# ASK A DOCTOR

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

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

- Autism is a clinical diagnosis, meaning that it is given based on the observations by the doctors during an office visit, or with additional psychological testing, such as the Autism Diagnostic Observation Scale (ADOS). That means that genetic testing is not used to diagnose autism; it is recommended after the diagnosis of autism has already been made to uncover potential causes.

- The decision about carrying out genetic testing should be a joint process with your medical team, and the decision to proceed is entirely in the hands of each family.

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

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

- Finding an explanation and underlying cause for the autism in a given family and putting an end to the diagnostic odyssey.

- Identifying risk for other behavioral and medical conditions associated with a given genetic change, such as cardiac or renal abnormalities, which may in turn impact clinical management, including additional workup and medication choice.

- Obtaining genetic counseling and risk assessment for family planning.

- Having a clearer picture of areas of strengths and vulnerabilities based on information from other families with the same genetic abnormality and accessing specific medical resources.

- Connecting with support groups of other families with the same genetic variant.

- Being eligible for clinical trials targeting people with a specific genetic variant. All the examples above encompass what we now call precision medicine - the ability to use precise individualized information for tailored clinical management.

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

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

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