Ask a Lurie Center Researcher: What does the latest research reveal about autism and its origins?

Autism Spectrum Disorder (ASD) has attracted significant attention from the scientific community over the past few decades as the number of diagnoses has increased[1]. Much of this research has been directed at understanding the root causes of autism. While there is still much we don't understand, we know that autism is highly heritable. However, genetic factors are not the whole story. In this article, we summarize what research is showing about the complex biology underlying ASD.

What role do genes play in autism?

Our genes influence nearly everything about us, and this includes likelihood of developing ASD. By studying families, researchers have learned that autism has a clear genetic basis. For identical twins, who share virtually all their DNA, if one twin has autism there is a 60-90% chance that the other twin will as well. On the other hand, for fraternal twins, who share about half their DNA, if one twin has autism there is a 3-30% chance that the other will as well. While these numbers are imprecise estimates, they clearly indicate a strong genetic link. Population based studies back this up. For example, a study in Sweden of over 7 million people estimated the heritability of ASD to be 83%, suggesting that genetic factors explain the majority, but not all, of the likelihood of ASD[2]. That said, for the majority of people on the autism spectrum, a specific genetic change cannot yet be identified.

Digging a bit deeper into the genetics of ASD, some genetic forms of autism can be caused by altering just one letter of the DNA code. These changes, called single nucleotide variants (or SNVs), can be present in the parental DNA or they may appear for the first time in the DNA of the child (called de novo variants). Other genetic changes consist of chunks of DNA that have been doubled or deleted, called a copy number variant. While the change of just one letter of the code, or just one gene, might seem insignificant, these types of changes can have profound effects on health and brain development.

Sometimes the genetic cause is not due to a single gene, but rather the combined effect of multiple genetic variations. On their own, each one of these genetic changes is insignificant, but together they can add up or interact in ways that increase the likelihood of autism. These more complex genetic causes are often influenced by additional factors, such as the environment in which the brain is developing, which can further shape how these genetic changes affect development.

In short, there is no singular genetic cause of autism. Over 400 genes have been linked to autism and large-scale cohort studies currently in progress, like the SPARK study, will add to this number and continue to clarify our understanding[3,4]. In the SPARK study, about 8–10% of autistic participants who join learn about a genetic change that may help explain their autism.
(See below for more information about how to enroll in this study).

What about environmental factors?
While genetics are a big factor in the development of autism, they do not tell the whole story. Environmental influences can compound genetic factors and make the development of autism more likely to occur. Environmental influences are wide ranging and include, but are not limited to, toxins in the air and water, the medications we take, the food we eat, the medical care we have access to, and our early life experiences. Environmental factors affecting autism have their greatest effects during gestation and in early life, which are critical periods of brain development.

Several environmental factors have been consistently linked to an increased likelihood of autism, as seen in the table below. In the late 1990s and early 2000s there was a theory that vaccines given to pregnant mothers or to infants may be responsible for autism. However, many studies have now repeatedly shown vaccines do not cause autism [5,6,7,8].

Environmental Factors Linked to ASD	Environmental Factors Studied and Unlikely to be Linked to ASD
Prematurity, very low birth weight, or traumatic birth	Vaccines
Maternal illness or infection during pregnancy	IVF (In vitro fertilization)
Advanced parental age	C-section
Maternal gestational diabetes	Plasticizers
Exposure to air pollution during pregnancy	Maternal cigarette smoking
Valproic acid (anti-seizure medication)	Maternal marijuana
	Breast feeding
	Terbutaline (Medication treating lung diseases)
	Fluoxetine (Prozac, antidepressant)
	Acetaminophen (Tylenol, pain reliever)
	Topiramate (anti-seizure medication)
	Antipsychotic medication

Table adapted from Halladay, A. (2025). Environment Cause of Autism with Dr. Alycia Halladay. The Complex Causes of Autism: Webinar Series

Importance of the gene-environment interaction
It can be helpful to think of the variety of genetic and environmental factors like drops of water in a bucket. If these factors accumulate, it becomes more likely that the bucket will fill up (and that autism will develop). This means that even two people with the same genetic material (i.e., identical twins) may have different outcomes because they experienced different environmental

factors during gestation (such as having different birth weights). Similarly, two people with similar gestational experiences (such as fraternal twins exposed to a serious maternal infection while in the womb) might end up with different outcomes or traits based on their different genetic backgrounds. This complex interplay is what makes autism so difficult to understand and predict.

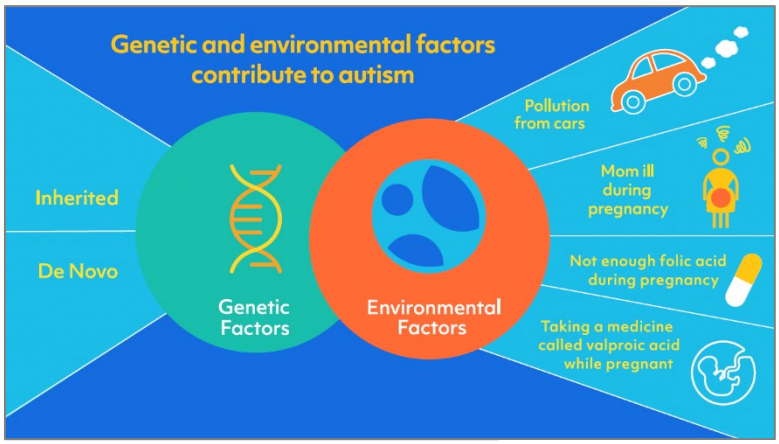

Image Credit: SPARK

What about the brain?

Genetic and environmental factors interact to determine how the brain forms. During gestation, when brain cells are being created, and early development, when brain cells are making connections with each other in response to an influx of new information, subtle individual differences can result in behavioral changes.

Some common differences in brain structure seen in autistic individuals may be that the brain is slightly smaller or larger than average, that brain cells are connected more or less frequently, or that messages between brain cells travel at a different speed than average[9,10,11]. However, these differences exist on a continuum throughout the entire population. You cannot tell whether or not someone has autism just by looking at an image of their brain, and this is one of the reasons why diagnosis is based on developmental history, input from parents, and professional observation.

While autism is a lifelong neurodevelopmental condition, many individuals benefit from supports and therapies such as Applied Behavioral Analysis (ABA) that help build skills and foster growth. These interventions may work in part because of the brain's ability to adapt and form new connections, a quality known as neuroplasticity. Neuroplasticity allows the brain to develop new ways of processing and responding to information throughout life in both autistic and non-autistic individuals[12].

In summary

There is no single cause of autism. As a spectrum disorder, autism reflects a wide range of differences in genetics, environment, and brain development, helping to explain the tremendous diversity seen among individuals with the same diagnosis.

Researchers continue to investigate the underlying causes of autism with the goal of uncovering patterns that can lead to more precise, personalized care. The more we understand about how

autism develops, the closer we get to interventions that are tailored to the unique needs of each person.

Although many questions remain, the progress made in recent decades is promising. With continued research and collaboration, there is real hope that these discoveries will translate into more effective supports and better outcomes for autistic individuals and their families.

By Audrey Carter, a Lurie Center for Autism Summer Research Intern.

ADDITIONAL RESOURCES

- National Institute of Child Health and Human Development
- Autism New Jersey Webinar Series

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Join the Largest Autism Research Study in the Country!

The Lurie Center for Autism is recruiting individuals of all ages with autism spectrum disorder (ASD) for SPARK. SPARK's mission is to improve the lives of people with autism by identifying the causes of autism and informing more effective therapies, treatments, services, and supports. Participation includes a 20-minute online registration. Children or adults with a professional diagnosis of autism and their families are asked to provide saliva samples, complete compensated surveys, and learn about other research opportunities. Samples are paid up to $50 and surveys up to $25. You will receive the SPARK newsletter and be invited to monthly webinars with experts about issues that matter to your family. Up to 10% of participants will receive genetic findings explaining the cause of their autism. Registration can be done entirely online at www.sparkforautism.org or in person with the MGB SPARK team.

To learn more and join the study, visit www.sparkforautism.org/MGB