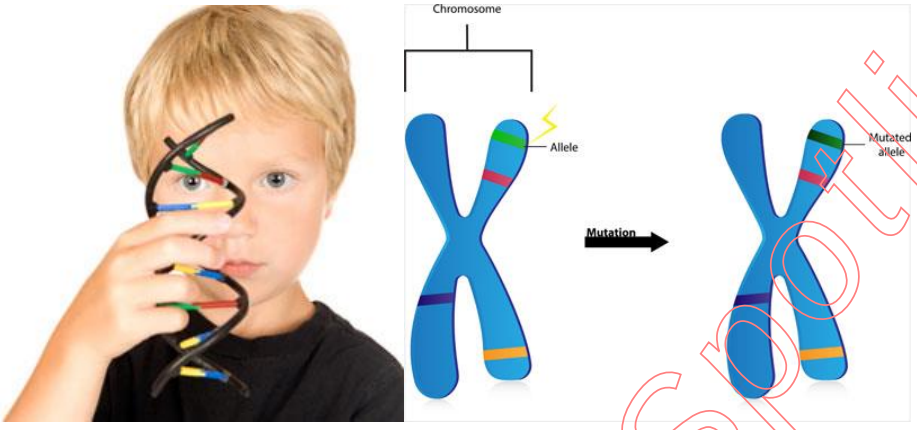
Genetic Science Spotlight

ASD: Power of Coverage from Genes to Exome to Genome, by The Hospital for Sick Children, Toronto



Risk factors associated with Autism Spectrum Disorder (ASD) susceptibility include genetic, environmental, and gene-environment (GxE) interactions. About 5-10% of cases of classic autism are due to an identifiable genetic disorder, which in many cases, has a known inheritance pattern and a specific recurrence risk. Identified genetic causes of autism can be classified as the cytogenetically visible chromosomal abnormalities (~5%), copy number variants (CNVs) (i.e., submicroscopic deletions and duplications) (10–20%), and single-gene disorders (~5%). Deletion, duplication and/or copy number variation (CNV) events in the region of chromosome 16p11.2 were observed in nearly 1% of multiplex families with autism and in more than 1.5% of clinical samples from subjects with developmental delay. These events are either as frequent as or more frequent than the most common known cause of autism. Nevertheless, a recent study led by The Hospital for Sick Children in Toronto, Canada, mentioned that discordant mutations were found in a marked portion, 69.4% of quartet ASD families with parents and 2 ASD siblings, when Whole Genome Sequencing (WGS) was employed. Comparing with Whole Exome Sequencing (WES), WGS data reveal evidence of association between CNV in non-coding regions and autism susceptibility. The authors also stressed the necessity of a full assessment of each individual's genome when determining the role of genetic factors in risk- or health-management strategies. Among the exome and/or genome sequencing data, the "many genes, common pathways" hypothesis may be the key to ASD pathogenesis, and a probable therapeutic target against the disorder.

Miles, J.H. (2011). Autism spectrum disorders—A genetics review. Genetics in Medicine 13, 278–294. doi:10.1097/GIM.0b013e3181ff67ba
Yuen, R.K.C., et al. (2015). Whole-genome sequencing of quartet families with autism spectrum disorder. NATURE MEDICINE 21, 185–191. doi:10.1038/nm.3792

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