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Study identifies genetic change in autism-related gene

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A new study from Bradley Hospital has identified a genetic change in a recently identified autism-associated gene, which may provide further insight into the causes of autism. The study, now published online in the *Journal of Medical Genetics*, presents findings that likely represent a definitive clinical marker for some patients' developmental disabilities.

Using whole-exome sequencing – a method that examines the parts of genes that regulate protein, called exons - the team identified a genetic change in a newly recognized autism-associated gene, Activity-Dependent Neuroprotective Protein (ADNP), in a girl with developmental delay. This change in the ADNP gene helps explain the cause of developmental delay in this patient. This same genetic change in ADNP was also found in a boy who was diagnosed with autism.

The ADNP gene plays an important role in regulation of early brain development. Recently, genetic changes in this gene have been found to cause a novel genetic syndrome associated with autism. Changes in this gene may be among the most common causes of autism.

"Genetic testing is a very powerful diagnostic tool for individuals with developmental delay," said Eric Morrow, M.D., Ph.D., director of the Developmental Disorder Genetics Research Program at Bradley Hospital and lead author of the study. "Through genetic testing, which is available to some in the clinical setting as well as in research, a medical diagnosis is possible for a large subset of patients."

Morrow continued, "Genetic changes in ADNP are highly associated with autism and are found in at least .17 percent of autism cases. In these patients, changes in this gene represent an important part of the medical cause for developmental delay and/or autism. The use of these genome-wide sequencing methods in patients with developmental disorders is one of the best examples of the applications of modern genomics in clinical practice."

This study represents one of the first publications resulting in part from Morrow's work with the Rhode Island Collaborative for Autism Research and Treatment (RI-CART), which is co-led by Morrow. Funding for RI-CART is provided in part by a grant from the Simons Foundation for Autism Research and also through support from the Brown Institute for Brain Science (BIBS), the Norman Prince Neuroscience Institute at Rhode Island Hospital, the Department of Psychiatry and Human Behavior at Brown University, Women & Infants Hospital and the Groden Network. This cross-disciplinary collaboration, including the work of Chanika Phornphutkul, M.D., director of Hasbro Children's Hospital's division of Clinical Genetics, and the paper's lead authors from several departments and training programs, represents an important development in research and clinical care for patients.

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