Whole genome sequencing opens a new way for the diagnosis and medical therapy for autism

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An international consortium, consisting of Autism Speaks, Duke University School of Medicine, The Hospital for Sick Children of Toronto, BGI and other institutes, has investigated the genetic variants in 32 families with Autism Spectrum Disorder (ASD). The results show that whole genome sequencing and analysis promise great value to identify de novo or rare inherited mutations that give rise to autism in ASD groups. The findings were published online today in *American Journal of Human Genetics*.

Autism is the fastest growing developmental disorder in the United States, or even the whole world. It is estimated that 1 in every 88 children in the United States is diagnosed with autism, which is greater than the prevalence of pediatric AIDS, cancer, and diabetes combined. It can rob the individual of typical development from childhood to adolescence to adulthood, and bring huge burden on the families.

The discoveries of genetic mutations can substantially increase people's understanding of the underlying biology of autism. In this study, researchers surveyed all the risk mutations in ASD patient groups by whole genome sequencing (WGS), and try to fully describe the genetic architecture of autism. The results may give critical insight into the molecular and cellular processes that may be preferentially targeted for disruption by genetic lesions in autism patients.

The study shows that the proportions of deleterious de novo mutations and X-linked or autosomal inherited alterations are higher than the previous reports with 19% and 31%, respectively. Researchers speculated the partial reason maybe the more comprehensive and uniform coverage afforded by WGS. Compared to exome sequencing technology, WGS also shows great advantages in efficiency and accuracy.

Researchers also identified the deleterious mutations variants in 4 novel, 9 known, and 8 candidate autism risk genes, including CAPRIN1 and AFF2 (both linked to FMR1 involved in fragile X syndrome), VIP (involved in social-cognitive deficits), and other genes such as SCN2A and KCNQ2 (also linked to epilepsy), NRXN1, and CHD7, which causes ASD-associated CHARGE syndrome.

"From diagnosis to treatment to prevention, whole genome sequencing efforts like these hold the potential to fundamentally transform the future of medical care for people with autism," stated Autism Speaks Chief Science Officer and study co-author Robert Ring, Ph.D.

Yingrui Li, CEO of BGI Tech, one of BGI's affiliates, said "Whole-genome sequencing may serve as a powerful tool to advance new effective treatments to improve the lives of individuals and families with autism. Early diagnosis is important for autism, which can help a child with autism make significant gains in language and social skills."

Provided by BGI Shenzhen