



NAAR Autism Genome Project: Fact Sheet

A Public/Private Research Partnership Uniting the World's Leading Autism Researchers to Locate the Genes Associated with Autism Spectrum Disorders

Who:

The project is a partnership between the National Alliance for Autism Research (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). Initially, the institutes have collectively committed \$2.5 million towards this project and NAAR, which began co-funding one of the largest autism genetics consortiums with the Nancy Lurie Marks Family Foundation in 2000, has committed \$2 million.

What:

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 about 170 of the world's leading genetic researchers from approximately 50 academic and research institutions focusing on autism and approximately 1,500 multiplex families (two children with autism spectrum disorders and their parents) from all over the world who are directly affected by autism spectrum disorders.

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); and the Autism Genetics Resource Exchange (AGRE).

Locating the genes believed to be involved with causing autism will not be easy. There are approximately 30,000 genes in the 46 chromosomes that make up the human genome. Autism is known as a complex disorder that is believed to involve many genes on several chromosomes.

To determine the genetic cause or causes of autism spectrum disorders, the **NAAR Autism Genome Project** aims to ultimately build a “map” of these genomic neighborhoods. This map will be used to determine which genes are causing specific *phenotypes* that are associated with autism.

The first phase of the project currently includes the completion of the largest genome screen for autism to date. The **NAAR Autism Genome Project** will conduct two genome scans: one based on DNA microarray technology and one based on microsatellite technology. A genome screen is a genetic analysis designed to identify intervals in the human genome that show the highest priority for further investigation. Intervals are like

“genomic neighborhoods” that appear to be the most likely places where the genes believed to be involved with autism exist on the genome.

The genome screen and follow-up genetic analysis will involve the use of *single nucleotide polymorphisms* (SNP) markers that are found across the human genome. SNPs (pronounced “snips”) are actually the difference between genetic sequences in two individuals. SNPs generally occur in human DNA at a frequency of one every 1,000 bases can be used to track inheritance in families. For this initiative, the SNP markers will help researchers more easily identify meaningful differences in each sample after the screen is completed.

To date, there have been several genetic analyses completed for autism, however results have varied and none have been performed on such a large scale as the **NAAR Autism Genome Project**. This initiative's unprecedented convergence of the largest sample set ever assembled, a consortium of leading researchers in the field and cutting edge genetics technologies offers the autism community the best opportunity to date to elucidate the genetic underpinnings of this devastating disorder.

Why:

Autism is largely considered the most heritable neuropsychiatric disorder. Locating the genes responsible for autism will likely enable researchers for the first time to gain a thorough understanding of what causes autism. Having a detailed understanding of the cause will better enable doctors to develop specific, more targeted medical treatments, preventions or a cure.

In addition, autism is a disorder with symptoms that are associated with other brain-based conditions. Determining the genetic causes of autism spectrum disorders has the potential to yield significant advances for other disorders involved in language & communication and behavioral sciences, such as attention deficit disorder, hyperactivity, speech impairments, fragile X, Turner Syndrome and some forms of mental retardation.

*Public/private collaborations between NAAR and the NIH focus on “team research” and “consortium building”— a major theme of the recently announced **NIH Roadmap Initiative for Medical Research** – and may become a research funding model for other disorders and diseases. NAAR provided the initial infrastructure and support that fostered a collaborative atmosphere to develop the NAAR Autism Genome Project. The NIH has now brought its support and expertise to the partnership, which has elevated the project to the next stage, illustrating a unique example of how the public and private sector can work together to enhance investments in medical research.*

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