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HOW CAN GENETICS HELP US UNDERSTAND AUTISM?

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Autism affects how people communicate, interact, and respond to things around them. People on the autism spectrum often have very strong interests, do some things repeatedly, and may feel most comfortable keeping their routines the same. Genetic tests can identify a cause for autism in up to one in three people. While these tests cannot diagnose autism, after a diagnosis is made, genetic testing can give doctors helpful clues to help families and find the right support. In this article, we explain key information about genetics, how genetic testing works, and how it can be used in autism. Taking a genetic test is a personal choice, and it is important

ZUNAIRA

AGE: 13

to talk with doctors or other healthcare professionals before making that decision. However, by helping people understand these topics, we hope to make it easier for families to ask questions, make choices, and get the support that fits them best.

WHAT IS AUTISM?

We all have different ways of existing in the world, and **autism** is one of those ways. Autism is a developmental condition that affects the way people communicate and relate to other people. Autistic people may also react differently to sounds, lights, smells, or textures. It is very common that people on the autism spectrum have intense interests and enjoy doing some things over and over again. The word "spectrum" is used in autism to reflect that each autistic person is unique, with different challenges, abilities, and talents—just like everyone else.

Doctors figure out if someone is autistic by talking to them and their family, asking questions about how they learn, play, and interact with others. Doctors might also use checklists or simple activities to understand how the person's brain works. Getting diagnosed with autism can help people get the support they might need to do their best at school, at work, or in everyday life.

AUTISM

A developmental condition that affects how people communicate and relate to others, and react to their environment (like sounds or textures). Autistic people also have intense interests, or enjoy doing certain things repeatedly.

GENOME

All the DNA in a living organism, including the genes (or words) that make up the complete set of genetic instructions and the spaces between and within those genes.

CHROMOSOMES

Thread-like structures inside cells made of DNA that act like a package for the genetic instructions, helping organize and protect them.

GENE

A small section of DNA with a specific meaning, like a word in a book, and job in helping the body grow and work properly.

GENETICS: OUR BUILDING BLOCKS

Scientists are doing research to better understand autism because there is still much that we do not know. However, we have already learned a lot about an important area that helps us understand autism better: genetics.

Genetics is the science that studies the building blocks of how any living being is made, including humans. We all have our entire set of genetic instructions, called our **genome**, in almost every cell in our bodies, and the genome tells our bodies how to grow and work. We can think of the genome as a library full of books. The human genetic library has 23 pairs of books, which we call **chromosomes**. We get one book in each pair from our biological mom and one from our biological dad. Each of those books has many words in it, which we call **genes**, and is written in a very special language, called DNA (Figure 1).

With such a big library, scientists have discovered hundreds of different genetic changes that explain why some people are autistic. Each of these changes is rare on its own, but together, they can be found in one out of every three people on the autism spectrum [1]. Most of the time, these genetic changes do not come from one of our parents but instead happen by chance—this is called *de novo*.

Figure 1

The library of life explained: how our bodies hold our genetic instructions. About one in three people on the autism spectrum have one of the many different genetic causes for autism.

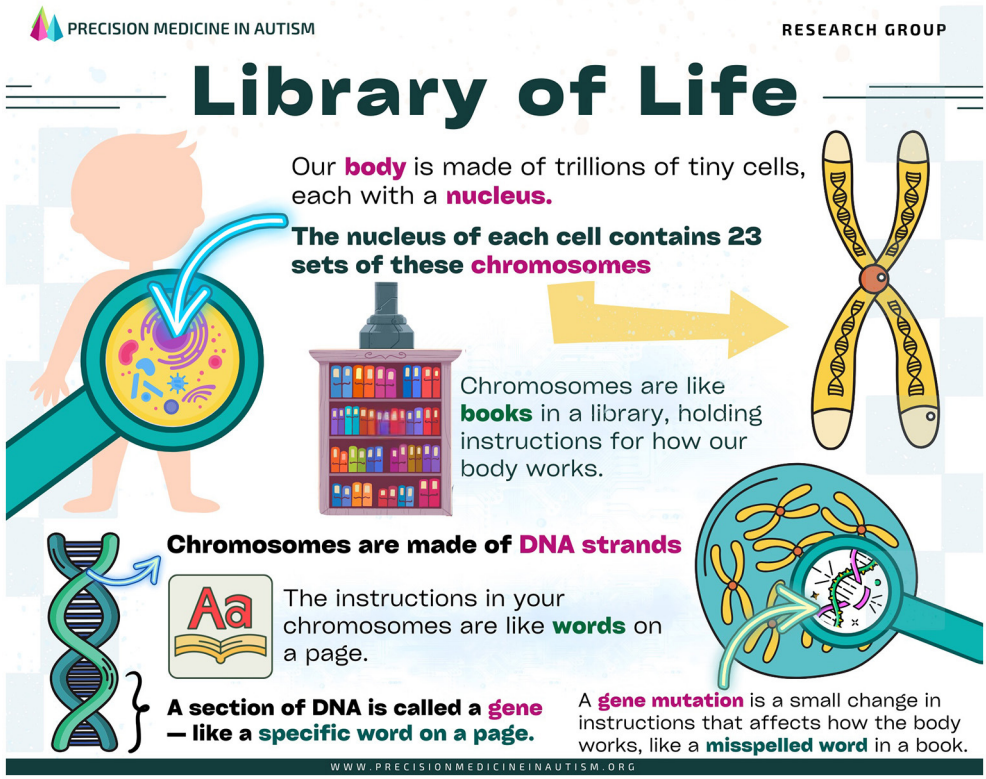

Figure 1

Although autism *cannot be diagnosed* by looking at a person's genetic library, it is very important to look at the genomes of people on the autism spectrum *after* a diagnosis is made. Finding a genetic change can help autistic people and their families understand the reason for this condition in their case, and can give doctors important information to help when needed.

CHROMOSOMAL MICROARRAY

A genetic test that looks at the amount of DNA across the entire genome, and can identify missing or extra pieces of genetic material that may include one or many genes.

HOW CAN WE LOOK AT THE GENOME?

We can look at the genome through genetic testing, which can be done as part of medical care by using a sample of saliva or blood. Genetic testing refers to any test that looks at the genetic library, and experts recommend offering specific genetic tests for people on the autism spectrum [2].

One of these tests is called **chromosomal microarray**. This test looks for any missing or extra pieces of DNA and tells us what specific part of the genetic library is involved. By looking at every single book, this test can detect both big and small changes and tell doctors whether a genetic change is likely the reason someone is autistic.

In addition to changes in the total amount of DNA, like missing or extra pieces, other changes affect only the spelling of the words in the genome. We need special tests to detect these spelling changes.

EXOME SEQUENCING

A genetic test that looks at the letters that make up the all the genes to identify any changes in spelling.

GENOME SEQUENCING

A genetic test that looks at the letters that make up all the genes, as well as the spaces within and between the genes, to identify any changes in spelling.

FRAGILE X TESTING

A genetic test that looks at the letters of one single gene on the X chromosome to identify a different kind of spelling change that causes Fragile X syndrome.

Exome sequencing and **genome sequencing** are tests that look at a person's entire genetic library, going over each of the words to find spelling changes. Exome sequencing focuses on reading all the words themselves, while genome sequencing also looks at the spaces between and inside the words in the genome.

Lastly, there is a special test called **Fragile X testing**, which looks at one specific gene because changes in this gene are tricky to find with other tests. Genetic changes in this gene cause Fragile X syndrome, which is one of the most common reasons why someone might be autistic [3]. This gene is located on the X chromosome, which can easily break during genetic testing—this is why it is called "Fragile X".

WHAT HAPPENS WHEN YOU HAVE A GENETIC CHANGE?

There are a couple of things to keep in mind to understand genetic changes and how they affect the body.

First, not all genetic changes affect how the body works. Only some changes cause health problems, these are called pathogenic changes. Think of them as a misspelled word in one of the books in the genetic library that gets in the way of understanding of the story. On the other hand, some changes do not cause any problems and are called benign changes. This is like when there is a different spelling of a word in one of the books that does not stop us from understanding the story.

Second, some genetic changes can affect more than one part of the body. For instance, the same genetic change can lead to autism *and* skin changes. Imagine a misspelled word in a book that changes the meaning of not only the sentence you are reading, but also the plot in other chapters. This is one reason why people on the autism spectrum might have other medical conditions, like seizures or heart problems.

Each genetic test helps doctors find answers in different ways. Results can help doctors and families learn about specific genetic changes that might explain autism. Results can also help doctors know if there are any health problems to look out for and how to make a care plan that fits each person's needs. However, it is important to remember that the decision about testing is entirely up to the person and their family, just like any other medical test. Families should discuss genetic testing with their doctor or medical team before making this important decision.

If you want to learn more about our work, visit www.precisionmedicineinautism.org. There, you will find a fun video explaining our "library of life" and plenty of other cool facts about genetics and autism!

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CONFLICT OF INTEREST: The authors declare that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

YOUNG REVIEWERS

MERIEM, AGE: 14
I am a bilingue student, I am interested in arts, and the correlation between art and science, I also also love communication and team work.

SAMI, AGE: 14
I am a student in a Canadian school, I am passionate about the animal world and the nature, I love design and illustration. I am a member of a robotic team.

YAHIA, AGE: 12
As a student in a French school, I have many interests beyond school domains, I am part of a robotics team and a judo competitive player. I love understanding natural phenomena and I love the teamwork.

ZUNAIRA, AGE: 13
I am Zunaira, a middle school student. I love reading, tennis and baking. I like learning about interesting scientific discoveries.

AUTHORS

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Dener is an autistic scientist who researches autism and genetics. While learning about autism in university, he found out he was autistic. This made him want to learn even more about autism. Now, he works with the PRISMA research group, researching how genetics can help us learn more about autism. In his free time, Dener loves listening to music and learning about linguistics.

MOLLY GOLDMAN
As a genetic counselor, Molly helps people understand how science, specifically genetics, relates to themselves and their own lives. She often provides resources and community connections to support each individual and celebrate their uniqueness. Molly enjoys her morning nature walks and is passionate about photography and playing board games with friends.

SILVANA GUERRERO

With a background in marketing engineering, Silvana works as a research assistant, closely engaging with the community to serve as a bridge between the public and the medical and scientific fields. She communicates complex information in a simple, friendly way that is easy to understand. Part of her role involves circulating information in different languages to ensure accessibility across diverse communities worldwide.

KEELY VACHON

Keely works as a research assistant with the PRISMA research group. She holds a Master of Science in pediatric medical sciences and helps find participants for research studies, aiding in connecting different research projects. She plays a key role in ensuring the success of the research.

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Carrie works as a research coordinator with the PRISMA research group, and is the mother of two autistic children. She holds a Master of Public Health degree in health behavior and health education from the University of Michigan School of Public Health, and she has been doing autism research for 10 years.

DANIEL MORENO-DE-LUCA

Daniel is a physician and scientist who studies the brain and the genetics of mental health and developmental conditions. He cares for individuals with autism, bipolar disorder, schizophrenia, and other mental health needs, especially when these are related to rare genetic changes. His research focuses on understanding these genetic causes to improve care and provide better guidance. He also shares knowledge about these topics with other clinicians and the general public. Together with his team, he works closely with the community to ensure their voices are heard loud and clear. *daniel.morenodeluca@ualberta.ca