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GENETIC PREDICTORS IN THE DEVELOPMENT OF AUTISM

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ABSTRACT
Autism spectrum disorders (ASD) comprise a group of neurodevelopmental conditions characterized by deficits in social communication, restricted interests, and repetitive patterns of behavior. In recent decades, particular attention has been devoted to the study of genetic factors that determine susceptibility to the development of ASD. The aim of this article is to analyze the role of genetic susceptibility in the formation of autistic disorders and to review current evidence on genetic mechanisms and their interaction with environmental factors.
KEYWORDS: *Autism spectrum disorders, genetic factors, polygenic inheritance, gene mutations, epigenetics, synaptic dysfunction, genomic variations.*

RELEVANCE

Autism spectrum disorders (ASD) represent one of the most significant medical and social challenges of our time due to the steady increase in their prevalence worldwide. According to international studies, the rate of ASD diagnosis has risen substantially over recent decades, driven both by improvements in diagnostic criteria and by the influence of a complex interplay of biological and environmental factors. Particular relevance is attributed to the study of genetic susceptibility in the development of ASD, as current scientific evidence indicates a high degree of genetic determination. At the same time, ASD are not classical monogenic disorders but arise from a complex interplay of multiple genes, epigenetic mechanisms, and environmental factors.

Understanding the genetic basis of autism is of critical importance for early diagnosis, risk prediction, and the development of personalized approaches to treatment and rehabilitation. Moreover, the study of hereditary predisposition contributes to improving genetic counseling for families in which cases of ASD have already been identified.

Thus, the study of the role of genetic susceptibility in the development of autism spectrum disorders represents a relevant direction in modern medicine and neuroscience, holding both theoretical and practical significance.

AIM OF THE RESEARCH

The aim of the reseach is to analyze the role of genetic susceptibility in the development of autism spectrum disorders (ASD), as well as to examine current data on the genetic mechanisms underlying ASD and their interaction with environmental factors.

MATERIALS AND METHODS

In this study, a review and analysis of contemporary scientific publications devoted to the investigation of the role of genetic susceptibility in the development of autism spectrum disorders were conducted.
The study materials included data from international and domestic scientific articles, systematic reviews, meta-analyses, as well as the results of genetic, clinical, and epidemiological studies published in peer-reviewed scientific journals.
The following research methods were employed in this study:
• systematic analysis of scientific literature;
• comparative analysis of data from various studies;
• generalization and synthesis of the obtained information;
• an analytical approach to assessing the role of genetic and epigenetic factors;
• interpretation of the results of twin and family studies.
Particular attention was paid to studies investigating the polygenic nature of inheritance, gene mutations, genomic variations, and epigenetic mechanisms, as well as their interaction with environmental factors.

RESULTS OF THE STUDY

The conducted analysis of scientific literature has demonstrated that autism spectrum disorders (ASD) have a strong genetic basis and are characterized by a high degree of hereditary predisposition. According to data from twin studies, the concordance rate of ASD in monozygotic twins is significantly higher (70–90%) than in dizygotic twins, which confirms the important role of genetic factors in their development.

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It has been found that ASD are not the result of mutations in a single gene but have a polygenic mode of inheritance. A large number of genes are involved in the development of the disorder, including SHANK3, NRXN1, MECP2, FMR1, and others, each contributing to the disruption of neural connections and synaptic function. In table 1 are presented main genes associated with ASD

Table 1.
Key genes associated with autism spectrum disorders (ASD)

Gene	Function	Association with ASD
SHANK3	Synapse formation	Impaired signal transmission between neurons
NRXN1	Synaptic adhesion	Defects in neural connectivity
MECP2	Regulation of gene expression	Associated with Rett syndrome
FMR1	Protein synthesis in neurons	Fragile X syndrome
CNTNAP2	Development of neural networks	Impaired speech and communication
SCN2A	Neuronal ion channels	Epilepsy and autistic symptoms

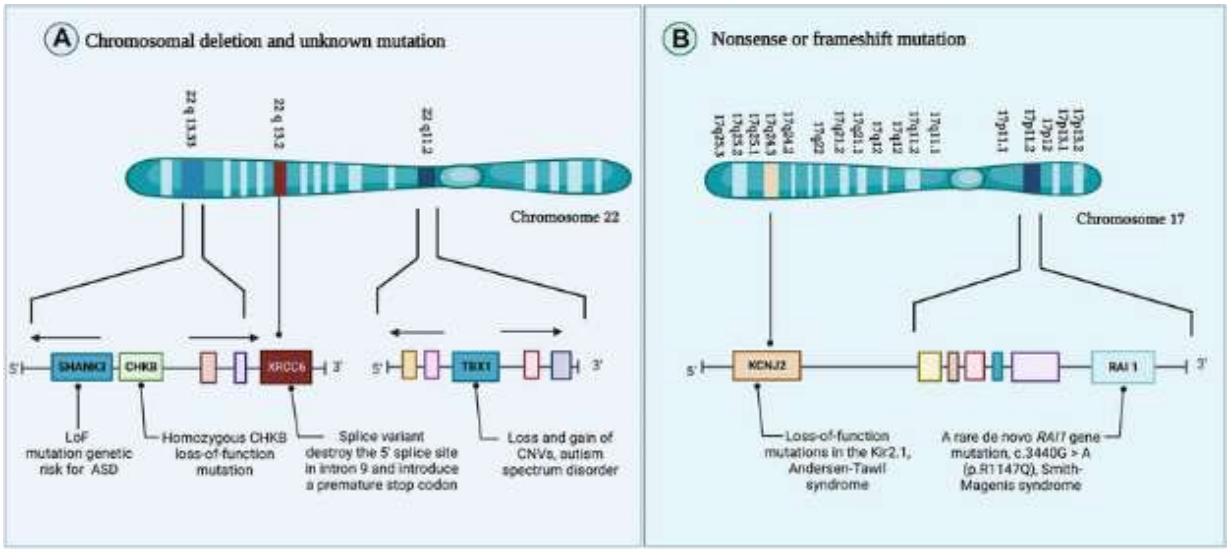


Figure 1. Schematic illustration of the chromosomal mutations or deletions in autism spectrum disorder. A The 22q11.2, 22q13.2 and 22q13.33 regions of the genome have unknown mutations and chromosomal deletions in ASD. Autism spectrum disease can be caused by a mutation in the TBX1 gene at 22q11.2. B Nonsense or frameshift mutations on chromosome 17 that affect two regions. Smith-Magenis syndrome is caused by a loss-of-function in the first locus of mutation RAI 1 gene, 17q11.2. Andersen-Tawil syndrome is caused by the second mutation area at 17q24.3 (Ghafouri-Fard et al., 2023)

It has also been established that genomic variations (CNVs) and epigenetic mechanisms, such as DNA methylation and gene expression regulation, play an important role. These processes can alter gene activity without changing their structure and contribute to the development of the ASD phenotype (table 2).

Table 2.
Heritability of ASD in monozygotic and dizygotic twins

Indicator	Monozygotic twins	Dizygotic twins
ASD concordance rate	70–90%	10–30%
Genetic similarity	100%	~50%
Environmental influence	Moderate	High

In addition, research findings confirm a significant interaction between genetic and environmental factors. Risk factors include intrauterine infections, fetal hypoxia, advanced parental age, and exposure to toxic substances during pregnancy.
Thus, the results of the analysis confirm that the development of ASD is a multifactorial process based on a complex interaction between genetic predisposition and environmental influences.

CONCLUSIONS

1. Autism spectrum disorders (ASD) have a strong genetic basis and are characterized by a high degree of hereditary predisposition.
2. The inheritance of ASD is polygenic, with multiple genes contributing to the development of the disorder, each exerting a small but significant effect on the pathogenesis.

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3. Mutations in individual genes, genomic variations, and epigenetic mechanisms that influence the regulation of gene expression all play an important role in the pathogenesis of ASD.
4. The development of ASD is determined not only by genetic factors but also by their interaction with environmental influences, including prenatal and perinatal factors.
5. The study of hereditary susceptibility is of important significance for early diagnosis, risk assessment of ASD development, and the development of personalized approaches to treatment and rehabilitation of patients.

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