

NEWS

Roots of language troubles, autism traits may diverge

BY NICHOLETTE ZELIADT

15 SEPTEMBER 2014





© Alloy Photography/Veer

The genetic and environmental factors that underlie difficulties with language differ from those that influence other autism traits, according to a new study of more than 3,000 twin pairs. The study, published 2 August in the *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, suggests that difficulty with language is not a core symptom of autism¹.

The core symptoms of autism are impaired social and communication abilities, and **repetitive and restricted behaviors** and interests. Until 2013, the diagnostic criteria for autism included a third category: language problems. In support of this, a few studies have identified genetic variants that **link language impairments to autism**.

But a more nuanced view has emerged in the past 20 years. “It’s now apparent that many people with autism have really quite good structural language, meaning they have large vocabularies, can produce sentences and identify meanings of words,” says **Catherine Lord**, director of the Center for Autism and the Developing Brain at New York-Presbyterian Hospital. Still, many people with autism **struggle with certain aspects of language**, such as conversational skills.

To resolve whether language difficulties are a core symptom of autism, the new study looked at whether this trait shares common origins with other symptoms characteristic of the disorder. The researchers went about this by looking at symptoms of autism in identical twins, who share essentially all of their genetic material, and fraternal twins, who share only half of their genes — as all non-twin siblings do — but who experienced the same uterine environment.

They found little concordance between language problems and other signs of autism in either type of twins.

“We expected high levels of autistic traits to correlate with low levels of language abilities, because language has been considered such a core part of the autism phenotype for so long,” says lead investigator **Angelica Ronald**, senior lecturer in psychological sciences at Birbeck, University of London. “But the correlations we found are really quite modest.”

Language link:

The findings support the decision to **exclude language difficulties** from the list of autism’s diagnostic criteria in the latest edition of the *Diagnostic and Statistical Manual of Mental Disorders*.

“This study suggests that that was a reasonable decision to make,” says **Annette Estes**, associate professor of speech and hearing sciences at the University of Washington in Seattle, who was not involved in the study. “It suggests that maybe language is more of a secondary phenomenon to autism spectrum disorders.”

The results also fit with the so-called ‘**fractionable** autism triad hypothesis,’ posited by Ronald and her colleagues in 2006². That theory suggests that the core symptoms of autism each **stem from distinct genetic causes**.

Ronald and her colleagues analyzed data on autism symptoms and language abilities in 3,222 pairs of 12-year-old identical or fraternal twins. The study included 35 participants diagnosed with autism. The data are part of the **Twins Early Development Study**, an ongoing project in the U.K.

Parents of the children filled out a 30-item questionnaire about their children’s autism traits, including social deficits, communication difficulties and repetitive and restricted behaviors and

interests. Each child completed four Internet-based tests of language abilities.

Each test assessed a different aspect of language ability, such as the richness of the participants' vocabularies, their understanding of the rules governing sentence structure, or syntax, and their ability to use language in a social context or to comprehend the non-literal meanings of words.

The researchers first looked at whether having autism traits corresponded to low scores on the language tests for each individual. "We know that autism is not just a case of having impaired language, but you might expect a degree of correlation between those two variables," Ronald says.

But that was not the case. The researchers found that the severity of an individual's autism symptoms does not predict his or her language ability.

The researchers then compared autism traits and language skills between the two members of a twin pair. They found that identical twins score more similarly to each other on both these aspects than do fraternal twins. This suggests that autism traits and language skills each have genetic components.

Trait tracking:

To estimate whether these sets of traits arise together, the researchers compared each twin's scores for autism traits with his or her co-twin's scores on the language tests.

If autism traits and language difficulties are inherited together, the two measures should track together more in identical twins than in fraternal twins; if shared environmental factors are at work, then the concordance for fraternal twins should be at least half of that calculated for identical twins.

But the researchers found little evidence for either scenario: The correspondence between the two measures is minimal, and does not differ between identical and fraternal twins. That suggests that language ability and other autism traits have few genetic and environmental influences in common.

This pattern holds even among those with the greatest impairments — individuals within the most severe 2.5 percent of the range of scores for autism traits and language difficulties.

"Maybe language is more of a secondary phenomenon to autism spectrum disorders."

The findings have important implications for researchers designing genetic studies. "We hypothesize that we're going to find different genetic variants that play a role in normal development of language from those genes that play a role in development of autistic tendencies," Ronald says.

Ronald cautions that the findings may not apply to cases of 'syndromic' autism, in which the disorder is associated with a known genetic defect, such as **tuberous sclerosis** or **fragile X syndrome**. "We need to check whether the findings are generalizable," Ronald says.

Other researchers echo these concerns.

"The number of individuals in the sample with an autism spectrum condition is relatively small," says **Joachim Hallmayer**, associate professor of psychiatry and behavioral sciences at Stanford University in California, who was not involved in the study. He also questions how well certain aspects of language ability, particularly the use of language in social settings, can be assessed over the Internet. "It's not easy to measure, actually."

What's more, the autism questionnaire used in the study, a modified form of the **Child Autism Spectrum Test (CAST)**, flags subtle symptoms that are not specific to the disorder, Lord says. Some of the participants with autism may also have been incapable of completing the computerized language tests, she adds.

Researchers could address some of these limitations by directly assessing language abilities and autism symptoms rather than relying on parent reports and Internet-based tests, says Estes. But these limitations aside, the results are surprisingly straightforward, she says. "A lot of times in autism research, you don't find such clear results."

References:

1. **Taylor M.J.** *et al. Am. J. Med. Genet. B. Neuropsychiatr. Genet.* **165**, 587-595 (2014) [PubMed](#)
2. Happé F. *et al. Nat. Neurosci.* **9**, 1218-1220 (2006) [PubMed](#)