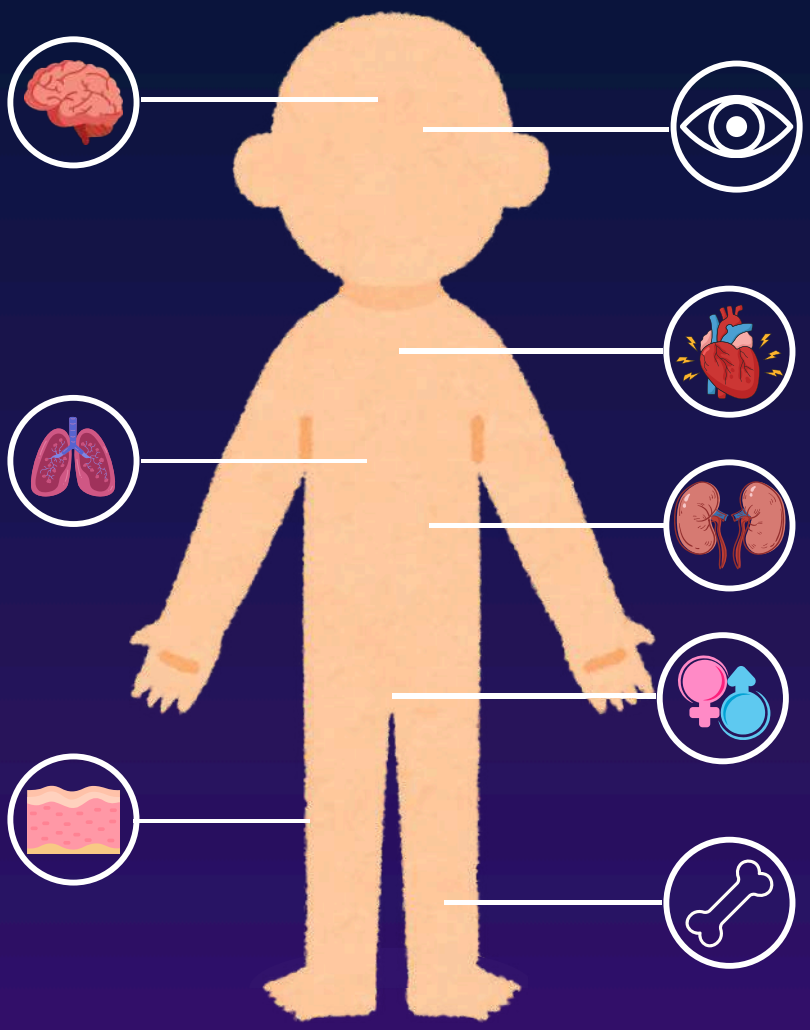
Have a question about autism, genetics, or precision medicine? Send us an email at prisma@ualberta.ca. In our next edition, we'll select one to answer!

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.

PRECISION MEDICINE IN AUTISM – RESEARCH PROGRAM

WHAT'S YOUR THING?



We'd love to learn about the hobbies, passions, and interests that bring you joy. If you or a family member is autistic, we'd love to hear your story. Submit the form using the QR code or email us at PRISMA@UALBERTA.CA

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Join the PRISMA community and stay up to date with our work in research, education, clinical care, and community. We are committed to working closely with the community, listening, sharing, and making information clear and accessible for everyone.

Follow us and visit our website to learn more about what we do and how you can get involved.

Have questions or want to connect? Contact us at **prisma@ualberta.ca or visit www.precisionmedicineinautism.org**