

## **What is the NAAR Autism Genome Project?**

The **NAAR Autism Genome Project** is a large-scale, collaborative genetics research project designed to map the human genome in the search for *autism susceptibility genes* – the genes responsible for the inherited risk for autism. This unprecedented endeavor is the largest research collaboration ever to focus on the genetics of autism and includes more than 170 of the world's leading genetic researchers from over 50 academic and research institutions throughout the United States, Canada, the United Kingdom, France, Sweden, Denmark and Germany. The project includes approximately 1,200 multiplex families (two children with autism spectrum disorders and their parents) from all over the world who are directly affected by autism spectrum disorders.

While autism was first identified more than 50 years ago, the disorder remains a medical mystery because the tools necessary to learn more about the genetics of autism simply did not exist. With the development of promising new technology and 6,000 genetic samples, the research “dream team” of the *NAAR Autism Genome Project* hopes to reveal genetic information about autism that was previously undetectable. Using the data that they find, scientists will identify those genes that demonstrate a likelihood of being linked to autism. These discoveries will ultimately allow the medical research community to develop medical treatments for those affected by the disorder on a case-by-case basis.

This project required unprecedented collaboration between the top autism researchers in North America and Europe. It is a credit to all the research organizations involved and also to NAAR's vision and coordination of efforts that this is now possible.

### **How will this study be conducted?**

- In mid-July, 6,000 genetic samples (four from each family) will be taken from 1,500 families that have two children with autism spectrum disorders and sent to the TGen for genotyping
- TGen will genotype each patient sample using Affymetrix' GeneChip® Mapping 10K Array, yielding over 10,000 genome-wide SNPs per patient
- The Affymetrix Mapping 10K Array will allow researchers to examine mutations in a person's DNA in finer detail than was previously possible
- In early fall, The SNPs will be analyzed by NAAR associated investigators
- Further studies will be conducted to identify related genes on a base by base level
- Ultimately, researchers hope to develop a diagnostic tool, followed by knowledge-based drug development

### **Why couldn't this be done before?**

The technology simply didn't exist until just under a year ago. Researchers were unable to analyze DNA at a sufficiently high resolution that would enable them to see the mutations that are at least in part responsible for autism.

Everything changed with the introduction of the Affymetrix GeneChip® Mapping 10K Array. It finally gave researchers the ability to see these tiny little genetic changes that will ultimately help them figure out what happens to the DNA of those affected with autism.

### **How did this project get started?**

The origins of the *NAAR Autism Genome Project* can be traced to 2000, with NAAR's initial support of the Autism Genetics Cooperative, a research consortium, which provided a forum for the researchers to meet and discuss unpublished data. NAAR and the Nancy Lurie Marks Family

Foundation co-funded this effort through 2003. In 2002, NAAR began working to bring top autism researchers together to find the genes that cause autism. Over the past two years, additional research consortiums joined NAAR's initiative to determine the genetic underpinnings of autism. In 2003, the National Institutes of Health (NIH) committed its support to the project. Later that year, NAAR and the NIH officially unveiled the *NAAR Autism Genome Project* at the Autism Summit Conference in Washington, D.C.

**Who is part of the NAAR Autism Genome Project?**

The project is a partnership between NAAR and four institutes of the National Institutes of Health (NIH): the National Institute of Mental Health (NIMH), National Institute of Child Health & Human Development (NICHD), National Institute of Neurological Disorders & Stroke (NINDS), and National Institute of Deafness and Other Communication Disorders (NIDCD). At the core of the *NAAR Autism Genome Project* are the investigators bringing it to life – a “collaboration of collaborations” composed of four main research teams:

- the Autism Genetics Cooperative (AGC)
- the International Molecular Genetic Study of Autism Consortium (IMGSAC)
- the Collaborative Programs of Excellence (CPEA)
- the Autism Genetics Resource Exchange (AGRE).