

From: **AUTISM SPEAKS**

Dana Marnane – 646-408-5749; dmarnane@autismspeaks.org

 Jane Rubinstein – 212-843-8287/516-993-0708; jrubinstein@rubenstein.com

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NEW AUTISM GENES DISCOVERED:

AUTISM SPEAKS AND THE WORLD’S LEADING AUTISM EXPERTS ANNOUNCE PHASE 2 RESULTS OF THE

AUTISM GENOME PROJECT

*Phase 2 Results of the Autism Genome Project*

*Published in* Nature

NEW YORK, N.Y. (June 9, 2010) – Autism Speaks, the world’s largest autism science and advocacy organization, and an international consortium of researchers, along with participating families, joined together to announce new autism genetic discoveries from the second phase of its collaborative study: the Autism Genome Project. The results were published today in the journal *Nature,* one of the world’s most respected peer-reviewed scientific publications.

The [Autism Genome Project](http://www.autismgenome.org/) (AGP) ([www.autismgenome.org](http://www.autismgenome.org)) is an international autism genetics research consortium co-funded by Autism Speaks, the Medical Research Council, Canadian Institutes of Health Research, Health Research Board (Ireland), Genome Canada, the Hilibrand Foundation and Autistica. Based on analysis of high-density genotyping data collected from 1,000 individuals with autism spectrum disorder (ASD) and 1,300 without ASD, the AGP reported that individuals with autism tend to carry more submicroscopic insertions and deletions called copy number variants (CNV) in their genome than controls. Some of these CNV appeared to be inherited, while others are considered de novo, or new, because they are found only in affected offspring and not in the parents. Taken together, more of the CNVs disrupt genes, previously reported to be implicated in intellectual disability without autism or in autism, than expected by chance.

The new AGP study also identified new autism susceptibility genes including SHANK2, SYNGAP1, DLGAP2 and the X-linked DDX53–PTCHD1 locus. Some of these genes belong to synapse-related pathways, while others are involved in cellular proliferation, projection and motility, and intracellular signaling, functional targets that may lead to the development of new treatment approaches.

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 in the autism community, as well as providing insights into possible common pathogenic mechanisms. 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.

“Piece by piece, we are discovering genetic mutations that can cause autism. These findings will provide answers for families about what contributed to their autism,” said Andy Shih, Ph.D., Autism Speaks vice president for scientific affairs. “Furthermore, as we have learned from examples involving other genetic risk factors of autism (e.g., Fragile X, Rett, TSC), these genetic findings help us understand the underlying biology of autism, which can lead to the development of novel treatments.”

The AGP consists of 120 scientists from more than 60 institutions representing 11 countries who formed a first-of-its-kind autism genetics consortium. The AGP began in 2002 when researchers from around the world decided to come together and share their samples, data, and expertise to facilitate the identification of autism susceptibility genes. This continuing collaboration and its unique scientific assets (e.g., large sample set and multidisciplinary expertise) created scientific opportunities that otherwise would not exist. The AGP is well positioned to build on these extraordinary assets as the field of autism genetics further investigates rare variants, requiring larger sample sets to identify more CNV. Additional support for Phase 2 of the AGP was provided by the National Institutes of Health. The first phase of the AGP, the assembly of the largest-ever autism DNA collection and whole genome linkage scan, was funded by Autism Speaks and the National Institutes of Health and completed in 2007.

**About Autism**

Autism is a complex neurobiological disorder that inhibits a person's ability to communicate and develop social relationships, and is often accompanied by behavioral challenges. Autism spectrum disorders are diagnosed in one in 110 children in the United States, affecting four times as many boys as girls. The prevalence of autism increased 57 percent from 2002 to 2006. The Centers for Disease Control and Prevention have called autism a national public health crisis whose cause and cure remain unknown.

**About Autism Speaks**

Autism Speaks is North America’s largest autism science and advocacy organization. Since its inception in 2005, Autism Speaks has made enormous strides, committing over $142.5 million to research through 2014 and developing innovative new resources for families. The organization is dedicated to funding research into the causes, prevention, treatments and a cure for autism; increasing awareness of autism spectrum disorders; and advocating for the needs of individuals with autism and their families. In addition to funding research, Autism Speaks also supports the Autism Treatment Network, Autism Genetic Resource Exchange and several other scientific and clinical programs. Notable awareness initiatives include the establishment of the annual United Nations-sanctioned World Autism Awareness Day on April 2 and an award-winning “Learn the Signs” campaign with the Ad Council which has received over $210 million in donated media. Autism Speaks’ family resources include the Autism Video Glossary, a 100 Day Kit for newly-diagnosed families, a School Community Tool Kit and a community grant program. Autism Speaks has played a critical role in securing federal legislation to advance the government’s response to autism, and has successfully advocated for insurance reform to cover behavioral treatments. Each year *Walk Now for Autism Speaks* events are held in more than 80 cities across North America. To learn more about Autism Speaks, please visit [www.autismspeaks.org](http://www.autismspeaks.org).

**About the Co-Founders**

Autism Speaks was founded in February 2005 by Suzanne and Bob Wright, the grandparents of a child with autism. Bob Wright is Senior Advisor at Lee Equity Partners, Chairman and CEO of the Palm Beach Civic Association and served as vice chairman, General Electric, and chief executive officer of NBC and NBC Universal for more than twenty years. He also serves on the boards of the Polo Ralph Lauren Corporation, RAND Corporation and the New York Presbyterian Hospital. Suzanne Wright has an extensive history of active involvement in community and philanthropic endeavors, mostly directed toward helping children. She serves on the boards of several non-profit organizations and is also Trustee Emeritus of Sarah Lawrence College, her alma mater. Suzanne has received numerous awards such as the CHILD Magazine Children’s Champions Award, Luella Bennack Volunteer Award, Spirit of Achievement award by the Albert Einstein College of Medicine's National Women’s Division and the Weizmann Institute of Science. In 2008, the Wrights were named to the *Time* 100 list of the most influential people in the world for their commitment to global autism advocacy.

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