

# Autism Consortium releases data on genes involved in autism to researchers worldwide

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The Autism Consortium, a group of researchers, clinicians and families dedicated to radically accelerating research and enhancing clinical care for autism, announced today that it has completed the first genome scan for Autism Spectrum Disorders (ASD) through its Autism Gene Discovery Project and has released the reference data set to a database that autism researchers around the world can use. The scan was conducted using new, high resolution technology developed by Affymetrix on genetic data from more than 3,000 children with ASD and their families.

“Today’s release of genetic and phenotypic data on autism marks a significant achievement for the autism research community,” said Thomas Insel, Ph.D., Director of the National Institute for Mental Health. “Progress in finding the causes and cures for autism spectrum disorders rests in large part on improving the rapid access and sharing of data and resources. That the Consortium is making the data available to the scientific community even before its own researchers have fully analyzed the information, demonstrates their high degree of commitment to and leadership in advancing autism research.”

Along with complementary data generated by Dr. Aravinda Chakravarti at Johns Hopkins and provided to the NIMH this week, these data provide the most detailed look to date at the genetic variation patterns in families with autism.

Mark Daly, PhD, a Consortium member with the Center for Human

Genetic Research at Massachusetts General Hospital and a senior associate member of the Broad Institute of Massachusetts Institute of Technology and Harvard, released genetic data from genome wide scans of DNA variation collected from 3,000 individuals who are either affected by autism spectrum disorders, or are family members of individuals with autism. DNA samples for this scan were provided by the Autism Genetic Resource Exchange (AGRE), a program of Autism Speaks, dedicated to accelerating the pace of autism research.

“We’re releasing raw genotype data so that other qualified researchers can take a look at it even as we’re still beginning our own analysis,” Daly said. “Autism Spectrum Disorders are extremely complex and only through collaboration with researchers with many specialized areas of expertise will we gain an understanding of what makes some children susceptible. That’s why we have been committed to providing the data to the research community as fast as we can. The new data will be deposited in the gene bank maintained by AGRE, which, in turn, will make the data available to qualified researchers.”

“It is really something of a landmark to have both data from his laboratory and mine available to autism researchers at virtually the same time,” said Dr. Chakravarti, who collaborated with Dr. Daly for many years. “We will each look carefully at the other’s findings as we continue to search for definitive information about which genes are important in causing autism spectrum disorders.”

The prepublication release of such a significant trove of data is a dramatic departure from the traditional less open culture of research science and a landmark achievement for the Consortium and its Autism Gene Discovery Project, the first comprehensive genetic association study to examine the entire human genome related to autism.

The number of individuals diagnosed with autism spectrum disorders has

significantly increased in recent years. Although there is some uncertainty about the role that better diagnosis, greater recognition of the disorders, and biological and environmental factors play; there is growing agreement in the research community that genes have a significant role in autism spectrum disorders. The release of the data from this screen is a significant step toward identifying the genes involved in ASD.

Researchers in the Autism Gene Discovery Project conducted the genome wide study using GeneChip® microarray technology made by Affymetrix Inc, of Santa Clara, California, in their search for autism spectrum disorder-related genes. The Affymetrix 500K Array offers a comprehensive view of the genome, enabling researchers to analyze 500,000 markers simultaneously and perform whole-genome analyses in large populations. The Affymetrix microarray chips were processed by researchers in the Broad Institute's Genetic Analysis Platform.

These tools enable researchers to search across the entire genome for areas of deletions or duplications of genetic material or for single nucleotide polymorphisms (SNPs), small changes in the DNA sequence. Daly, a statistical geneticist, and his colleagues wrote two mathematical algorithms to analyze the genome scan for specific genes involved in autism spectrum disorders, or for the absence of genes that ought to be present. Among other things, the Autism Consortium's Gene Discovery Group found what they believe may be important new information on the genes involved in autism spectrum disorders. The next step is to conduct an association analysis to determine the role that the genetic variations identified in the scan play in autism spectrum disorders.

According to Daly, the speed at which he collected genome wide data from such a large sample could not have been accomplished without the funding or the collaboration from the Autism Consortium, which contributed the \$1 million needed to complete the project saving the

researchers months or years seeking grants. The grant was made possible through a gift from the Anne and Paul Marcus Family Foundation which provided the funds for the Gene Association Study as well as for the purchase of the entire set of samples from AGRE.

“Autism spectrum disorders present major challenges for families, communities, and the health care system,” said Peter Barrett, president of the Autism Consortium. “Our job is to accelerate new ways to diagnose and treat autism spectrum disorders by breaking down barriers. In this case, we are working to foster collaboration between scientists from multiple institutions. Autism spectrum disorders are the focus here, not any single scientific group’s accomplishment. What we've shown within a year is that when people work together toward a common goal, we can speed up our understanding of these disorders and move towards better ways to help individuals with autism spectrum disorders and their families.”

Source: The Autism Consortium

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