PRECISION MEDICINE IN AUTISM - RESEARCH PROGRAM

Genetic Testing

Physician Resources

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PRECISION MEDICINE IN AUTISM - RESEARCH PROGRAM

GENETIC TESTING QI PROJECT

This interactive document provides links to our website with essential resources for each step, including educational videos, pamphlets, test information, Connect Care instructions, and referral forms. At PRISMA, we've included some supplemental materials to support you in your genetic testing journey. We encourage you to explore these resources and use the checklist to ensure that all key aspects of pre-test counselling are covered.

Our team is available to assist you in ordering genetic testing and to offer counseling for any positive results through our Genetic Counselling & Genomic Psychiatry service. If you have any questions or would like more information, please don't hesitate to reach out to us by emailing prisma@ualberta.ca.

PRECISION MEDICINE IN AUTISM - RESEARCH PROGRAM

Meet the Team Behind the QI Project

The multidisciplinary team supporting genetic testing in autism spectrum disorder

GENETIC COUNSELLOURS

Genetic counsellors play a key role in supporting individuals and families in understanding genetic testing related to autism.

JULIA HEATON
Clinical Genetic Counsellor

MOLLY GOLDMAN
Research Genetic Counsellor

PRAJJITA BARDOLOI
Adult Psychiatrist

DANIEL MORENO DE LUCA
Child, Adolescent, and Adult Psychiatrist

ADE ORIMALADE
Adult Psychiatrist

PSYCHIATRISTS

The role of these three members is to help us learn more about genetics and genetic testing in the context of autism spectrum disorder.

This group is leading the effort for genetic testing "mainstreaming" empowering clinicians to independently implement genetic counselling in neurodevelopmental care

ONLINE SUPPORT

Can we indicate that online support is available via the handbook and website and that the clinics are available for any additional questions.

BO AND DINA
The role of these two new members is to help us learn more about genetics and genetic testing in the context of autism spectrum disorder.

Pre-Test (ASD)

1. Genetic Testing Checklist

Supports physicians in discussing genetic testing and ensuring informed decision-making

DIAGNOSTIC CRITERIA

Patient has met criteria for ASD genetic testing

Clinical diagnosis of autism spectrum disorder or global developmental delay, intellectual disability, or speech delay AND Incomplete, outdated or absence of genetic testing

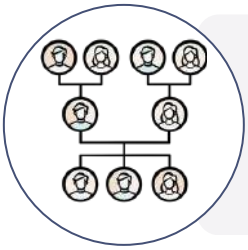
GENETIC TESTING

Understanding Genetic Testing for ASD

1. **ASD is diagnosed clinically**. A mixture of environment and genetic factors underly the condition. Over 100 genes, however, are known to play a role. Genetic testing can identify an underlying cause in 40% of individuals. Knowing genetic changes that cause a particular case of ASD could connect patient with organizations and families with the same mutation and guide personal medical management or future planning.

2. **Standard of Care:** Multiple medical professional societies including the Canadian Paediatric Society (CPS), American Academy of Pediatrics (AAP), American College of Medical Genetics (ACMG) and American Academy of Clinical Psychiatrists (AACP) have recommended genetic testing, specifically CMA as the standard of care.

3. **Genetic testing is always a choice**. Genetic counseling remains available should you want to discuss this in more detail.



FAMILY HISTORY

Complete three generation pedigree: (How To Draw Your Family History Pedigree Video)

UNDERSTANDING BASIC GENETICS

1. Share educational video
   a. English
   b. Spanish
2. Provide pamphlet
   a. Key Points: Genetic testing is a test that looks in your cells to identify an underlying genetic component to ASD
   b. Genes make us who we are.
   c. Tests: Chromosomal Microarray & ASD/ID Panel
   d. Genetic results may be **positive (pathogenic), negative (benign) or uncertain (VUS- variant of uncertain significance).**
   e. VUS result may or may not be associated with phenotype. Indicates evidence is insufficient or conflicting and not associated with disease. Classification of VUS might change over time.
   f. Genetic testing is protected by the Genetic Non-Discrimination Act (GNDA 2020)

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Genetic Testing Checklist

2.Clinical Action

Supports physicians in discussing genetic testing and ensuring informed decision-making

ORDERING CHROMOSOMAL MICROARRAY

- **ON CONNECT CARE**

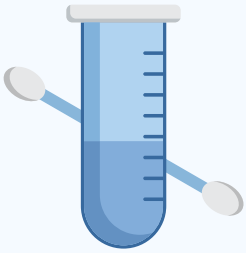
Chromosomal Microarray (CMA): Information for Ordering

- **OUTSIDE CONNECT CARE**

Cytogenetics Requisition form (CMA): GRC requisition for ASD /ID panel Testing

INDIVIDUALS SHOULD HAVE A NEGATIVE CMA BEFORE PROCEEDING WITH ASD PANEL TESTING.

ASD/ID PANEL TESTING



Ordering Panel Testing for Autism or Intellectual Disability

Two parts are needed for every panel test These steps don't need to be done in order.

No order is needed in ConnectCare and no blood draw is needed for the patient.

TIMELINE

Genetic test results are usually available in **6-8 weeks after** the blood sample is received in the lab

RESULTS DISCLOSURE

1. NEGATIVE: Please discuss results with the patient and assure they received a copy of their results as well as an appropriate handout. Results will live in EHR.

2. VUS: Please discuss results of VUS and assure they understand & receive a copy of their results. Results will live in EHR and should be reassessed with lab every two years. Consider clinical picture to see if genetics referral is warranted.

3. **POSITIVE:** Patients with a pathogenic results should be referred for an appointment at the Genetic Counselling Consultation to ensure that results are reported under the guidance of a genetic professional and management is reviewed. Results will live in EHR. Referrals to the Genetic Counselling/Genomic Psychiatry consult service using this form.

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Connect CARE System

Ordering CMA

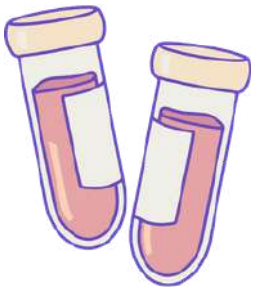
A structured form that supports physicians in ordering DNA storage through Connect Care

1

"Open the order 'Cytogenetic Analysis.' This can also be found by searching 'Chromosomal Microarray' or 'CMA.'"

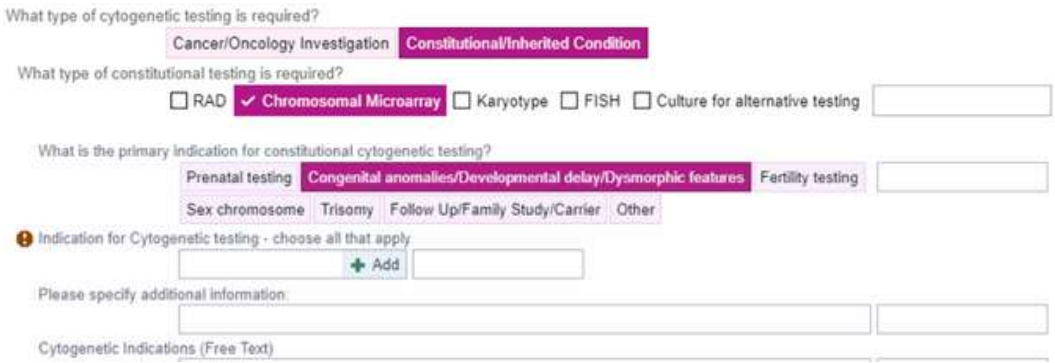
2

Testing is performed on blood. Confirm that pre-test counselling has been provided.



3

Select Constitutional/Inherited Condition and check only "chromosomal microarray"



4

Provide reason for testing and choose from the options provided. Be sure to specify whether the patient has: **autism spectrum disorder, intellectual disability speech delay, global developmental delay** from the "Indication for **Cytogenetic testing"** drop down box to ensure testing is accepted by the lab.

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How to Set Up Your GeneDX Account

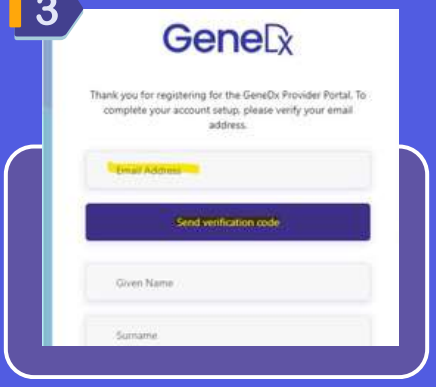
Step-by-step guidance to set up your GeneDX account for secure access and use.

1 Visit the www.genedx.com and select 'Provider Sign In,' then click 'Login' in the top right corner of the screen.

2

Select **"Sign Up Now"** to begin the registration process.

3



Enter your **email address** and select **"Send Verification Code"**.

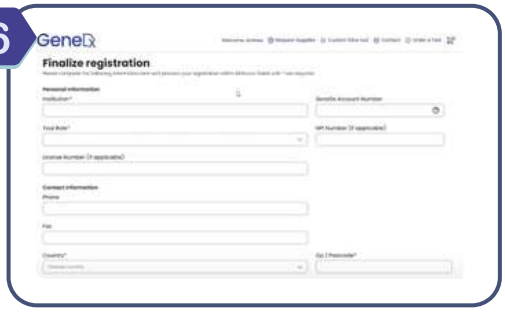
4

A **verification code** will be sent to your inbox. Enter it in the **"Verification Code"** field. Once verified, enter your name and create a password.

5

Once you click "Create," you will be taken to our main page. Please click on "Login"

6



Complete the 'Finalize Registration' page:"

- Enter institution name (**Edmonton Adult Neurodevelopmental Disorders Clinic**)
- Enter account number (**GC089**)
- Provide your role (**include PracID and License Number for ordering providers**)
- Enter **institution address, phone, and fax number**
- Select 'Save'

7 Upon completion, GeneDX will be notified to review the request and grant access as appropriate.

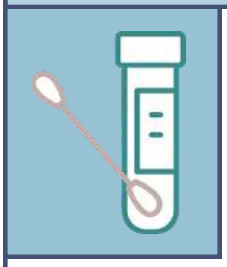
PRECISION MEDICINE IN AUTISM - RESEARCH PROGRAM

Ordering Panel Testing for Autism or Intellectual Disability

On Saliva (Buccal Swab)

Two parts are needed for every panel test. These steps don't need to be done in order.
No order is needed in Connect Care and no blood draw is needed for the patient.

1.FUNDING APPLICATION



Funding for the test is requested using the PDF GRC requisition. A partially completed version of this form is provided "Genetic Resource Centre" PDF requisition.

Add in patient and provider information on page 1, and a brief description of your patient's clinical features. If you've completed your GeneDx portal order already you can provide the order ID at the bottom of the first page, instructions for this are here. Once this form is completed, it should be emailed to the Genetic Resource Centre, GRC@albertaprecisionlabs.ca .The requisition must include a brief note.

Hello, Please find attached an application for autism/ID panel testing for @NAME@ @ULI@. Testing is requested on saliva as the patient has difficulty with blood collection . Kits will be provided to the family. Thank you!

2.PORTAL ORDER

The first time you order this testing, you will need to create an account with GeneDx. Reach out to the PRISMA team, we're happy to help you set this up, or watch this video that walks you through the process!

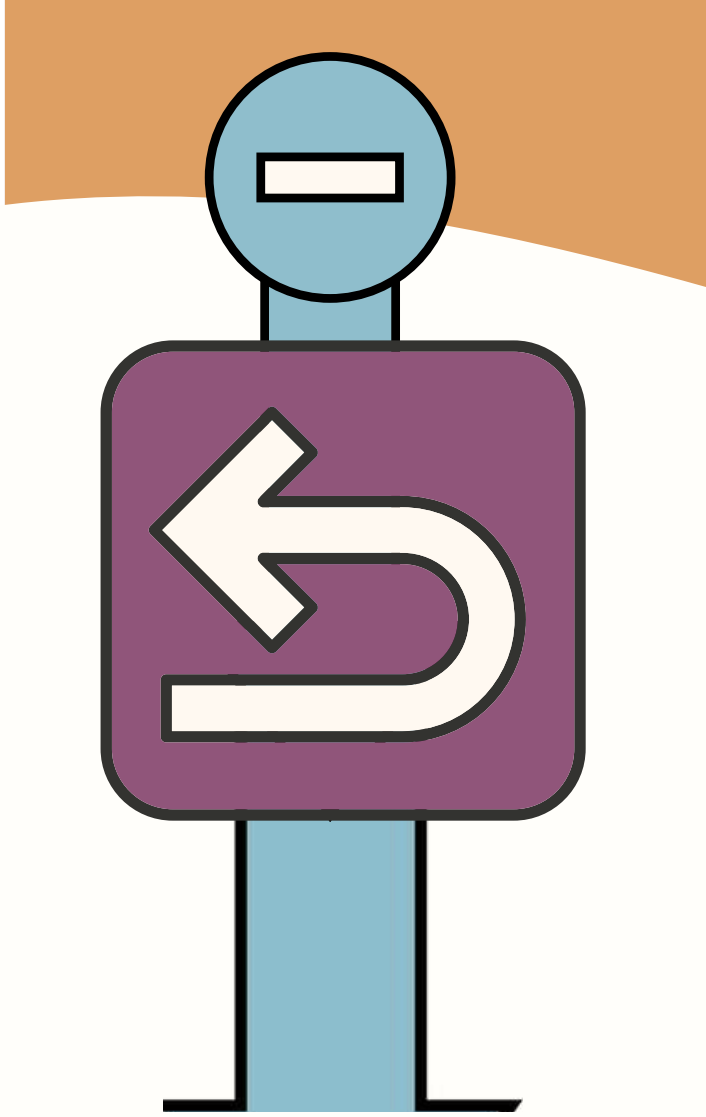
Log in to Provider Tools & Resources | GeneDx Provider Portal and search up test code **952: Autism/ID Xpanded Panel.** The team at GeneDx are available for any challenges you might run into. Some tips for completing your portal order:

- Share the order with "Amy Davis" to give the Genetic Resource Centre access to your order so they can upload the funding approval
- ICD-10 codes are optional, but a note or clinical letter needs to be included
- Testing has the highest yield when completed as a trio of your patient and their parents. If parents are available, have their names and dates of birth ready to add to the order
- You can ask GeneDx to send cheek swab collection kits to your patient's home address in the patient information section

3.TIMELINE

Genetic test results are usually available in 6-8 weeks after the sample is received in the lab

Return of Negative Results



- Even when genetic testing is negative, **genetics still play a role in most cases of autism and neurodevelopmental disorders.** Many cases are multifactorial, resulting from an interaction of several genes with environmental factors. A helpful counselling tool is the **"Jar Model"** of multifactorial inheritance developed by ***Dr. Jehannine Austin's team:*** Figure - PMC (nih.gov)[1]
- **A Variant of Uncertain Significance (VUS)** indicates evidence is insufficient or conflicting and **is not associated with disease.** VUS may or may not relate to phenotype. VUS should **not guide medical management.** It is possible for the classification of VUS to change over time and should be periodically reevaluated.
- **For either a negative or VUS result,** discussion with the family should convey **that no genetic diagnosis has been made by tests performed,** however this **can not rule out all possible genetic conditions.** For example, most single-gene disorders would not be detected by chromosomal microarray.
- **Consider referral to genetic services** for coordination of further testing if any of the below criteria are met.
  - **Multi-system involvement** (e.g. presence of congenital anomalies or significant physical health concerns)
  - **Multiple family members similarly affected**
  - **Parents of the affected individual are consanguineous** (related by blood)
  - **Family is highly interested in pursuing further testing** (e.g. for reproductive risk assessment for other family members) and patient has moderate to severe functional impact

Letter / Consult Note Template for Negative Results Return

Template is written to be saved as a **Connect Care smart phrase.** Please feel free to adapt and modify to suit your practice. This letter is to summarize genetic testing completed to date for @NAME@. Genetic testing was offered due to their diagnosis of *** and these investigations have now been completed. The results of genetic testing completed for @FNAME@ are summarized below:

- **Chromosomal Microarray:** No clinically significant gains or losses identified.
- **ASD/ID Panel:** No significant changes were found on the genes associated with the GeneDX ASD/ID panel.

We discussed that these results **greatly reduce the likelihood of a genetic diagnosis** for @FNAME@ but **cannot rule out a genetic cause beyond the detection limits of available testing.** In the absence of a genetic diagnosis, ***(diagnosis) is thought to be of **multifactorial etiology**. Multifactorial disorders result from an interaction of several genes with environmental factors. Recurrence risks are estimated from empiric evidence as well as the number of affected individuals in the family, and how they are related. **Management should continue to be based on @FNAME@'s presenting symptoms.**

Letter / Consult Note Template for VUS Results Return

Template is written to be saved as a **Connect Care smart phrase.** Please feel free to adapt and modify to suit your practice. This letter is to summarize genetic testing completed to date for @NAME@. Genetic testing was offered due to their diagnosis of *** and these investigations have now been completed. The results of genetic testing completed for @FNAME@ are summarized below:

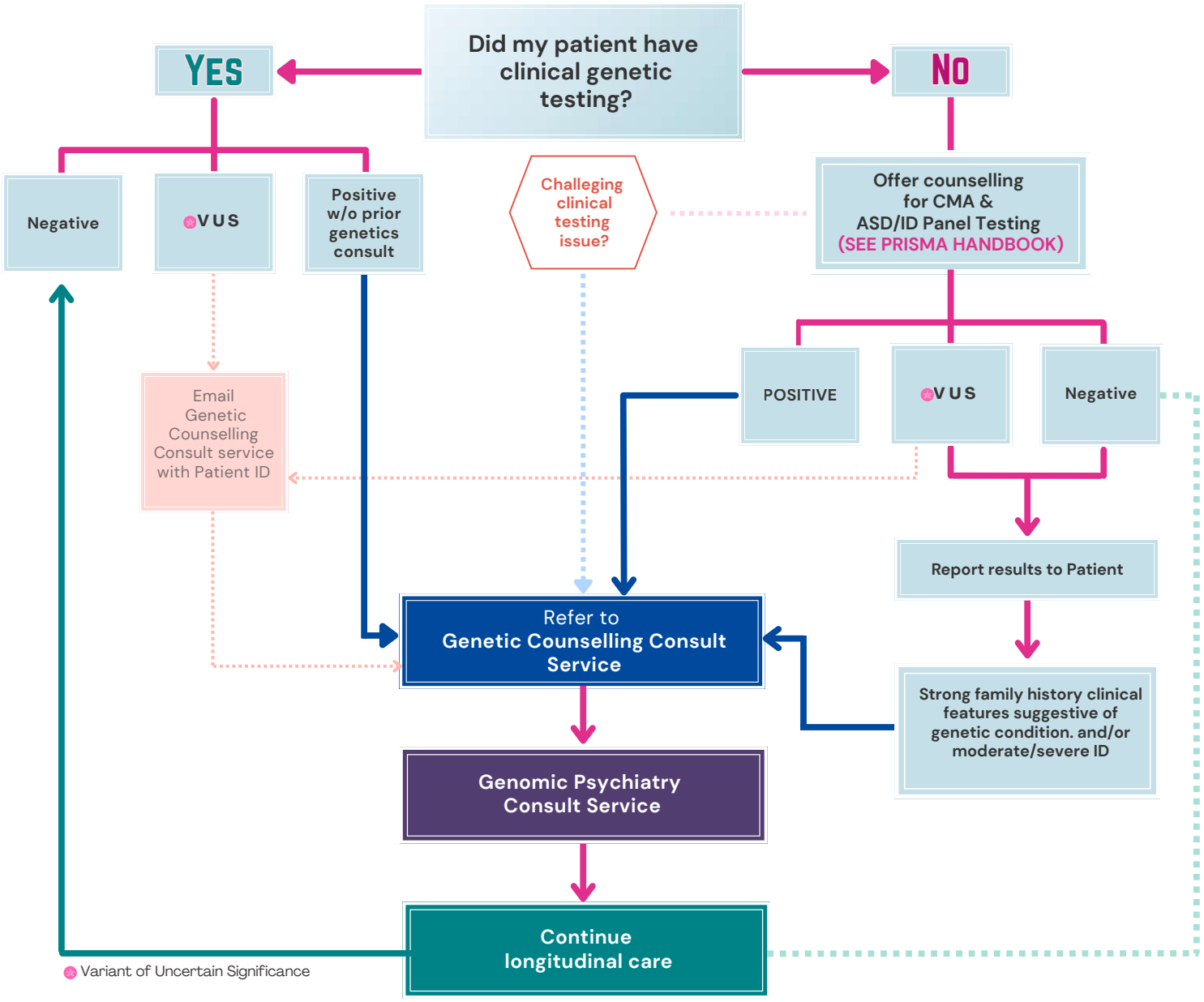
- **Chromosomal Microarray:** A VUS, namely _____ , was identified that has not been previously described in literature. The **clinical significance of this copy number variant is unknown** at this time, **does not alter immediate medical management. Re-evaluation of this finding with ordering laboratory is recommended every two years.**
- **ASD/ID Panel:** No significant changes were found on the genes associated with the GeneDX ASD/ID panel.

We discussed that these results **greatly reduce the likelihood of a genetic diagnosis** for @FNAME@ but **cannot rule out a genetic cause beyond the detection limits of available testing.** In the absence of a genetic diagnosis, ***(diagnosis) is thought to be of **multifactorial etiology.** Multifactorial disorders result from an interaction of several genes with environmental factors. Recurrence risks are estimated from empiric evidence as well as the number of affected individuals in the family, and how they are related. **Management should continue to be based on @FNAME@'s presenting symptoms.**

Austin JC. Evidence-Based Genetic Counseling for Psychiatric Disorders: A Road Map. Cold Spring Harb Perspect Med. 2020 Jun 1;10(6):a036608. doi: 10.1101/cshperspect.a036608. PMID: 31501264; PMCID: PMC7263094.

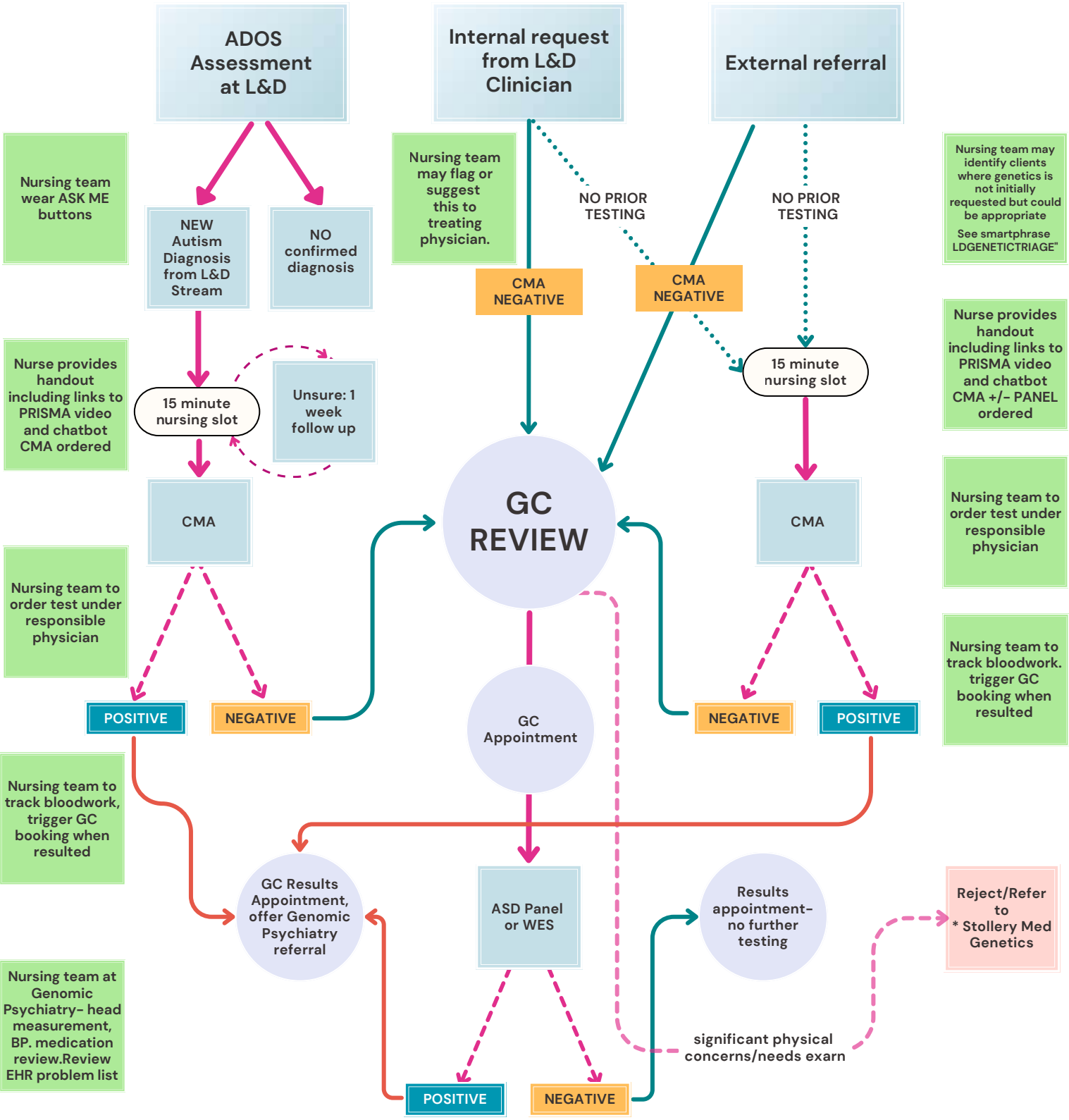
CLINICAL WORKFLOW

CHILD/ADOLESCENT PSYCHIATRIST, DEVELOPMENTAL PEDIATRICIAN CHILD/ADULT NEUROLOGIST, PRIMARY/FAMILY PHYSICIAN IN COLLABORATION WITH NURSING TEAM



This flow diagram outlines the pathway for **Genetic Counseling and Genomic Psychiatry Consultation services** for individuals with autism and other neurodevelopmental conditions. It illustrates how patients may be referred, assessed, and supported through genetic evaluation, result interpretation, and clinical follow-up, in collaboration with multidisciplinary care teams.

NURSING STAFF INTEGRATION INTO GENETICS



PRECISION MEDICINE IN AUTISM - RESEARCH PROGRAM

EDUCATIONAL RESOURCES

WHAT IS INCLUDED

The following materials are provided as supportive resources throughout the genetic testing process. They include educational videos, background information on genetic testing, research-based resources, and communication tools to support discussions with families

PRINTED RESOURCES

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

ASK ME" buttons

"ASK ME" buttons are available for clinicians and healthcare providers who currently discuss, or are interested in discussing, genetic testing with their patients. Wearing these buttons helps create an open and welcoming environment, encouraging patients and families to ask questions and start conversations about genetics and its role in their care.

These buttons are a simple way to signal approachability and support informed, patient-centered discussions around genetic testing.

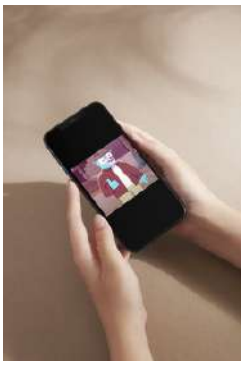
Newsletters

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.

PHYSICAL - QI HANDBOOK

FAQ BROCHURE



CHAT BOT

If you would like to request any of this physical material, please contact us at prisma@ualberta.ca.

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=Es8g8u_tp-g - A

SCAN ME!

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 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!

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 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 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.com**

PRISMA
PRECISION MEDICINE IN AUTISM

Where you'll meet Bo, DiNA, and the rest of their friends!

WATCH NOW

SCAN ME!

We are eager to launch our new video aimed at people on the autism spectrum and their families, explaining in a clear and engaging way key concepts like autism, genetics and genetic testing.

Our video is now available in English and Spanish! Take a look at the links below

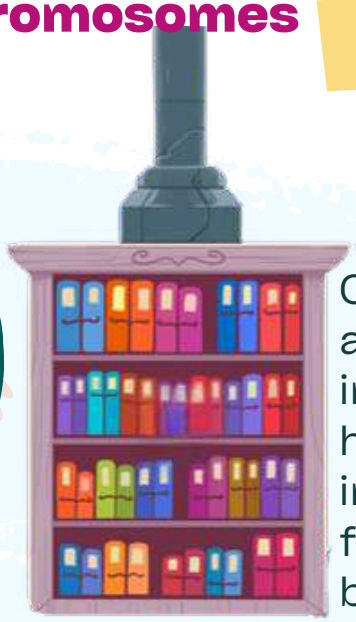
English: https://youtu.be/iXvd_GEdNCE

Spanish: https://youtu.be/mirvSuo3aS4

Library of Life

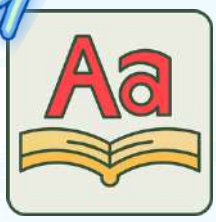
Our **body** is made of trillions of tiny cells, each with a **nucleus.**

The nucleus of each cell contains 23 sets of these chromosomes

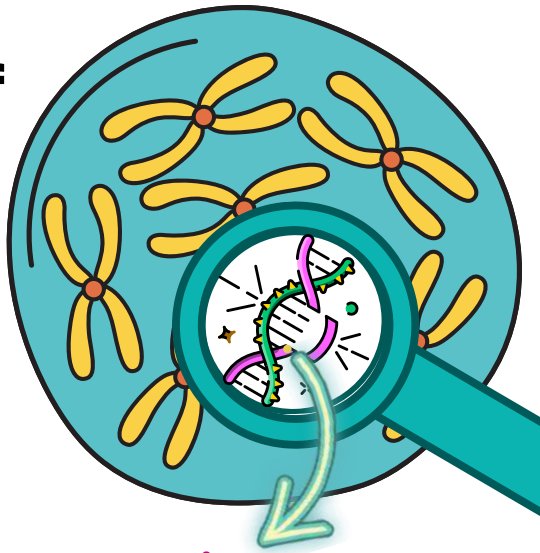


Chromosomes are like **books** in a library, holding instructions for how our body works.

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.

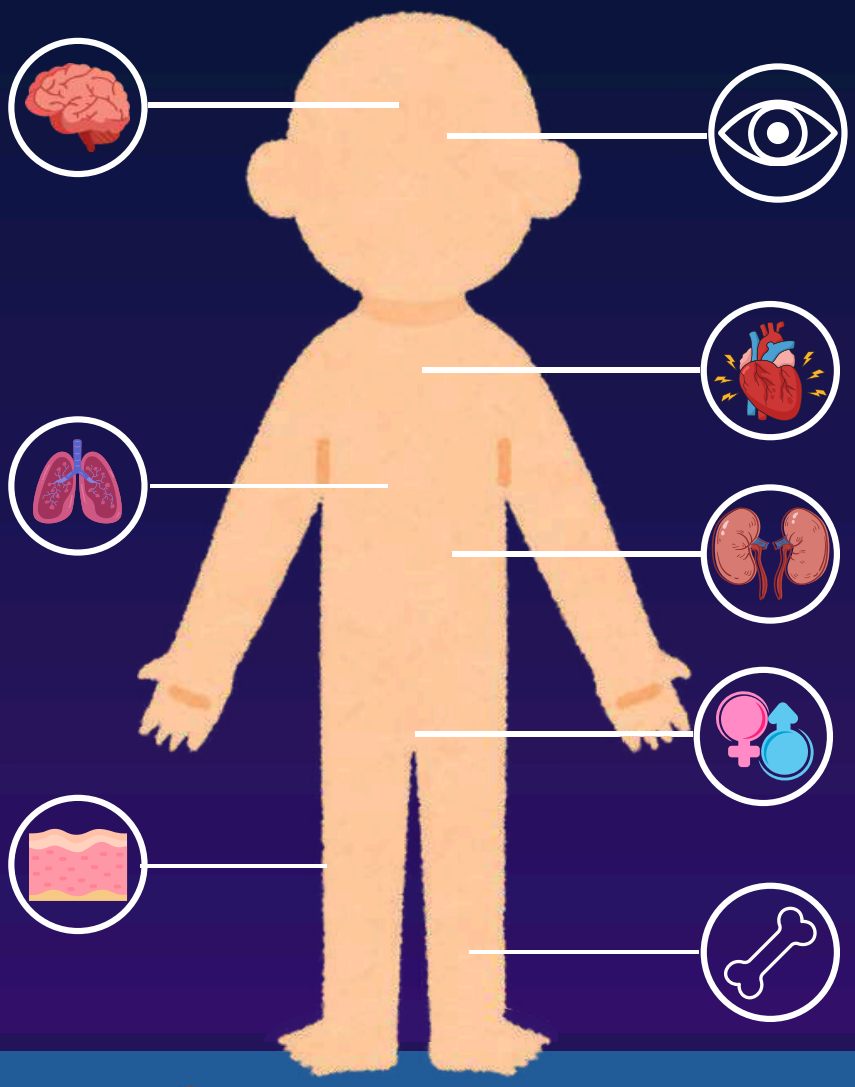
A **gene mutation** is a small change in instructions that affects how the body works, like a **misspelled word** in a book.

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 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.

QUALITY IMPROVEMENT

WHAT IS INCLUDED

The following resources support continuous quality improvement by collecting feedback and promoting research participation to improve access to and implementation of genetic testing in clinical practice.

PRECISION MEDICINE IN AUTISM - RESEARCH PROGRAM

GENETIC TESTING QI PROJECT SURVEY

We encourage you to reflect on this process by completing a short survey: **Genetic Testing QI: Clinician Feedback.** Visit tinyurl.com/3ut2vuzn or scan the QR code.

Your successes and challenges will help us shape the future of this important service, thus improving patient care.

If you have any questions or would like more information, please don't hesitate to reach out to us by emailing **prisma@ualberta.ca**

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

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

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