New autism susceptibility genes identified

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Mount Sinai researchers and the Autism Genome Project Consortium (AGP) announced today that they have identified new autism susceptibility genes that may lead to the development of new treatment approaches. These genes, which include SHANK2, SYNGAP1, DLGAP2 and the X-linked DDX53PTCHD1 locus, primarily belong to synapse-related pathways, while others are involved in cellular proliferation, projection and motility, and intracellular signaling. The findings were published today in *Nature* by researchers at the Seaver Autism Center for Research and Treatment at Mount Sinai School of Medicine, together with an international consortium of researchers who make up AGP.

"As we continue to uncover genetic mutations that can cause autism, we are gaining further insights that will ultimately lead to earlier diagnosis and better treatments," said Joseph Buxbaum, PhD, Director of the Seaver Autism Center and Professor of Psychiatry, Neuroscience and Genetics and Genomic Sciences at the Mount Sinai School of Medicine.

The study results are based on analysis of high-density genotyping data collected from 1,000 individuals with autism spectrum disorder (ASD) and 1,300 without ASD. These findings further support an emerging consensus within the scientific community that autism is caused in part by many "rare variants," or genetic changes found in less than one percent of the population.

While each of these variants may only account for a small fraction of the cases, collectively they are starting to account for a greater percentage of individuals with autism. They are also providing insights into possible common pathological mechanisms.

Findings show that the DNA of individuals with ASDs has more copy number variants (CNVs) ? rare submicroscopic insertions and deletions ? disrupting genes, including genes previously reported to be associated with autism, but also other genes such as those involved in intellectual disabilities. The overlap between autism susceptibility genes and genes previously implicated in intellectual disabilities further supports the hypothesis that at least some genetic risk factors are shared by different psychiatric developmental disabilities. Finally, identification of these biological pathways points to new avenues of scientific investigation, as well as potential targets for the development of novel treatments.

"It is an exciting development to see Dr. Buxbaum and colleagues identify genes that have been linked to intellectual disabilities but not previously implicated in autism now be linked to that condition as well," commented Bruce D. Gelb, MD, Director of the Child Health and Development Institute at Mount Sinai. Dr. Gelb, who was not affiliated with this study, said further, "This landmark study also provides a template for future research into the genetics of many other important human

disorders."

SOURCE [1]

Links:
[1] http://www.eurekalert.org/pub_releases/2010-06/tmsh-nas060810.php