Medical Xpress

Researchers Identify Autism Gene

March 13 2007

Yale School of Medicine autism experts Fred Volkmar, M.D. and Ami Klin are part of a global research consortium from 19 countries to identify a gene and a region of a chromosome that may lead to autism in children.

The findings are published online today in *Nature Genetics* and also will be published in the journal's March print edition. They are based on the largest-ever autism genome scan. Over 120 scientists from over 50 institutions who formed the Autism Genome Project (AGP) performed the research. The AGP began in 2002 when researchers from around the world decided to collaborate and share their samples, data and expertise to aid in identifying autism susceptibility genes.

Funded by Autism Speaks, a national non-profit dedicated to increasing awareness of autism and raising money to research the disorder, and the National Institutes of Health, these are the preliminary findings from the AGP's first phase.

The consortium used "gene chip" technology to look for genetic similarities in autistic individuals culled from almost 1,200 families. They also scanned the DNA to search for copy number variations, which are submicroscopic insertions and deletions of genetic material that scientists believe may be linked to autism and other diseases. The researchers found neurexin 1, part of a family of genes that plays a role with the neurotransmitter glutamate, which has been previously linked to autism. They also found a gene on chromosome 11 that may be linked to autism susceptibility. That gene has not yet been pinpointed.

Researchers speculate that there may be fiver or six major genes and as many as 30 other genes involved in autism. If a child has more of these genes, there is a higher chance of being born with autism or a more severe form of the disease.

Autism is a complex brain disorder that inhibits a person's ability to communicate and develop social relationships, and is often accompanied by extreme behavioral challenges. Autism Spectrum Disorders are diagnosed in one in 150 children in the United States, affecting four times as many boys as girls. The diagnosis of autism has increased tenfold in the last decade.

Phase Two of the Autism Genome Project was also announced to continue the effort to discover the genes that cause the disorder. This second phase represents a $14.5 million, three-year investment by Autism Speaks, the British Medical Research Council, the Health Research Board of Ireland, Genome Canada and its partners, Canadian Institutes for Health Research, Southwest Autism Research and Resource Center, and the Hilibrand Foundation.

Source: Yale University