THE AUTISM CONSORTIUM

Media Contact:
Kathryn Morris
845-635-9828
kathryn@kmorrispr.com

First Comprehensive Study to Discover the Genetic Causes of Autism

-- The Autism Consortium Brings Together Funding, New Technology, and Experts for Landmark Research to be Conducted with Harvard-MIT's Broad Institute --

BOSTON, MA, AND CAMBRIDGE, MA, AUGUST 15, 2006 – The Autism Consortium, a scientific and clinical collaboration involving 11 Boston area institutions dedicated to research leading to breakthroughs for autism, today announced the initiation of the first comprehensive genetic association study to examine the entire human genome related to autism, The Autism Gene Discovery Project. For the first time ever, the Project brings together new genetic analysis technology, developed by Affymetrix, with the patient samples and experts to conduct a comprehensive research effort to definitively identify the set of genes that cause autism. The project will be initiated this month.

While the clinical evidence is increasingly clear that genes play an important role in autism, [1] [2] [3], definitive links to specific genes contributing to autism have not been fully elucidated. Family studies have indicated a strong genetic contribution to autism, including observations that siblings of autistic children are 10 to 60 times more likely to be autistic, and studies of twins showing that in identical twins there is about a 75% rate of both twins having autism [1] [2] [3] . The inheritance pattern is complex and suggests that a number of genes are involved, but studies to date have not been large enough to discover the specific genes that produce susceptibility to the disorder.

"This landmark project will lay the groundwork for understanding the molecular mechanism(s) and disease pathway(s) that underlie autism, something that is desperately needed to accelerate research for the growing number of children and families affected by this devastating disorder," said Chris Walsh MD, at Children's Hospital Boston, and Chair of the Autism Consortium's Executive Committee of Researchers and Clinicians. "Once the molecular mechanism and disease pathways are better understood, further research can be pinpointed to improve screening and predictive tests for autism, better diagnosis of autism, and gene targets to be used to develop treatments for autism."

The Autism Gene Discovery Project brings together new genetic analysis technology, (one of) the largest collections of biological samples of autistic children, and experts to conduct the research, including Dr. Rudy Tanzi of the Massachusetts General Hospital

who also serves on the CAN Scientific Advisory Board and the AGRE Steering Committee, Dr. Chris Walsh of Children's Hospital Boston and Drs. Mark Daly and James Gusella of MGH Center for Human Genetic Research and the Broad Institute.

- The new analytic technology was selected from Affymetrix, the GeneChip® Human Mapping 500K Array, which now enables the comprehensive measurement of polymorphisms and gene copy number in DNA samples of children with autism.
- The Study will use 3,700 samples provided by the Autism Genetic Resource Exchange (AGRE), a DNA repository and family registry for autism sponsored by the Cure Autism Now Foundation.
- Experts from the Autism Consortium will conduct the research in collaboration with the Broad Institute of MIT and Harvard.

The Project will begin within a month, and the Autism Consortium hopes to have data available from the Autism Gene Discovery Project within six months to contribute to the field of genetic research related to autism. The database of results will be made publicly available to anyone conducting research in the field of autism.

"The Autism Gene Discovery Project embodies the mission of the Autism Consortium, as we aim to bring together world-recognized leaders in science and clinical medicine to conduct collaborative research focused on breakthroughs in autism," said Peter Barrett, Chairman of the Board of Directors of the Autism Consortium. "This project demonstrates the advantages of the Autism Consortium. We can integrate cutting edge science and leading clinical medicine critical to solving the full complexity of this disorder. Through this project and other initiatives, the participating members of the Autism Consortium are committed to interdisciplinary teamwork and inter-institutional collaboration with the goal of seeing our discoveries translated into improved treatments for children with autism in the very near future. We know speed matters."

The Broad Institute of Harvard and MIT will be performing the genotyping and join scientists from the Autism Consortium to perform genetic analyses. "We are thrilled to be working with the Autism Consortium to uncover the genetic contributions to this devastating disease, "said Dr. Mark Daly, of Massachusetts General Hospital's Center for Human Genetic Research and Broad's Program in Medical and Population Genetics. "This study, along with a smaller ongoing effort at Johns Hopkins led by Dr. Aravinda Chakravarti using the same technology, will provide dramatic new opportunities for gene discovery by creating a reference data set for the entire research community."

About Autism

Autism is the fastest growing disability in the United States. A child is diagnosed with autism every 21 minutes and it now affects one in every 166 children in the US. Autism spectrum disorders (ASDs) are a group of developmental disabilities defined by significant impairments in social interaction and communication and the presence of unusual behaviors and interests. Many people with ASDs also have unusual ways of

learning, paying attention, or reacting to different sensations. Since the diagnosis of ASD relies on the description of behavior, there is no straightforward clinical diagnosis or simple blood test. ASD begins before the age of 3 and lasts throughout a person's life. It occurs in all racial, ethnic, and socioeconomic groups and is four times more likely to occur in boys than girls.

About the Autism Consortium

The Autism Consortium is dedicated to facilitating the advancement, understanding and treatment of Autism and related developmental brain disorders. The Autism Consortium brings together scientists and clinicians interested in developmental brain disorders at Beth Israel Deaconess Medical Center, Boston University and Boston University Medical Center, the Broad Institute of MIT and Harvard, Cambridge Health Alliance, Children's Hospital Boston, Harvard University and Harvard Medical School, Massachusetts General Hospital, Massachusetts Institute of Technology, and Tufts-New England Medical Center-Floating Hospital for Children.

The Autism Consortium serves as an umbrella entity that facilitates, directs and funds the collaborative work conducted by its member scientists and clinicians to advance research on autism.

In addition to Drs Daly, Gusella, Tanzi and Walsh, other Autism Consortium investigators involved in autism gene discovery include Drs. Louis Kunkel and Isaac Kohane of the Children's Hospital Boston and David Pauls, Lars Bertram and Susan Santangelo of the Massachusetts General Hospital.

For further information about the Broad Institute, go to http://www.broad.mit.edu.

1. Folstein S and Rutter M (1977) Infantile autism: a genetics study of 21 twin pairs. J. Child Psychol Psychiatry 18:297-321,
2. Baily et al. (1995) Autism as a strong genetic disorder: evidence from a British twin study. Psychol Med 25:63-77,
3. Lauritsen M and Ewald H (2001) The genetics of autism. Acta Psychiatr Scand 103:411-427.