Perspective
Volume 9:1,2021
DOI: 10.37421/jcmg.2021.9.168

Journal of Clinical & Medical Genomics

ISSN: 2472-128X **Open Access**

Repetitive DNA-Potential Autism Genes

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Repetitive DNA is called as tandem repeats. A link has been found between repetitive DNA and autism spectrum disorder. Above 50 diseases are known to be caused by repeat sequence in a single gene. But the role of tandem repeats in polygenic diseases, have more-complex genetics. A new approach of tandem repeats associated with one condition, autism spectrum disorder.

Diagnosis of ASD (Autism spectrum disorder): (1) persistent deficits in social communication and social interaction across several contexts; and (2) restricted, repetitive patterns of behavior, interests, or activities. Monozygotic twins were more likely to share a diagnosis than dizygotic twins. However, this is only for 10–20% of ASD cases, and patients with similar pathogenic variants may be diagnosed on different levels of the disorder.

Autism spectrum disorder (ASD) is highly prevalent. ASD is genetically heterogeneous and may be caused by both de novo gene and inheritable variations. It is characterized by communication deficits, atypical neurodevelopment, atypical social functioning, repetitive behaviours and restricted interests. Understanding of ASD mechanisms is very crucial for both comprehending ASD and for developing novel therapies. Initially it was assumed to be environmental origin, and improved understanding of the role of genetics in human health. The risk of a child with ASD found to be proportional to the percentage of the genome shared with sibling or parent who is affected.

Tandem repeats genome-wide has been optimized from past few years. Studies found that tandem repeats collectively contribute to autism disorder; none of the genes are associated individually with the condition. The studies are not able to explain the complete mechanisms by which tandem repeats which contribute to autism disorder. Several expansions occurred in DNA that drives to the expression of genes which involves in the foetal brain development. Many of the tandem repeats are highly responsive to the environmental signal which leads to the altered epigenetic modifications.

More broadly, the repeatome, and tandem repeats, in particularly should now be studied systematically across the range of common human disorders, including diabetes, cancer, and brain disorders like depression and schizophrenia. Genome-wide association studies (GWAS), link the genetic variants to traits and disorders, and improved understandings of polygenic disorders, but the substantial gaps remain. However, to investigate at gene-level resolution, early studies are used to the candidate approach. Sequencing technology quickly confirmed that etiology of Autism Spectrum Disorder was multigenic and highly heterogeneous, with few of pathogenic variants present in a significant percentage of afflicted individuals. It is known that average case is a product of many susceptibility-increasing variations.

How to cite this article: Bondada, Anusharadha . "Repetitive DNA-Potential Autism Genes". J Clin Med Genomics 9 (2021) doi: 10.37421/jcmg.2021.9.168

Received: *January 06, 2021;* ***Accepted:*** *January 21, 2021;* ***Published:*** *January 28, 2021*