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**MAPPING THE GENETIC LANDSCAPE OF AUTISM SPECTRUM  
DISORDER: A HOMOEOPATHIC PERSPECTIVE — A NARRATIVE  
REVIEW**

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**ABSTRACT:**

**Background:** Autism Spectrum Disorder(ASD) is a neurodevelopmental disorder with impairments in two areas mainly social communication and restricted and repetitive behaviours. It is typically visible during the second to third year of their life. They also show a lack of appropriate interest in social communication early in their life in severe cases. It is a multifaceted disorder with a complex aetiology. Among all the causes being found, genetic component along with environmental risk factors has been most suggested. Based on the

recent studies more than 200 susceptibility genes have been identified. **Objectives:** This article focus on the influence of genetic component and environmental factors in developing ASD. **Methods:** In this review, literature search was conducted in standard psychiatric textbooks and Google Scholar to assess the impact of related genetics and associated environmental factors on the development of autism. There are various studies that have investigated the relation between genetic factors and ASD. **Results:** This article discusses the role of genetics as a causative factor and also environmental factors that triggers the development of autism spectrum disorder in individuals are being discussed in this article.

**KEYWORDS:** Autism Spectrum Disorder, Neurodevelopmental disorder, Genetic Component, Environmental factors

### **INTRODUCTION:**

Autism Spectrum Disorder is a group of lifelong developmental disorders characterized by their effect on social and communication skills, as well as by a restricted, stereotyped, repetitive interests and activities.<sup>(1)</sup> Before the DSM-5, five overlapping disorders captured the spectrum such as the Autistic disorder, Asperger disorder, Childhood Disintegrative disorder, Rett syndrome, and Pervasive Developmental disorder not otherwise specified. These differed by the level of severity, specific syndrome, and in some cases underlying pathology.<sup>(2)</sup> A genetic basis for ASD is now well established, and with the availability of high-throughput microarray and sequencing platforms, major advances have been made in our understanding of genetic risk factors which reveals rare variants in genes having important roles in synapse formation, function and maintenance.<sup>(3, 4)</sup>

### **METHODOLOGY:**

This narrative review was compiled from standard psychiatric textbooks and peer-reviewed journal articles which are indexed in Google Scholar and PubMed. Relevant literatures from 2005 to 2024 was reviewed.

### **RESULTS:**

From 1940s to the present, understanding the causes of ASD has developed along with the progress in the field of genetics and biology. Some specific genetic markers were identified in children diagnosed with ASD. Studies has advanced from linkage analysis to identifying susceptibility loci in connection with ASD. <sup>(5)</sup>

### **GENETIC SYNDROMIC ASSOCIATIONS IN AUTISM**

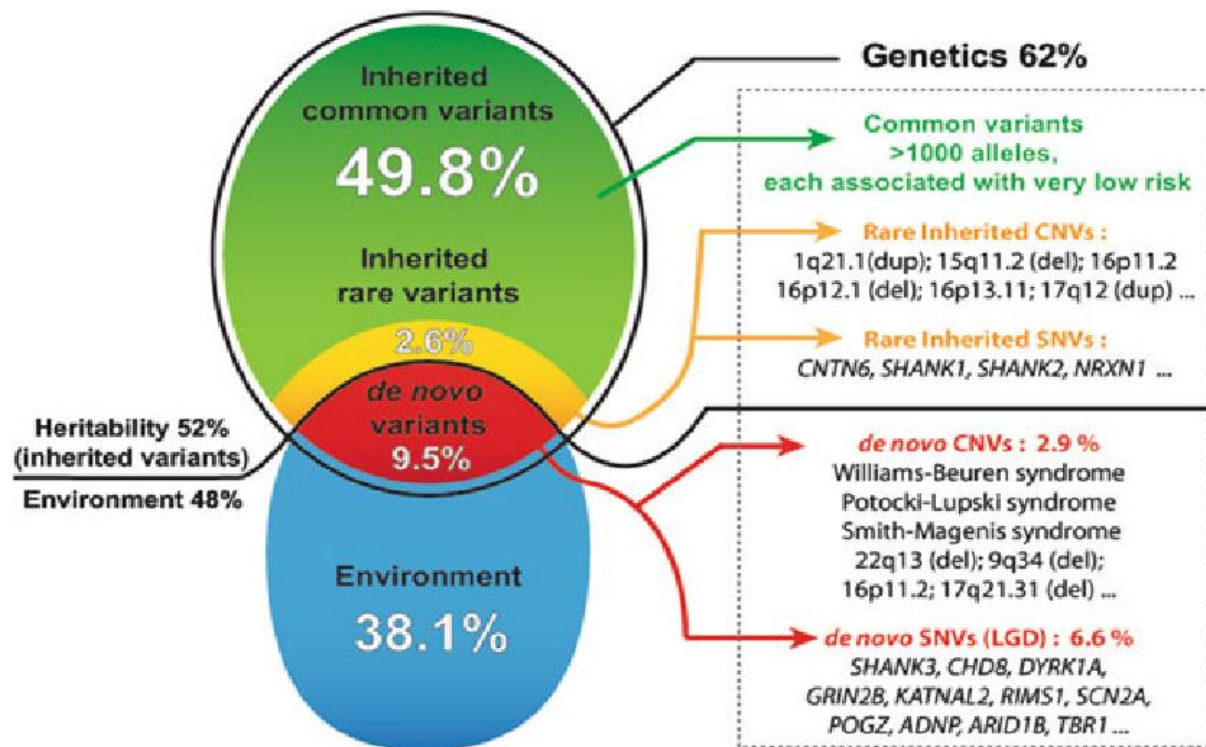
Autism Spectrum Disorder exhibits more prevalence on individuals with genetic syndromes than general population.<sup>(6)</sup> Standardized assessments shows an association between ASD and genetic syndromes while in depth investigations have shown qualitative differences in ASD-like symptoms across different syndrome groups.<sup>(7)</sup> Some of the genetic syndromes like Fragile X syndrome, Down syndrome, Neurofibromatosis, Prader-Willi syndrome etc account for almost more than 10% of all the ASD cases when compared to the general population<sup>(8)</sup>

### **GENE INVOLVEMENT IN DISEASE**

Rare point mutations and analysis of chromosomal abnormalities have been identified where many genes associated with ASD are showing strong effects. Some genes related to ASD have well-defined functional roles, especially those involved in synaptic function, such as the SHANK family of proteins, Neuroligin, and Neurexin, along with proteins associated with Fragile X Syndrome. In addition, other ASD-related genes have also been identified that do not yet show a clear mechanism for the neurodevelopmental dysfunction, including genes that regulate chromatin modification and overall gene expression.<sup>(9,10)</sup> An analysis of lymphoblastoid cell lines showed an elevation of the gene transcript of FOXP1 gene expression in the pathogenesis of ASD.<sup>(11)</sup>

### **ENVIRONMENT AND GENES**

Exposure to air pollution during pregnancy and the first year of life has been suggested as a potential risk factor for Autism Spectrum Disorder. These effects may occur through epigenetic mechanisms similar to those observed with valproate exposure. The risk increases with exposure to pollution, especially during the first trimester of pregnancy and studies have reported association of ASD with air pollutants like nitrogen dioxide and particulate matter (PM2.5 and PM10) at birth and during early life.<sup>(12)</sup> Studies suggest an increased prevalence of ASD through advancing parental age, valproate intake, toxic chemical exposure, maternal diabetes, increased steroidogenic activity etc<sup>(13)</sup>. Furthermore, factors like vaccination, maternal smoking, thimerosal exposure are unrelated to ASD development.<sup>(14)</sup>



**FIG 1: Genetic and Environmental factors.**

## ADVANCED GENETIC TESTING

ASD presents a complex behavioural phenotype, and this variability reflects diverse genetic influences that interact with environmental factors. Therefore, genetic testing has become an essential component in the diagnostic evaluation of these children. Common genetic testing methods for evaluation include array comparative genomic hybridization, microarray analysis, DNA sequencing (using the Sanger method or next-generation sequencing), and polymerase chain reaction. However, no study directly evaluated the impact of these genetic tests on health outcomes. Most available clinical studies assessing their utility indirectly consist of case series that report the diagnostic yields of these tests.<sup>(15)</sup>

## HOMOEOPATHY AND ASD

Autism spectrum disorder is identified as a lifelong disabilities, an appropriate early intervention can reduce the disability and helps them living up to an expected lifestyle and enable them to do their daily activities on their own. The scope of recovery depends on the stage and age at which it is diagnosed in an individual. The intervention of ASD with individualized homeopathic remedies on well-suited similarities has proven effective in previous studies in reducing varied ASD features.<sup>(16)</sup> But based on the current evidence, no

direct clinical studies have been conducted to map the genetic factors to ASD measuring the effect of homoeopathy on those genes.

### **DISCUSSION:**

Autism Spectrum Disorder is genetically diverse and results from both inherited mutations and new genetic variations. Over the past decade, researchers have identified almost hundreds of genes linked to the significant challenges in communication, social understanding, and behaviour which are the core domains of ASD.<sup>(17)</sup> In addition, it highlights the extreme sensitivity of the developing human brain to toxic environmental exposures or pollution, along with previous studies that directly associate prenatal environmental exposures with autism.<sup>(18,19)</sup>

### **CONCLUSION:**

Autism Spectrum Disorder arises from a interplay between genetic predisposition and environmental influences. Advances in genomic research have identified numerous genes associated with ASD, and studies on environmental exposures have highlighted their potential role in modifying neurodevelopmental outcomes. Future research focusing on gene–environment interactions will be crucial for unravelling the underlying mechanisms of ASD and developing effective therapeutic and preventive approaches. And further studies should be promoted to assess the effect of homoeopathy in genetic variations.

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