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Researchers discover many genetic keys needed to unlock autism

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Hundreds of small genetic variations are associated with autism spectrum disorders, including an area of DNA that may be a key to understanding why humans are social animals, according to a multi-site collaborative study led by researchers at Yale University.

Published in the June 9 issue of the journal *Neuron*, the study reinforces the theory that autism, a disorder that develops in early childhood involving impairments in social interaction, language deficits and distinctive behaviors, is not caused by one or two major genetic defects, but by many small variations, each associated with a small percentage of cases.

The study—led by Matthew State, M.D., Ph.D., the Donald J. Cohen Associate Professor of Child Psychiatry, Psychiatry and Genetics—looked at more than 1,000 families in which there was a single child with an autism spectrum disorder, an unaffected sibling and unaffected parents. The team, including postdoctoral fellow and first author Stephan Sanders from Yale, compared individuals with autism to their siblings to determine what types of genetic changes distinguished the affected child from the unaffected child.

"Thanks to an ambitious collaboration among a large group of autism researchers from around the country, supported by the Simons Foundation, we were able to focus on an ideal study population," said State, who is co-director of the Yale Program on Neurogenetics. "It made all the difference in our ability to identify several regions of the

genome clearly contributing to autism."

One of the most intriguing of these findings points to the same small section of the genome that causes Williams syndrome—a developmental disorder marked by high sociability and an unusual aptitude for music.

In autism, there is an increase in the chromosomal material, an extra copy of this region, and in Williams syndrome, there's a loss of that same material," said State. "What makes this observation particularly interesting is that Williams syndrome is known for a personality type that is highly empathetic, social, and sensitive to the emotional state of others. Individuals with autism often have difficulties in the opposite direction. This suggests that there is an important key in that region to understanding the nature of the social brain."

State and his team also found about 30 other regions in the genome that are very likely contributing to autism and are focused on about six of those regions that showed the strongest evidence. "We're now moving on to a second phase of the study looking at an additional 1,600 families and should be able to identify multiple new regions that are strongly implicated in autism," he said.

Sanders and State are optimistic about the new findings, pointing out that genetics is the first step to understanding what's really going on at the molecular and cellular level of the brain. "We can use these genetic findings to begin unraveling the underlying biology of autism," said Sanders. "This will help tremendously in the effort to identify new and better approaches to treatment."

Two other studies published in the June 9 issue of *Neuron* report on the same families studied by State, Sanders and their co-authors. One of these, by a group at Cold Spring Harbor Laboratory in New York, paints a very similar picture—that autism is a highly genetically diverse

disorder and that sporadic changes in the structure of the genome present only in the affected individuals and not in other families often play a key role. The other study, by researchers at Columbia University, suggests that although hundreds of genes may be involved in autism, they appear to disrupt a common molecular network involved in the mobility of brain cells and development of synapses between them.

More information: Christopher Mason, Rahul Dhodapkar, Vikram Fielding-Singh, Daniel Fishman, Sindhuja Kammela, Brian O'Roak, Rebecca Pottenger and Ilana Yurkiewicz. *Neuron*, Vol. 70, Issue 5 (June 9, 2011).

Provided by Yale University

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