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!