

Researchers develop mouse model to help find how a gene mutation leads to autism

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Researchers from Mount Sinai School of Medicine have found that when one copy of the SHANK3 gene in mice is missing, nerve cells do not effectively communicate and do not show cellular properties associated with normal learning. This discovery may explain how mutations affecting SHANK3 may lead to autism spectrum disorders (ASDs). The research is currently published in *Molecular Autism*.

"We know that SHANK3 mutation plays a central, causative role in some forms of [autism spectrum disorders](#), but wanted to learn more about how it does this," said Joseph Buxbaum, PhD, Director of the Seaver [Autism](#) Center and Professor of Psychiatry, Neuroscience and Genetics and Genomic Sciences at Mount Sinai School of Medicine. "These data provide critical insight into the mechanism behind the development of the cognitive and social changes associated with autism."

Previous research has shown that [gene mutation](#) in SHANK3 is associated with delayed language abilities, learning disability, and ASDs. A team of researchers at the Seaver Autism Center for Research and Treatment at Mount Sinai School of Medicine and the Intramural Research Program of the National Institute of Mental Health wanted to better understand the connection between the SHANK3 mutation and subsequent brain and behavioral difficulties. They examined mice genetically engineered to lack one copy of SHANK3, similar to patients who have a mutation in one copy of SHANK3, and compared the nerve cell activity of these mice with that of mice in a control group that did not have the mutation. They also examined social behaviors in these

mice.

Mount Sinai scientists looked at brain activity in vitro and worked with the NIMH Laboratory of [Behavioral Neuroscience](#), led by Jacqueline Crawley, PhD, to evaluate behavioral differences in the two groups of mice. The research team observed impaired communication between nerve cells in the mice with the SHANK3 mutation. They also found altered functional and structural plasticity in nerve cells, which is a cellular measure of the flexibility that occurs during learning, and in the synapses—the points of contact between [nerve cells](#). Behavioral observations indicated reduced male-female social interactions in the SHANK3 mutant mice. The studies identify clear brain targets that can implicate drugs that can be therapeutic.

"These results have helped us determine a pathological mechanism behind neurodevelopmental disorders like autism," said Dr. Buxbaum. "Currently, the only therapeutic options for people with ASDs are to treat the symptoms of the disease, like anxiety or aggression. Armed with this breakthrough, we can begin testing drug compounds that treat the disease at its root cause, improving nerve cell communication. We hope and expect that, like other developmental disorders such as Fragile X syndrome, the use of mouse models will lead directly to clinical trials that can benefit patients."

Provided by The Mount Sinai Hospital

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