NIH Joined by Advocacy Groups to Fund Research on Autism Susceptibility Genes

Five institutes at the National Institutes of Health (NIH) and three private autism organizations have formed a consortium to pursue their common goal of understanding a devastating disorder. This public-private partnership has funded five grants representing three projects to identify genes that may contribute to the development of autism and Autism Spectrum Disorders. The National Institute of Mental Health will administer the $10.8 million awards over the next five years.

The participating NIH institutes are The National Institute of Child Health and Human Development (NICHD), The National Institute on Deafness and Other Communication Disorders (NIDCD), The National Institute of Environmental Health Sciences (NIEHS), The National Institute of Mental Health (NIMH), and The National Institute of Neurological Disorders and Stroke (NINDS). The voluntary organizations contributing funds are Cure Autism Now (CAN), National Alliance for Autism Research (NAAR), and the Southwest Autism Research & Resource Center (SARRC).

Autism is a neurodevelopmental disorder that causes severe and pervasive impairment in thinking, feeling, language, and the ability to relate to others. In an average year, one to six new cases of autism arise per 1,000 children. Experts believe that as high as 90 percent of the variance in the disorder is due to hereditary factors, and research suggests a strong interaction between environmental factors and multiple unknown genes. As researchers gain a better understanding of the genes responsible for autism, they may be better able to distinguish between the different variants of the disorder and to develop targeted therapies and interventions to treat them.

“This initiative seeks to expand our knowledge of the genetic factors involved in this disorder that affects so many families, said NIMH Director Thomas R. Insel, M.D. “New technologies in gene research can allow scientists to better understand the role genes play in the development of autism, and eventually lead to better treatments.”

Five grants have been awarded to three teams of investigators:

- A three-site collaborative project, involving Rutgers University, the University of Medicine and Dentistry of New Jersey, the Robert Wood Johnson Medical School, and University of Iowa, for a project entitled “Identification and Functional
Assessment of Autism Susceptibility Genes” with investigators Linda Brzustowicz, M.D., James Millonig, Ph.D., and Veronica Vieland, Ph.D., respectively.

- Cold Spring Harbor Laboratory for a project entitled, “Determining the Genetic Basis of Autism by High-Resolution Analysis of Copy Number,” directed by Jonathan Sebat, Ph.D.

- Emory University, for a project entitled, “Identifying Autism Susceptibility Genes by High-Throughput Chip Resequencing,” directed by Michael Zwick, Ph.D.

The three-site collaborative project uses novel statistical methods, fine mapping of candidate regions across the genome, and animal models in the search for autism susceptibility genes.

Cold Spring Harbor Laboratory’s proposal also is highly innovative, capitalizing on the recent discovery of the existence and extent of the high variability of the human genome. Dr. Sebat will examine gene sequence data in order to identify genes and other genomic elements that have either been deleted (micro deletions) or repeated (some many times over). These extensive gene alterations may contribute to the range of Autism Spectrum Disorders.

The Emory project seeks to explore forms of familial inheritance through the X chromosome — because more males than females are diagnosed with autism. Zwick will use a highly innovative technology with great promise for DNA sequencing. It is hoped that this study might lead to insights into autism and a related disorder, Fragile X syndrome.

These new initiatives complement a large autism genetics project funded by NIMH earlier this year and awarded to Johns Hopkins University for research led by Aravinda Chakravarti, Ph.D. This effort employs state-of-the-art gene chips to study variation across the genome in children with autism. This study has already yielded interesting results that point to candidate genomic regions on chromosomes 7, 10, and 19. These signals will provide important clues to the three new genetics projects funded by the consortium.*

These studies would not be possible without the support of the NIMH Human Genetics Initiative** and its large data and sample repository which is available to investigators who study the genetics of autism. It is the largest such facility for mental disorders in the world and collects data and blood samples, makes cell lines, stores DNA and clinical data for autism and other mental disorders.


*For information about similar projects in genetic research on autism, visit http://www.nimh.nih.gov/press/autismgenetics.cfm.

**For more information about the NIMH Human Genetics Initiative, visit www.nimhgenetics.org.

NIMH is one of the 27 components that make up NIH, the Federal Government’s primary
agency for biomedical and behavioral research. NIH is part of the Department of Health and Human Services.

The National Institutes of Health (NIH) — The Nation's Medical Research Agency — includes 27 Institutes and Centers and is a component of the U. S. Department of Health and Human Services. It is the primary Federal agency for conducting and supporting basic, clinical, and translational medical research, and it investigates the causes, treatments, and cures for both common and rare diseases. For more information about NIH and its programs, visit http://www.nih.gov.