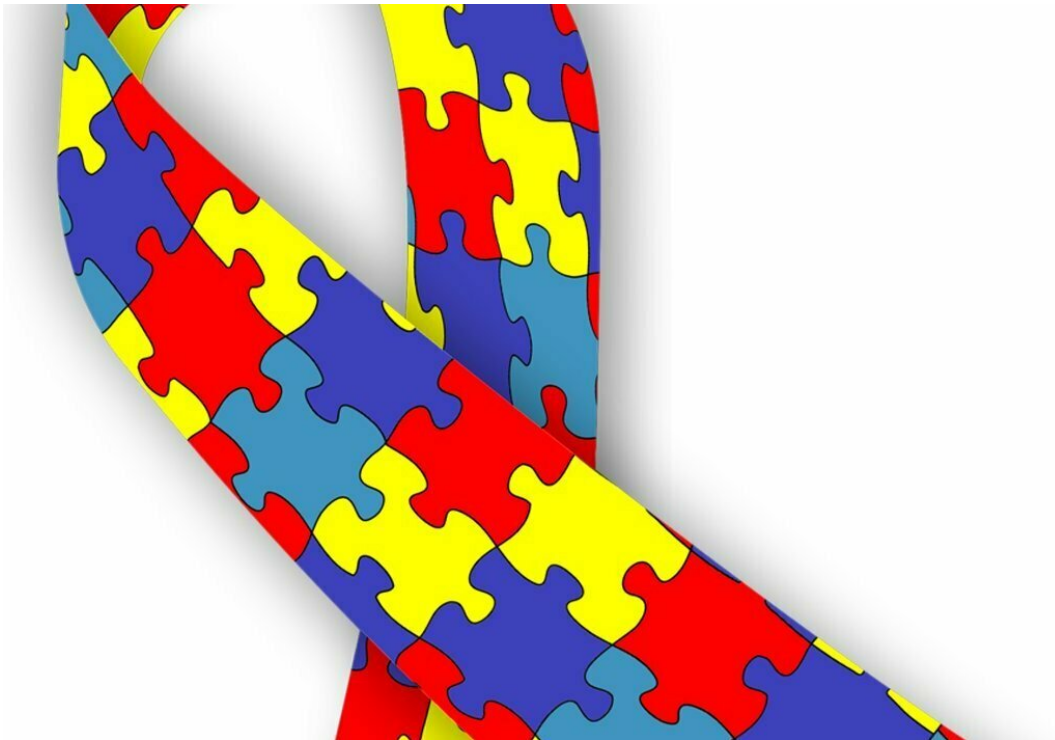


Study finds only 3% of individuals with autism receive recommended genetic tests

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A study analyzing data from the Rhode Island Consortium for Autism Research and Treatment (RI-CART) found that only 3% of individuals diagnosed with autism spectrum disorder reported having fully received clinical genetic tests recommended by medical professional societies.

The results bring to light a dissonance between professional recommendations and [clinical practice](#), the researchers behind the study say.

Autism spectrum disorder is one of the most strongly genetic neuropsychiatric conditions. Medical professional societies—such as the American Academy of Pediatrics, the American College of Medical Genetics, and the American Academy of Child and Adolescent Psychiatry—recommend offering chromosomal microarray testing and Fragile X testing for patients diagnosed with autism. The tests can identify or rule out genetic abnormalities that could have implications in a patient's diagnosis and [clinical care](#).

The study, published in *JAMA Psychiatry* on May 13, analyzed 1,280 participants with [autism spectrum disorder](#) based on medical records and self-reported data from the time period of April 2013 to April 2019. The participants are enrolled with RI-CART, a public-private-academic collaborative focused on advancing research and building community among individuals with autism spectrum disorder in Rhode Island and their families. The study's goal was to determine the current state of clinical [genetic testing](#) for autism in this cohort, said authors Dr. Daniel Moreno De Luca and Dr. Eric Morrow.

Of the 1,280 participants, 16.5% reported having received some genetic testing, with 13.2% stating they received Fragile X testing, and 4.5% reporting that they received chromosomal microarray testing. However, only 3% of participants reported having received both recommended tests.

"I had the impression that the frequency of recommended genetic testing was not going to be very high based on the patients I encounter clinically, but 3% is actually lower than I thought it would be," said Moreno De Luca, an assistant professor of psychiatry and human behavior at Brown

University, who is affiliated with the Carney Institute for Brain Science, and a psychiatrist at Bradley Hospital. "A higher proportion has had either [test](#) individually, and the proportion of people with chromosomal microarray is higher in recent calendar years, which is a hopeful glimpse for people who are being diagnosed recently and who may be younger. However, this underscores that there is still significant work to be done, especially for adults on the autism spectrum."

In the study, researchers examine possible reasons for the gap between clinical practice and the recommendations from medical professional societies. Age was among the most prominent, as people with autism in older age groups are less likely to be tested. According to the study, adults with autism were generally unlikely to have undergone the clinical genetic tests.

The researchers also found that patients diagnosed by subspecialist pediatricians were more likely to report genetic testing as compared to those diagnosed by psychiatrists and psychologists.

"This paper is really about how you implement clinical genetic tests in the clinical diagnostic setting," said Dr. Eric Morrow, an associate professor of biology at Brown and director of the Developmental Disorders Genetics Research Program at Bradley Hospital. "There is rapid progress from research, and then there's the doctor and [health systems](#) that need to translate that to clinical practice. The clinics need to set up more support to educate clinicians and families about genetics and autism. Generally, this is done by genetic counselors who may be rare in autism clinics."

Furthermore, the researchers found that nearly 10% of participants who received an autism spectrum disorder diagnosis between 2010 and 2014 reported receiving chromosomal microarray testing, one of the more modern genetic tests. Compared to those in the study who received a

diagnosis in years before 2010, this showed an increase in self-reported testing.

"There is a more hopeful message that conveys that the success in implementing clinical genetic testing is increasing," said Morrow, who is affiliated with the Carney Institute, co-leads the Autism Initiative at the Hassenfeld Child Health Innovation Institute at Brown and directs the University's Center for Translational Neuroscience.

Based at Bradley Hospital in East Providence, the team behind RI-CART represents a partnership between researchers at Brown, Bradley Hospital and Women and Infants that also involves nearly every site of service for people on the [autism](#) spectrum and their families in Rhode Island.

As a next step, the researchers behind the *JAMA Psychiatry* study are conducting a separate study to understand in greater detail the factors that could be influencing the rate of genetic testing.

"Challenges can be found on the patient and families side, on the physician side, and on the systemic side with institutional requirements and many other potential barriers," said Moreno De Luca. "We want to address each of those factors independently."

More information: *JAMA Psychiatry* (2020). [DOI: 10.1001/jamapsychiatry.2020.0950](https://doi.org/10.1001/jamapsychiatry.2020.0950) , jamanetwork.com/journals/jamapsychiatry/fullarticle/2765958

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