Unraveling the Genetic Basis of Neurodevelopmental Disorders.

Paula Arrifano*

Department of Molecular Biology, Federal University of Para, Belem, Brazil

Autism spectrum disorder (ASD) is a complex neurodevelopmental disorder that affects communication, social interaction, and behavior. While the causes of ASD are still not fully understood, research has shown that genetic factors play a significant role in the disorder's development. One area of genetic research that has garnered recent attention is the link between hemizygous variants and autism. Hemizygosity refers to the presence of only one copy of a particular gene, rather than the usual two copies. This can occur when a person has a deletion of one copy of a gene or when a gene is located on the X chromosome, and a male only inherits one copy of the gene. Since males have only one X chromosome, they are particularly susceptible to hemizygous variants. Several studies have identified a link between hemizygous variants and autism. For example, a 2017 study published in Nature Genetics identified a significant association between hemizygous variants and autism risk in a large cohort of individuals with ASD. Another study published in the American Journal of Human Genetics identified several hemizygous variants associated with autism in a group of 1,000 families with at least one child with ASD [1].

One of the genes that has been linked to hemizygous variants and autism is the SHANK3 gene. SHANK3 is a protein that plays a crucial role in the development and function of synapses, the connections between nerve cells in the brain. Mutations in the SHANK3 gene have been identified in individuals with ASD, and hemizygous deletions of the gene have been linked to severe forms of the disorder. Other genes that have been linked to hemizygous variants and autism include NRXN1, CNTNAP2, and PTCHD1. These genes are involved in various aspects of brain development and function, including synapse formation and communication between nerve cells. The identification of hemizygous variants associated with autism has important implications for the diagnosis and treatment of the disorder. Genetic testing can identify individuals who carry these variants and may be at increased risk for developing ASD. This information can help families make informed decisions about reproductive planning and can guide the development of personalized treatment plans for individuals with ASD [2].

The link between hemizygous variants and autism is an area of active research that has important implications for the diagnosis and treatment of the disorder. While the causes of ASD are complex and multifactorial, the identification of specific genetic variants associated with the disorder is a significant step forward in our understanding of the disorder's biological basis. As our understanding of the genetic basis of ASD continues to evolve, it is likely that new insights and discoveries will lead to improved diagnostic and therapeutic approaches for individuals with ASD. Chromosomes are thread-like structures made up of DNA and protein that are found in the nucleus of eukaryotic cells. These structures contain the genetic information that is passed down from one generation to the next, and they play a critical role in cell division and the transmission of genetic traits [3].

Each chromosome is made up of a single, long molecule of DNA that is tightly coiled around proteins called histones. The DNA molecule is divided into a series of units called genes, which contain the instructions for the production of specific proteins that perform essential functions within the body. Humans have 23 pairs of chromosomes, for a total of 46 chromosomes. One member of each pair is inherited from the mother, and the other is inherited from the father. The first 22 pairs of chromosomes are known as autosomes, and the 23rd pair is known as the sex chromosomes, which determine an individual's biological sex. Chromosomes play a crucial role in cell division, which is necessary for the growth and development of an organism [4].

During cell division, the chromosomes duplicate themselves, and then the two copies of each chromosome separate into different cells. This process, known as mitosis, allows for the production of new cells that are genetically identical to the parent cell. Another type of cell division, known as meiosis, occurs during the formation of sex cells, such as eggs and sperm. During meiosis, the chromosomes duplicate themselves, and then the pairs of chromosomes exchange segments of DNA in a process called recombination. The resulting cells have only one copy of each chromosome, which is why sex cells are also known as haploid cells. Chromosomal abnormalities can occur when there are errors in cell division. For example, a cell may end up with an extra copy of a chromosome, or a portion of a chromosome may be deleted or rearranged. These abnormalities can lead to genetic disorders, such as Down syndrome, Turner syndrome, and Klinefelter syndrome [5].

References

1. Hyman SL, Levy SE, Myers SM, et al. Identification, evaluation, and management of children with autism spectrum disorder. Pediatrics. 2020;145(1).
2. van't Hof M, Tisseur C, van Berckelear-Onnes I, et al. Age at autism spectrum disorder diagnosis: A systematic

***Correspondence to:** Paula Arrifano, Department of Molecular Biology, Federal University of Para, Belem, Brazil, E-mail: arrifanogabrela@gmail.com

***Received:** 02-Mar-2023, Manuscript No. AAGMB-23-90410; **Editor assigned:** 03-Mar-2023, PreQC No. AAGMB-23-90410(PQ); **Reviewed:** 16-Mar-2023, QC No. AAGMB-23-90410; **Revised:** 19-Mar-2023, Manuscript No. AAGMB-23-90410(R); **Published:** 27-Mar-2023, DOI:10.35841/aagmb-7.2.138*

***Citation:** Arrifano P. Unraveling the Genetic Basis of Neurodevelopmental Disorders. J Genet Mol Biol. 2023;7(2):138*

review and meta-analysis from 2012 to 2019. Autism. 2021;25(4):862-73.

3. Johnson CP, Myers SM. Identification and evaluation of children with autism spectrum disorders. Pediatrics. 2007;120(5):1183-215.
4. Zwaigenbaum L, Bauman ML, Choueiri R, et al. Early intervention for children with autism spectrum disorder under 3 years of age: recommendations for practice and research. Pediatrics. 2015;136(1):S60-81.
5. Chiarotti F, Venerosi A. Epidemiology of autism spectrum disorders: a review of worldwide prevalence estimates since 2014. Brain Sci. 2020;10(5):274.

Citation: *Arrifano P. Unraveling the Genetic Basis of Neurodevelopmental Disorders. J Genet Mol Biol. 2023;7(2):138*