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Genetic Implications and Behaviour Analysis of Children with Autism Spectrum Disorder

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Introduction

Autism Spectrum Disorder (ASD) is one of the most common neurodevelopmental disorders, affecting roughly one out of every 59 children. ASD is very genetically diverse, with both inheritable and de novo gene variants being possible causes. Hundreds of genes have been discovered in the last decade that contribute to the severe communication, social cognitive, and behaviour problems that patients frequently experience. However, these pathogenic variants account for just 10–20% of ASD cases, and patients with comparable pathogenic variants can be diagnosed at different degrees of the spectrum.

This illness is marked by persistent deficiencies in social interaction and communication throughout a wide range of life areas, as well as confined and repetitive behavioural patterns. The DSM states that symptoms of autism spectrum disorder must appear during the early stages of development and cause severe impairment in crucial areas of life, such as social and occupational functioning [1].

Autism spectrum disorder (ASD) behaviour analysis

The outward appearance of an autistic youngster is usually normal. These children, on the other hand, have an uneven development profile that may be detected in the first three years of life and lasts until adulthood. The Triade of Social Impediments is defined by a tight and consistent pattern of intelligence levels ranging from mental retardation to exceptional performance in some cognitive domains (such as music, arts, mathematic, or memory) or savant abilities. Savant abilities can occur in autistic children, despite the fact that 80% of autistic children display mental impairment. However, the global intelligence ratio is low. It's important to note the distinction between mental retardation and autism: The first has a consistent developmental deficit, whereas the last has an uneven profile with varying levels of commitment [2].

The World Health Organization's International Categorization of Diseases and the American Academy of Psychiatry's Manual of Diagnosis and Statistics of Mental Diseases are two classification and diagnosis systems that can help identify autism from other disorders (DSM-IV). The name Child Autism was replaced with Autistic Disturbance in these systems, officially distinguishing it from Asperger syndrome.

Autism is characterised by echolalia (the child repeats the same sound over and over), inability to play symbolic games, prenominal inversion (the use of the third person instead of the first), abnormal voice sound, absence or deficit of speech, peculiar relation within animate objects, stereotyped interests, repetitive behaviours, deficit in social, emotional, and interests sharing, and difficulty establishing visual and physical contact. Hyperactivity, attention deficit, impulsivity, aggressivity, and self-aggressivity are all common symptoms among these people. People with Asperger's syndrome have social problems and narrow interests. Their speaking abilities, on the other hand, stay intact or are above normal for their age.

Individuals with Global Disturbance of Non-Specified Development have alterations in one or more developmental domains, but these are insufficient to be classified as autistic or Asperger disturbances since they are infrequent or mild.

Diagnosis

Because genetic markers for this disorder are still unavailable, diagnosis is usually based on a thorough examination of the patient's history and family inquiry (cognitive and behavioural capacities) using the Autism Diagnostic Interview-Revised (ADI-R) and the Autism Diagnostic Observation Schedule (ADOS).

Autism is thought to be caused by a brain aberration that has yet to be identified and is linked to a genetic origin. Because ASDs have such a diverse origin, no clear medical test or solution for these illnesses has yet been found. As a result, only a few cases of conclusive diagnosis occur before the age of twenty-four months (a well-established diagnosis is normally done between 3 and 6 years of age).

Clinical genetic tests are also available, and the most common technique entails first and foremost a chromosome microarray analysis, which allows for the detection of chromosome copy number variation as well as the presence of large chromosomal deletions or duplications. The second method involves performing molecular DNA tests for individual genes or even full genome sequencing. Patients can be tested for Fragile X syndrome, for example, by analysing a particular gene or, in the presence of a specific feature/condition, by searching for a set of genes linked to those traits [3].

Several genetic illnesses have been linked to an elevated risk of ASDs, accounting for 5-10% of all identified cases. Because the frequency of autism is higher in men, some studies have shown that the X chromosome's hereditary influence increases vulnerability to ASDs.

X-Fragile syndrome (XFS)

Patients typically exhibit developmental issues such as speech and motor delay, as well as autistic behaviours. Because the key implicated gene is located on the X chromosome (Xq27.3), males are more likely to acquire X-fragile syndrome. In the vast majority of cases (99%), the disease is caused by the loss of function of the FMR1 gene, which is caused by the presence of numerous copies of the triplet repeat CGG in the gene's 5' untranslated region (5' UTR). The FMR1 protein (FMR1P - fragile X mental retardation 1 protein) is produced by the FMR1 gene and is primarily found in the cytoplasm of neurons. It is involved in important processes such as ribosomal translation and synapse structure and function maturation, as well as acting as a suppressor in the post-synaptic region.

Autism is very common in XFS people, with a prevalence of 25-33% [4]. The FMR1 protein has the ability to influence the expression of numerous genes implicated in the development of autism. The existence of a particular biomarker, the aberrant growth of the CGG repeat on the X chromosome, distinguishes XFS from autism.

Down and Pradel-Willi/Angelman syndrome

The most common chromosomal aberration in patients with ASDs is Down syndrome (7-10% of people with ASDs are carriers), followed by the one seen in the 15q11-13 region (present in 1-4% of autistic patients). When the 15q11-13 region is duplicated or inverted, it causes a high rate of epilepsy in children, as well as muscle hypotony and motor coordination issues, as well as strong or moderate mental impairment, absent or delayed speech, and severe hyperactivity. Maternal or paternal imprinting illnesses will emerge if deletions occur in the referenced chromosomic regions: Angelman and Prader-Willi syndromes are two different types of syndromes.

Rett syndrome

Rett syndrome, which is linked to the MeCP2 gene, is an X-chromosome dominant condition that is thought to be one of the causes of a small number of autism cases. In most cases, this syndrome is fatal in boys. Exons 2, 3, and 4 of the aforementioned gene are the most often mutated.

Epilepsy

Epilepsy episodes occur in a casual form in people with ASDs, according to some researchers. Data reveal that people with ASDs have a higher risk of epilepsy, which is caused by a Central Nervous System aberration defined by neuropathological alterations [5]. Because CNS dysfunctions greatly increase the likelihood of having epilepsy, one-third of autistic people develop it. The percentage of autistic children with epilepsy ranges from 5% to 46%.

Mental retardation

The AGTR2 gene, which is found on the X chromosome (Xq22-q23), appears to be implicated in the development of mental impairment in autistic people, as it is deleted in these people. The Xq13-q21 region, which comprises genes from the neuroligin family, is, however, considerably more crucial in the development of this disease. Neuroligins play a critical role in cellular connection between neuronal cells via adhesion molecules.

Conclusion

There is still a lot to learn about autism spectrum disorders, as there are likely to be numerous susceptibility factors at play, some of which have yet to be uncovered. It's critical to find genetic variants that haven't been found by traditional genotyping research by focusing on several loci analysis at the same time. The criteria for diagnosing autism spectrum disorders have been improved. Identification of genetic variations responsible for distinct autism symptoms would be the perfect diagnosis. The role of genetics in autism is evident, but it is insufficient to explain this complicated condition without taking into account environmental influences.

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