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Gene mutation is linked to autism-like symptoms in mice, UT Southwestern researchers find

DALLAS – Feb. 23, 2010 – When a gene implicated in human autism is disabled in mice, the rodents show learning problems and obsessive, repetitive behaviors, researchers at UT Southwestern Medical Center have found.

The researchers also report that a drug affecting a specific type of nerve function reduced the obsessive behavior in the animals, suggesting a potential way to treat repetitive behaviors in humans. The findings appear in the Feb. 24 issue of the *Journal of Neuroscience*.

“Clinically, this study highlights the possibility that some autism-related behaviors can be reversed through drugs targeting specific brain function abnormalities,” said Dr. Craig Powell, assistant professor of neurology and psychiatry at UT Southwestern and the study’s senior author.

“Understanding one abnormality that can lead to increased, repetitive motor behavior is not only important for autism, but also potentially for obsessive-compulsive disorder, compulsive hair-pulling and other disorders of excessive activity,” Dr. Powell said.

The study focused on a protein called neuroligin 1, or NL1, which helps physically hold nerve cells together so they can communicate better with one another. Mutations in proteins related to NL1 have been implicated in previous investigations to human autism and mental retardation.

In the latest study, the UT Southwestern researchers studied mice that had been genetically engineered to lack NL1. These mice were normal in many ways, but they groomed themselves excessively and were not as good at learning a maze as normal mice.

The altered mice showed weakened nerve signaling in a part of the brain called the hippocampus, which is involved in learning and memory, and in another brain region involved in grooming.

When treated with a drug called D-cycloserine, which activates nerves in those brain regions, the excessive grooming lessened.

“Our goal was not to make an ‘autistic mouse’ but rather to understand better how autism-related

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genes might alter brain function that leads to behavioral abnormalities,” Dr. Powell said. “By studying mice that lack neuroligin-1, we hope to understand better how this molecule affects communication between neurons and how that altered communication affects behavior.

“This study is important because we were able to link the altered neuronal communication to behavioral effects using a specific drug to ‘treat’ the behavioral abnormality.”

Future studies, Dr. Powell said, will focus on understanding in more detail how NL1 operates in nerve cells.

Other UT Southwestern researchers participating in the study were co-lead authors Jacqueline Blundell, former postdoctoral researcher in neurology, and Dr. Cory Blaiss, postdoctoral researcher in neurology; Felipe Espinosa, senior research scientist in neurology; and graduate student Christopher Walz.

Researchers at Stanford University also contributed to this work.

The research was supported by Autism Speaks, the Simons Foundation, the National Institute of Mental Health, BRAINS for Autism, and the Hartwell Foundation.

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