MedicalXpress

# Autism Genome Project delivers genetic discovery

April 24 2014

A new study from investigators with the Autism Genome Project, the world's largest research project on identifying genes associated with risk for autism, has found that the comprehensive use of copy number variant (CNV) genetic testing offers an important tool in individualized diagnosis and treatment of autism.

Funded primarily by Autism Speaks, the world's leading autism science and advocacy organization, the Autism Genome Project involved more than 50 research centers in 11 countries. The report, published today in the *American Journal of Human Genetics*, delivers on the 10-year project's objective to provide practical methods for earlier diagnosis and personalized treatment of autism.

"With the publication of this study, we should step back to recognize and celebrate the pioneering achievements of the AGP and what they have accomplished in helping to launch the field of genomic risk discovery in autism," says Autism Speaks Chief Science Officer Rob Ring. "The AGP has generated information that holds the potential to guide medical care for certain individuals with autism today. They have demonstrated that science can work for families, and Autism Speaks is proud to have been a supporter of the work all along the way."

The study involved CNV testing of 2,446 families affected by autism and 4,768 individuals unaffected by neurologic or psychiatric disorders. Overall, CNVs were significantly more common in the participating families affected by autism. And, the CNV testing uncovered dozens of

cases where autism-linked gene changes were associated with additional health risks warranting medical attention.

In nine of the families affected by autism, CNVs involved a gene that indicates elevated risk for seizures and epilepsy. "This result warrants an immediate referral to a neurologist," explains senior author Stephen Scherer of the Toronto's Hospital for Sick Children and the University of Toronto. Similarly, CNV testing indicated a high risk for muscular dystrophy in several of the autism families and identified syndromes associated with heart problems in others.

CNVs are genetic changes that involve duplication or deletion of entire segments of DNA. They do not typically show up on standard genetic tests which search for "spelling mistakes" in the DNA letters that compose a gene. Those standard tests identify a clear genetic autism link in only 15 to 20 percent of the cases.

"This report and its extensive supplements should become a new guidebook for medical geneticists working with families affected by autism," Dr. Scherer says.

In addition, the study added dozens of genes to the growing list of those that contribute to the development of autism. Surprisingly, the autism genes identified through CNV testing had little overlap with those detected using standard exome gene sequencing, yet researchers say they affect the same brain pathways.

"These gene discoveries will help guide further research on autism subtypes and their treatment," Dr. Scherer says.

In response, the investigators urge medical geneticists to add CNV testing to the standard gene tests for autism and to consult medical recommendations for the many autism-linked syndromes that CNV

testing can reveal. CNV testing is currently available, though it's not typically part of standard genetic testing for autism. Whole genome sequencing is the next step in genetic testing for Autism but is not yet widely available in ordinary medical settings. Autism Speaks has taken a world-leadership position in in this direction with its Autism Ten Thousand Genomes (Aut10K) program.

Provided by Autism Speaks

Citation: Autism Genome Project delivers genetic discovery (2014, April 24) retrieved 2 May 2024 from https://medicalxpress.com/news/2014-04-autism-genome-genetic-discovery.html

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.