

Study finds first-ever genetic animal model of autism

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By introducing a gene mutation in mice, investigators have created what they believe to be the first accurate model of autism not associated with a broader neuropsychiatric syndrome, according to research presented at the American College of Neuropsychopharmacology annual meeting.

This animal model could help researchers better understand abnormal brain function in autistic humans, which could help them identify and improve treatment strategies. Broader neuropsychiatric conditions include Fragile X, the most common cause of inherited mental impairment, and Rett Syndrome, a childhood neurodevelopmental disorder characterized by normal early development followed by slowed brain and head growth, seizures, and mental retardation.

Autism is a neuropsychiatric disorder characterized by repetitive behaviors and by impairment in social interactions and communication skills. These symptoms can coexist with either enhanced or decreased cognitive abilities and skills.

“Prior to this study we knew next to nothing about the mechanisms of autism in the brain,” says study researcher Craig M. Powell, M.D., Ph.D., assistant professor of neurology and psychiatry at the University of Texas Southwestern Medical Center at Dallas. “With this research, we can study changes in the brain that lead to autistic behaviors and symptoms, which may help us understand more about progression and treatment of the disorder.”

The research team, led by Thomas Südhof, M.D., professor and chairman of neuroscience at UT Southwestern, replaced the normal mouse *neuroligin-3* gene with a mutated *neuroligin-3* gene associated with autism in humans. By doing so, the team was able to create a gene in the mice that is similar to the human autism disease gene. While the result amounted to a very small change in their genetic makeup, it perfectly mimicked the same small change occurring in some patients with human autism.

Dr. Powell studied the genetically altered mice and found that, when examined in behavioral tests that may reflect key signs of autism, they showed decreased social interaction with other mice; other traits, such as anxiety, coordination and pain sensitivity, were unaffected. These social interaction deficits, Dr. Powell says, are hallmark features of human autism. In addition, the mice showed enhanced spatial learning abilities, which may resemble the enhanced cognitive abilities in autistic savants (people who have a severe developmental or mental handicap as well as extraordinary mental abilities).

“These findings could be especially helpful in identifying novel treatment approaches. We already know that inhibitory chemical synaptic transmission from one neuron to the next is increased in this mouse model. Now we can test drugs that decrease this effect directly in the mice and ask whether this reverses their social interaction deficits,” Dr. Powell says. “For now, the mainstay of autism treatment is still behavioral therapy. The earlier we can get patients involved with behavioral interventions, the better off people with autism will be.” Dr. Powell adds that the model gives researchers insight into mouse brains which share important parallels with brains of living humans, which can only be studied in limited ways with the use of new brain imaging tools.

Source: American College of Neuropsychopharmacology

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