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Autism and language impairment genetically linked

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Lorenzo Miodus-Santini an 11-year-old sixth-grader from Princeton, who was classified as autistic at only 13 months old, was never a big talker. As an infant he didn't babble or coo. When he was a toddler beginning to speak, he would learn one word but forget another.

His older brother, Christian, a 15-year-old high school sophomore, shared some similar characteristics – difficulty with reading, processing words and speaking clearly. Doctors said he had language impairments but was not autistic.

New research published online today in the *American Journal of Psychiatry*, by scientists at Rutgers University and The Research Institute at Nationwide Children's Hospital in Ohio, reveals that there is a genetic link connecting family members with autism like Lorenzo Miodus-Santini to those like his brother, Christian, who have specific language impairment characterized by speech and language difficulties that can't be explained by cognitive or physical problems.

The research project leader Linda Brzustowicz, Rutgers professor and chair of the Department of Genetics, in the School of Arts and Sciences, says that genes in a narrow region of two chromosomes (15q23-26 and 16p12) responsible for oral and written language impairments can result in similar behavioral characteristics with one family member developing autism and the other having only language difficulties.

Specific language impairment is one of the most common learning

disabilities, affecting an estimated 7 percent of children. It is not considered to be an autism spectrum disorder. Autism effects one in 88 children nationally – with nearly five times as many boys than girls diagnosed – about half of whom have some degree of language impairment.

"In this group of families we are trying to find genetic factors that might connect them," says Brzustowicz, who collaborated on the study with Christopher W. Bartell, principal investigator in the Battelle Center for Mathematical Medicine at Nationwide Children's Hospital. "This research is important because it is hard to understand autism until we find the genes that might be involved."

While scientists don't believe that there is one single gene that causes autism but rather a number of genes that increase the risk, Brzustowicz and her team of researchers are working to identify genetic patterns in these families in order to help gain a better understanding of the mechanisms that lead to autism, a developmental brain disorder that appears in the first three years of life.

In the Rutgers autism study, 79 families – mostly from New Jersey and Pennsylvania – with one child with autism and at least one with specific language impairment underwent extensive in-home testing. Besides taking blood samples for genetic testing, family members including parents, children, and grandparents and in some cases even uncles, aunts and cousins underwent a battery of tests to assess grammar, vocabulary and language processing.

"Our results indicate that there are shared patterns of DNA and visible behavioral characteristics across our group of study families," says Judy Flax, an associate research professor working on the study with Brzustowicz.

In addition to the language findings, researchers also found strong evidence of a genetic link in the areas of obsessive-compulsive, repetitive behaviors and social interaction skills, other symptoms associated with autism.

Brzustowicz says the next step will be to sequence the whole genome of those who participated in the study in order to compare the families to see if scientists can pinpoint any specific genes or mutations that are common to all.

It is part of a long-term collaboration between scientists from Rutgers and Nationwide Children's Hospital, as well as scientists, data experts and physicians from Rutgers, Saint Peter's University Hospital in New Brunswick and the Rutgers University Cell and DNA Repository (RUCDR).

Brzustowicz and her team have been studying the genetic influences of autism on families for the past decade – recently receiving a $2.2 million five-year grant from the state last year. They are opening the study to new families with autism as they continue the study over the next four years.

"This is just the beginning," says Brzustowicz. "We are finding evidence of genetic similarities with the hopes of being able to identify targets that might respond to pharmacological treatments."

Provided by Rutgers University

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