Researchers identify first drug to demonstrate therapeutic effect in a type of autism

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Researchers from Mount Sinai School of Medicine have identified a drug that improves communication between nerve cells in a mouse model of Phelan-McDermid Syndrome (PMS). Behavioral symptoms of PMS fall under the autism spectrum disorder category. The research will be presented Friday at the International Meeting for Autism Research (IMFAR) in Philadelphia.

Previous research has shown that a gene mutation in the brain called SHANK3 can cause absent or severely delayed language abilities, intellectual disability, and autism. Mount Sinai researchers developed mice with a mutant SHANK3 gene and observed a lapse in communication between nerve cells in the brain, which can lead to learning problems. This communication breakdown indicated that the nerve cells were not maturing properly.

The researchers then injected the mice with a derivative of a compound called insulin-like growth factor-1 (IGF1), which is FDA-approved to treat growth failure in children. After two weeks of treatment, nerve cell communication was normal and adaptation of nerve cells to stimulation, a key part of learning and memory, was restored.

"The result of IGF1 treatment of these mice is an exciting development on the road to ultimate therapies for individuals with PMS," said Joseph Buxbaum, PhD, Director of the Seaver Autism Center for Research and

Treatment at Mount Sinai School of Medicine. "If these data are further verified in additional preclinical studies, individuals with a SHANK3 mutation may benefit from treatments with compounds like this one."

Dr. Buxbaum and his team at the Seaver Autism Center will continue to evaluate the efficacy of IGF1 in mice. Patrick Hof, MD, Professor of Neuroscience at Mount Sinai School of Medicine, will specifically evaluate the effects of the compound on neuroanatomical changes. Additionally, Jacqueline Crawley, PhD, Senior Investigator at the National Institutes of Health, will study the effects on behavioral changes in the mice.

Provided by The Mount Sinai Hospital