AUTISM SPECTRUM DISORDER: BEHAVIORAL AND GENETIC ASPECTS

TRANSTORNO DO ESPECTRO DO AUTISMO: ASPECTOS COMPORTAMENTAIS E GENÉTICOS

TRASTORNO DEL ESPECTRO AUTISTA: ASPECTOS CONDUCTUALES Y GENÉTICOS

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ABSTRACT

Autism Spectrum Disorder (ASD) is a neurodevelopmental disorder characterized by persistent deficits in communication and social interaction, associated with restricted and repetitive patterns of behavior, interests, or activities. This study aims to present a review of the main conceptual, clinical, and genetic aspects related to autism, highlighting scientific advances that have contributed to a broader understanding of the disorder. Over time, the conception of autism has undergone important changes, and it is currently understood as a spectrum that involves different levels of manifestation and need for support. Clinically, ASD manifests itself from early childhood and may include difficulties in verbal and nonverbal communication, impairments in social interaction, stereotyped behaviors, restricted interests, and sensory alterations. Early identification of the disorder is considered fundamental, as it allows for more effective therapeutic interventions and favors better outcomes in the cognitive, social, and communicative development of the individual. In the field of genetics, recent research indicates that ASD has a strong hereditary component, involving multiple genes associated with the development and functioning of the nervous system. Rare and common genetic variants, spontaneous mutations, and epigenetic alterations contribute to the clinical heterogeneity observed among individuals on the spectrum. Furthermore,

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environmental factors can interact with genetic predisposition, influencing the risk of developing the disorder. Therefore, ASD should be understood as a multifactorial and complex condition, whose continuous investigation is essential to expand knowledge about its biological mechanisms, improve diagnostic strategies, and develop more effective therapeutic interventions.

Keywords: Autism. Neurological Development. Genes. Epigenetics.

RESUMO

O Transtorno do Espectro Autista (TEA) é um transtorno do neurodesenvolvimento caracterizado por déficits persistentes na comunicação e na interação social, associados a padrões restritos e repetitivos de comportamento, interesses ou atividades. O presente estudo tem como objetivo apresentar uma revisão sobre os principais aspectos conceituais, clínicos e genéticos relacionados ao autismo, destacando avanços científicos que contribuíram para ampliar a compreensão do transtorno. Ao longo do tempo, a concepção do autismo passou por mudanças importantes, sendo atualmente compreendido como um espectro que envolve diferentes níveis de manifestação e necessidade de suporte. Clinicamente, o TEA manifesta-se desde a primeira infância e pode incluir dificuldades na comunicação verbal e não verbal, prejuízos na interação social, comportamentos estereotipados, interesses restritos e alterações sensoriais. A identificação precoce do transtorno é considerada fundamental, pois possibilita intervenções terapêuticas mais eficazes e favorece melhores resultados no desenvolvimento cognitivo, social e comunicativo do indivíduo. No campo da genética, pesquisas recentes indicam que o TEA possui forte componente hereditário, envolvendo múltiplos genes associados ao desenvolvimento e funcionamento do sistema nervoso. Variantes genéticas raras e comuns, mutações espontâneas e alterações epigenéticas contribuem para a heterogeneidade clínica observada entre os indivíduos no espectro. Além disso, fatores ambientais podem interagir com a predisposição genética, influenciando o risco de desenvolvimento do transtorno. Dessa forma, o TEA deve ser compreendido como uma condição multifatorial e complexa, cuja investigação contínua é essencial para ampliar o conhecimento sobre seus mecanismos biológicos, aprimorar estratégias diagnósticas e desenvolver intervenções terapêuticas mais eficazes.

Palavras-chave: Autismo. Desenvolvimento Neurológico. Genes Autismo. Epigenética Autismo.

RESUMEN

El trastorno del espectro autista (TEA) es un trastorno del neurodesarrollo caracterizado por déficits persistentes en la comunicación y la interacción social, asociados a patrones de comportamiento, intereses o actividades restringidos y repetitivos. Este estudio tiene como objetivo presentar una revisión de los principales aspectos conceptuales, clínicos y genéticos relacionados con el autismo, destacando los avances científicos que han contribuido a una comprensión más amplia del trastorno. Con el tiempo, la concepción del autismo ha experimentado cambios importantes y actualmente se entiende como un espectro que implica diferentes niveles de manifestación y necesidad de apoyo. Clínicamente, el TEA se manifiesta desde la primera infancia y puede incluir dificultades en la comunicación verbal y no verbal, deficiencias en la interacción social, comportamientos estereotipados, intereses restringidos y alteraciones sensoriales. La identificación temprana del trastorno se considera fundamental, ya que permite intervenciones terapéuticas más efectivas y favorece mejores resultados en el desarrollo cognitivo, social y comunicativo del individuo. En el campo de la

genética, investigaciones recientes indican que el TEA tiene un fuerte componente hereditario, que involucra múltiples genes asociados con el desarrollo y el funcionamiento del sistema nervioso. Las variantes genéticas raras y comunes, las mutaciones espontáneas y las alteraciones epigenéticas contribuyen a la heterogeneidad clínica observada entre las personas con trastorno del espectro autista (TEA). Además, los factores ambientales pueden interactuar con la predisposición genética, influyendo en el riesgo de desarrollar el trastorno. Por lo tanto, el TEA debe entenderse como una condición multifactorial y compleja, cuya investigación continua es esencial para ampliar el conocimiento sobre sus mecanismos biológicos, mejorar las estrategias diagnósticas y desarrollar intervenciones terapéuticas más eficaces.

Palabras clave: Autismo. Desarrollo Neurológico. Genes. Epigenética.

1 INTRODUCTION

Autism is a term originating from the Greek *autos* and means "turned to oneself". The year 1908 represents a historical milestone in autism when it was first used by psychiatrist Eugen Bleuler (1857-1939) to describe the escape from reality into an inner world (inner withdrawal), a common behavior in schizophrenic patients (Cunha, 2020). However, autism, as it is known today, can be considered a "technically recent phenomenon" that permeates discussions of its causes, treatment and public policies, and whose diagnosis is significant both for autistic people and for their families (Marfinati; Abrão, 2014; Fernandes; Costa e Silva, 2023).

The history and conceptualization of autism go back to its description and diagnostic categorization in which behaviors and characteristics configure a specific picture. Its psychopathological diagnosis is based on the formulations of Leo Kanner (2012, p. 168), a psychiatrist considered the father of autism.

Kanner observed, in the manifestations of early childhood children, a strong desire for solitude as a form of self-regulation (intense sensory and emotional stimuli, affectivity and stress control) and resistance to change, preferring predictable routines and actions as a way to reduce anxiety and stress (Bialer; Voltolini, 2022). Kanner conceived autism as a disorder distinct from schizophrenia, in which, among other aspects of child development, communicative difficulty stands out.

From 1908 onwards, there were several advances in the delimitation of the characteristics and diagnosis of autism (Marfinati; Abraão, 2014; Brazil, 2015a, b; Dias, 2015; Brazil, 2020; Fernandes; Costa e Silva, 2023).

In 1943, the Austrian-American psychiatrist Leo Kanner made the first diagnoses of autism, disassociating it from schizophrenia and treating it as a behavioral syndrome. In that

year, he published his first discoveries about autism in his study entitled "*Autistic disturbances of affective contact*", outlining the main characteristic of autism: the child's inability to communicate and relate to other people, from the first years of life (Marfinati; Abrão, 2014).

In 1944, Hans Asperger, a psychiatrist and researcher from Vienna, exposed clinical conditions similar to autism, describing four children who presented the central issue – disorder in the relationship with the environment: poverty of gestural and facial expressions, restlessness; stereotyped, aimless, rhythmic movements; artificial speech; fields of interest different from those presented by other children of the same age, abstraction and inventiveness, unconscious imitation of adult behaviors; field of emotions without affective poverty, but with qualitative alteration and disharmony (hypersensitivity of instincts, "extreme egocentrism" and lack of sense of humor). Asperger pointed out the higher prevalence of autism in boys, who had little empathy, restricted interests and a peculiar way of talking.

In 1952, the American Psychiatric Association publishes the first edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-1), in which autism appears referenced, establishing specific standards, becoming a world reference for researchers and clinicians in the segment. The Manual provided nomenclatures and standard criteria for whether the diagnosis of mental disorders was established, and the various symptoms of autism were classified as a subgroup of childhood schizophrenia.

It was in 1978, however, that British psychiatrist Michael Rutter classified autism as a cognitive development disorder. He proposed that it was: a delay, social deviations and communication problems and not just an intellectual disability; unusual behaviors presented as stereotyped movements and mannerisms; it began even before 30 months of age. From his studies, autism came to be recognized as a specific condition, classified as an Pervasive Developmental Disorder (PDD), in which multiple areas of brain functioning are affected by autism and related conditions.

From the 1980s onwards, the concept and psychoanalytic conceptions of autism were defined in psychiatric manuals, becoming part of the Pervasive Developmental Disorders (PDD) – ICD-10 (Brasil, 2015a). The subsidiary criteria for the diagnosis of autism involved changes over time, being described in nosological categorization manuals, such as the *Diagnostic and Statistical Manual of Mental Disorders* (DSM) and the *International Classification of Diseases and Related Health Problems* (ICD) (Fernandes; Tomazell; Girianelli, 2020).

In Brazil, in 2012, Law No. 12,764 (Berenice Piana Law) was sanctioned , which institutes the National Policy for the Protection of the Rights of Persons with Autism Spectrum Disorder. This law represents a significant legal framework that sought to guarantee rights to

people with ASD, such as access to early diagnosis, treatment, therapies and medication offered by the Unified Health System (SUS), education and social protection, work and services aimed at equal opportunities (Sousa, 2021).

Launched in 2013, the *DSM-5* now encompasses the various subcategories of autism in a single diagnosis: Autism Spectrum Disorder (ASD), that is, a single spectrum with different levels of severity and deficits in two central domains: a) deficiencies in social communication, social interaction, and communication; b) repetitive, stereotyped, restricted, and behaviors of interests and activities (Paoli; Machado, 2022). In this version of the DSM-5, the denomination used is "autism spectrum disorders", located in the group of "neurodevelopmental disorders", which encompasses autistic disorder (autism), Asperger's disorder, childhood disintegrative disorder, Rett's disorder and pervasive developmental disorder not otherwise specified by the DSM-IV (APA, 2014; Brazil, 2015a).

In 2015, Law 13.146 (Brazilian Law for the Inclusion of Persons with Disabilities) was enacted, which created the Statute of Persons with Disabilities, increasing the protection of people with ASD. The Law defines a person with a disability as "one who has a long-term impairment of a physical, mental, intellectual or sensory nature" (Brasil, 2015b, art. 2). The Statute of Persons with Disabilities (Law 13,146) and the National Policy for the Protection of the Rights of Persons with Autism Spectrum Disorder (Law No. 12,764/2012) are considered important symbols in the defense of equal rights for the disabled, in the fight against discrimination and in the regulation of accessibility and priority care (Setúbal; Fayan, 2016).

In 2020, Law 13.977 (or Romeo Mion Law) came into force, which created the Identification Card for Persons with Autism Spectrum Disorder (Ciptea), with free issuance by states and municipalities (Brasil, 2020). The card is a substitute for the medical certificate and has the function of facilitating access to rights provided for in Law No. 12,764/2012, known as the Berenice Piana Law (Brazil, 2012).

In 2022, the version of the International Statistical Classification of Diseases and Related Health Problems, the ICD-11, follows what had been proposed in the DSM-5 and adopts the nomenclature Autism Spectrum Disorder. The new version encompasses all previous diagnoses classified as Pervasive Developmental Disorder (PDD) to allow an individualized and more accurate assessment of autism according to the need for support, in addition to making the diagnosis more inclusive and aligned with individual needs (Freitas *et al.*, 2023).

From the point of view of care, autistic people did not have support outside the health field and were cared for by philanthropic (such as Apae and the Pestalozzi Society) and educational entities, or by non-governmental organizations that offered social assistance

services, for example, family associations. Few had access to Mental Health services, in psychiatric or university hospitals, with multiprofessional treatment, but without articulation with a territorial network of services, as recommended by the National Mental Health Policy. Other services to autistic people were provided in a traditional outpatient setting, accompanied by a psychiatrist or neurologist, with drug treatment. Only recently, autism has entered the official political agenda of health and, with the publication of Ordinance No. 336/2002 (Brasil, 2002), the Center for Psychosocial Care for Children and Adolescents (CAPSI) has been consolidated "as a privileged equipment for psychosocial care for children with autism within the scope of the SUS, although it is not exclusively aimed at this clientele" (Brasil 2015b, p. 29).

According to Brazilian law, "the person with autism spectrum disorder is considered a person with a disability" (Brasil, 2012, art. 1, § 2) and has specific rights, such as priority care, free access to health, education and social assistance, and Law No. 13,146/2015 (Inclusion Law) encompasses people with autism, expanding their rights (Brasil, 2015b).

The Diagnostic and Statistical Manual of Mental Disorders*:* DSM-5 describes autism, also known as Autism Spectrum Disorder (ASD),[12] as a general condition of people with behavior characterized by low social interaction, deficit in social communicative ability (verbal and non-verbal), repetitive, restricted and stereotyped behaviors, sensory and motor issues, conduct control, little interest in performing the proposed activities (APA, 2014), and may have peculiar interests, such as music, history, politics, philosophy, among others (Paoli; Machado, 2022).

ASD is part of the group of neurodevelopmental disorders, with symptoms and level of impairment significantly varying from person to person. According to the DSM-5 Diagnostic and Statistical Manual of Mental Disorders, neurodevelopmental disorders group conditions that begin in the developmental period and manifest early, even before the child enters school. The disorder is characterized

> [...] by developmental deficits that cause impairments in personal, social, academic or professional functioning. Developmental deficits range from very specific limitations in learning or in the control of executive functions to global impairments in social skills or intelligence. It is frequent [...] the occurrence of more than one neurodevelopmental

[12] The 11th Revision of the International Classification of Diseases (ICD-11) was published in 2022. The ICD-11 adopted the nomenclature Autism Spectrum Disorder (ASD) to encompass all diagnoses previously classified as Pervasive Developmental Disorder (PDD), under code 6A02, including: childhood autism, Asperger's syndrome, and childhood disintegrative disorder. By unifying the diagnosis, the ICD-11 enables subdivisions of ASD and assists in a more accurate diagnosis and the development of individualized treatment plans. Rett syndrome, however, has its own code (LD90.4) in the ICD-11, and is no longer considered a member of the autism spectrum (Brasil, 2024). The Note establishes the implementation of ICD-11 in Brazil as of January 2027, although the World Health Organization (WHO) has adopted it as of January 2022.

> disorder; for example, individuals with autism spectrum disorder often have intellectual disability (intellectual developmental disorder), and many children with attention-deficit/hyperactivity disorder (ADHD) also have a specific learning disorder. In the case of some disorders, the clinical presentation includes symptoms of both excess and deficits and delays in reaching the expected milestones. (APA, 2014, p. 31)

Autism spectrum disorder (ASD) is identified by two areas: a) persistent deficits in social communication [adaptive functioning] and social interaction, and b) presence of restricted and repetitive patterns of behavior, interests, or activities (Fernandes; Costa e Silva, 2023). The main symptoms can appear in early childhood, however, it is emphasized that the diagnosis is complex and variable in intensity.

2 CLINICAL BEHAVIORAL CHARACTERISTICS

ASD originates in the first years of life, although it does not manifest uniformly at this stage. Some children have apparent and not very suggestive symptoms at birth. Most autism symptoms are identified between 12 and 24 months of age, but in general, they are usually diagnosed at 4 or 5 years of age (Cardoso *et al*., 2019).

Autism Spectrum Disorder, or simply autism, is a neurological disorder that affects the individual's social environment (lack of interaction, including social isolation), their communication, and repetitive behavior patterns. The earlier it is diagnosed in the child, the better their development will be in the areas of communication, learning, and behavior reduction (Araújo, 2022). The stage of functional impairment varies according to the severity of the autistic condition, the level of development and chronological age, the characteristics of the individual and his environment that are evident in the developmental period, because the signs can be masked through interventions and compensations (APA, 2014).

Commonly, autistic children manifest a high degree of loneliness and other traits: extreme disinterest, does not respond to external stimuli, has "impaired communication, mutism, echolalic language, obsessive insistence on sameness, anxiety in the face of new situations, repetitive rituals, fascination with objects and disinterest in people, distressing reaction" to the presence of others (Barroso, 2019, p. 1234). Without responding to external stimuli, autistic children practically do not develop social interaction, live in their own universe, although they maintain an intelligent relationship with objects and memory above the ordinary and need predictability translated into routine, sameness and monotony (Maranhão; Pires, 2017; Tamanaha *et al*., 2022).

Other components for ASD include: impaired reciprocity, nonverbal communication, motor stereotypy, repetitive behaviors and interests (intense and unusual), sensory sensitivities, speech (language) and cognition alterations, frequent atypical sensory responses (Barros; Source, 2016; Meneses and Silva, 2020; Arvigo; Schwartzman, 2021; Silva *et al*, 2025). ASD may also be associated with other psychiatric disorders, such as attention deficit hyperactivity disorder, depression, and anxiety, and medical conditions, such as epilepsy and genetic disorders (Cardoso *et al.*, 2019).

Children with ASD have a unique triad, found in "difficulty and qualitative impairments of verbal and non-verbal communication, in social interactivity and in the restriction of their cycle of activities and interests", and the symptomatology may bring together "stereotyped movements and mannerisms", in addition to "variable intelligence pattern and extremely labile temperament" (Pinto *et al.*, 2016, p. 2). The symptomatology also includes results bordering on stress such as "difficulty concentrating and paying attention, nervousness,

anxiety, depressed mood, sadness, intense fear, sudden changes in mood or feeling" for no apparent reason, obsessions and cognitive inflexibility, withdrawal from oneself (isolation), relationship difficulties, nightmares, among others (Almeida *et al*., 2021, p. 92). What is certain is that the early identification of risk signs and screening for the disorder in early childhood is essential, since the sooner it is identified and treatment is initiated, the better the results for cognitive development and language (Almeida *et al*., 2021, p. 91).

Childhood autism is a serious developmental disorder that encompasses socio-communicative impairments (Mercado, 2022) such as language regression (Backes; Zanon; Bosa, 2017). It also has the consequence of compromising the acquisition of some of the skills indispensable for human life, with impairments in social interactions, deficiencies in verbal and non-verbal communication, and the limitation of activities and interests. This whole picture directly interferes in social relationships (including family relationships), in the school environment, in the relationship between student and teacher, and even between special students and those considered normal (Bianchi; Abrão, 2023, p. 5261), although he may have special abilities, clumsy motor movements, lack of empathy, extreme sincerity, and few friends (Paoli, Machado, 2022).

Adolescence is a transition between childhood and adulthood and presents itself as a period with unique challenges for all young people and, in particular, for young people with ASD. In adolescence, autistic people may suffer a behavioral decline, with a decrease in language skills and sociability, have higher levels of anxiety and depression related to the degree of self-awareness and their inability to relate socially when trying to establish friendships, talk and interact with other adolescents and even adults. The lack of ability to interact makes it easier for autistic people to become victims of psychological and physical abuse – a reason for frustrations that make them more introspective (Serbai; Priotto, 2021).

Although the symptoms of autism tend to improve with advancing age, in adolescence there are still marked difficulties in social relationships and communication, also due to the expansion of interest and greater demands inherent to this stage of life, in relation to autonomy, psycho-affective development and educational and social processes. Despite this, adolescents with ASD, when adequately stimulated, tend to achieve a higher degree of self-sufficiency in adulthood in the long term (Serbai; Priotto, 2021).

Adolescents with ASD are confronted with the presence of numerous issues (search for autonomy, insertion in social groups, construction of love partnerships) that are reduced throughout life. It is worth remembering that characteristics of the individual and the environment around them will show the variation of functional impairment, since the

manifestations of the disorder essentially depend on the severity of the autistic condition (Cardoso *et al*., 2019).

The persistence of deficits in adolescence (in communication, social interaction, etc.) marks reciprocal social movements and inclusion in adolescent groups. However, the search for autonomy (personal care, school demands) and independence (displacement from one place to another, personal care) can generate distancing from parental figures, although it is an important foundation in the construction of identity. These factors, added to the individualized search in the sphere of love, intensify the obstacles in the sharing of interests, emotions or affections: the autistic adolescent must break these obstacles in the affective approximation made difficult by the lack of verbal and non-verbal communication, for the initiation, maintenance and understanding of relationships, as well as behavior adjusted to social contexts (groups of friends, standard social coexistence, *e.g*.) and control of reactivity to sensory stimuli in the environment (Saad; Bastos; Souza, 2020).

Adolescents must have adequate family (emotional and material) and psychological support in order to be able to deal with situations in which social demands exceed their limited natural capacities: in the construction of social and professional functioning and activities performed in the family context; in the perception of affectivity (relationships of affection) among family members; in the reduction of stress levels and excessive overload in care (Freitas *et al*., 2020).

In adults, Autism Spectrum Disorder (ASD) is often neglected, given that, predominantly associated with childhood, its identification is difficult because its typical signs are masked over the years and its diagnosis in adults usually occurs after some episode of dysregulation in the professional or social sphere (Duarte; Ribeiro; Nazaré, 2024).

In fact, the behavioral changes manifested in the autistic individual can be present at any age. Adults with ASD present: absence of intellectual deficit and have more fluent language and adequate levels of intelligence, although they may commonly present difficulties in social interaction and difficulties in acquiring new adaptive skills. This can occur through mechanisms of copying neurotypical behaviors that mask autistic symptoms and camouflage social interactions, by imitating gestures, sounds, and facial expressions in certain situations, despite the fact that the adult autistic person has social skills of a limited, restricted, and rehearsed nature, and expresses himself in monologues (Ronzani *et al*., 2023).

As autism is a neurodevelopmental disorder, it is characterized by difficulty in communication, social interaction and restrictive or repetitive behaviors, with varying degrees of severity and manifestation, being a permanent condition that includes clinical

manifestations that require a holistic approach; to this end, adaptive functioning and social demands are taken into account, and the condition of adult autism tends to be less evident than in children (Alves, 2024). However, adults with ASD may face ongoing challenges, with persistent symptoms (such as aggression and social difficulties), creating significant impacts on the social and health services available to this population. In adults, late diagnosis tends to compromise emotional and social development, cause tension, isolation and stress, limit opportunities for insertion in the labor market and in society (Duarte; Ribeiro; Nazaré, 2024). It can generate a greater degree of anxiety due to the perception of differences in relation to others, however, it can also constitute an opportunity for self-acceptance and understanding to improve quality of life and reduce anxious behaviors or behaviors considered inappropriate (Carvalho; Neres; Nascimento, 2024).

Fabretti *et al*. (2024) reinforce the presence, in adults, of "challenges in social interactions, atypical communication, repetitive behaviors, and sensory sensitivities", that is, they highlight the clinical repercussions of the "recurrent presence of comorbidities, especially anxiety and depression, in addition to challenges in terms of physical and academic health". Anxiety and depression appear as protagonists that aggravate the clinical panorama of autistic adults who have to face frequent "challenges in dealing with social and environmental demands", difficulties in social engagement and interpersonal interactions that usually contribute to the development of mental disorders.

In addition, adult autism has to overcome physical health challenges, including the presence of simultaneous medical conditions and medical comorbidities, such as gastrointestinal disorders and autoimmune diseases, which increase the complexity of management (Brasil, 2015a). Difficulties in socialization should also be highlighted, resulting in isolation and reduced opportunities in the construction of meaningful interpersonal relationships; the obstacles (cognitive inflexibility, sensory hypersensitivity, adaptive difficulties to change) that compromise performance and limit growth opportunities (Paiva *et al*., 2025).

3 DIAGNOSIS

Early diagnosis of ASD, especially between 2 and 6 years of age, is extremely important to: optimize the development of the infant and his quality of life, enable a positive change in the condition and stimulate the child (improve social and communicative skills, for example); increase the results of therapies and treatments performed by the multidisciplinary team; increase the chances of success in the appropriate treatment of other diseases (current or supervening) through interventions personalized and timely supports in the early stages of

the disease, before it progresses; prevent possible more significant harm to the child in the future and expand awareness among parents and caregivers. In addition, it reduces the cost of care and medication and unwanted and challenging behaviors (Doubrawa; Menezes, 2023; Ramos *et al*., 2024; Mota *et al*., 2025).

Early diagnosis in children can identify autistic characteristics such as "intense relationship with inanimate objects; intelligent physiognomy; cognitive potential; changes in communication; possible special abilities, such as memory; peculiar interests; obstinacy for rituals and unaltering the environment" (Paoli; Machado, 2022, p. 548).

Among the benefits provided by the early diagnosis of autism, Girianelli *et al*. (2023) mention: reduction of costs associated with late interventions, better treatment effectiveness and overcoming challenges, deeper understanding of the individual needs of the autistic person, significant additional advantages for family members, caregivers and the affected individuals themselves (improvement in social and academic/school adaptation), expansion of social, communicative (verbal and non-verbal) and motor skills, improvement of quality of life, establishment of support and favorable support throughout life to face specific challenges (associated with cognitive, social, sensory, and behavioral development), possibility of implementing rapid and efficient therapeutic strategies, positive influence on neural plasticity throughout development.

Leite *et al*. (2024) and Mota *et al*. (2025) corroborate these benefits and adduce others: it favors and enhances the possibilities of intervention in the early stages of child development, the acquisition of repertoire, the development of cognitive (verbal language and communication), sociocognitive (shared attention) and behavioral (social skills) skills, better guidance of parents and caregivers (psychoeducation and development of management strategies).

A possible diagnostic delay – due to various factors such as ethnicity, low family income, lack of stimulation in childhood, lack of information and perception of parents/guardians about child development and signs of autism – may evidence a cementation of the symptoms in the long term (worsening of clinical signs) and the consequences of the disease: insensitivity to pain or sensory hypersensitivity, without fear of danger (it causes accidents), inability of the brain to change its structure based on extrinsic stimuli in early childhood (neuroplasticity), ineffectiveness of interventions, deficient adaptation and rehabilitation of the child, impairment of cognitive, social, sensory and behavioral development (Silva; Araújo; Dornelas, 2020; Doubrawa; Menezes, 2023).

4 GENETIC FACTORS

4.1 AUTISM GENETICS: OVERVIEW

Autism Spectrum Disorder (ASD) is characterized by persistent difficulties in social communication and restricted, repetitive patterns of behavior, interests, or activities. In recent years, significant advances in the field of genetics have contributed to understanding the biological mechanisms underlying ASD, revealing that genetic factors play a central role in the etiology of the disorder (Ramaswami *et al*., 2018).

Heritability studies indicate that between 50% and 90% of the risk of developing ASD can be explained by genetic factors, suggesting a substantial contribution of hereditary variants and de *novo mutations* (Sanders *et al*., 2018). Genome-wide association analyses (GWAS) have identified multiple risk-related *loci* for ASD, including genes involved in synaptic regulation, neuronal development, and modulation of brain plasticity (Grove *et al*., 2019).

In addition, investigations have pointed out that ASD is genetically heterogeneous, with rare variants of great effect, such as chromosomal deletions or duplications (CNVs), coexisting with common variants of small effect (Satterstrom *et al*., 2020). This genetic diversity may explain the wide clinical and phenotypic variability observed among individuals on the spectrum.

Research also suggests that changes in gene expression during brain development, especially in the prenatal period and in the first years of life, play a relevant role in the manifestation of ASD. Transcriptomic studies reveal abnormal patterns of gene coexpression in neural networks of different forms, mainly affecting regions associated with sociability and language (Werling *et al*., 2020).

From a translational point of view, the identification of genetic variants associated with ASD opens up possibilities for the development of biomarkers and personalized therapies. Although still far from routine clinical practice, genetic analysis is already used in some contexts to assist in differential diagnosis and family genetic counseling (Lord *et al*., 2020).

However, the researchers warn that ASD cannot be understood exclusively by genetics. Environmental factors, such as antenatal complications, exposure to pollutants, and maternal infections, interact with genetic predisposition to modulate risk (Karimi *et al*., 2017). Thus, understanding ASD requires a multifactorial approach, integrating genomic, epigenetic, and environmental data.

Recent literature points to the need for broader and more diverse studies, especially involving populations underrepresented in current research, in order to expand the generalizability of findings and reduce biases (Grove *et al*., 2019). The integration of large international genetic databases, combined with advances in artificial intelligence for data

analysis, promises to accelerate discoveries and open new frontiers in understanding the biology of autism.

In summary, the evidence points to ASD resulting from a complex network of genetic and environmental interactions, with multiple biological pathways converging to changes in brain development and functioning. The elucidation of these mechanisms is essential to advance in more effective diagnostic and therapeutic strategies.

Studies of the autism genome have shown that although *de novo* mutations occur in many different genes, these genes tend to form densely connected protein-protein interaction networks (PPI) converging on common biological pathways, like a cascade. By integrating data from six studies using *StringDB* (an online database that gathers information on interactions between proteins), it was observed that networks based on mutated genes in probands were significantly more connected than random networks or networks based on sibling or synonymous mutations, showing that the fruit of mutations does not come from chance, nor from a family bias. In addition, two major functional blocks were evidenced: one of postsynaptic synaptic proteins (e.g., SYNGAP1, DLG4, GRIN2A/B, NLGN1, NRXN1) and another involving WNT signaling (CTNNB1, DLL1, TBL1XR1) and chromatin remodeling (CHD8), suggesting that heterogeneous mutations converge to central processes in neuronal development and functioning (Krumm *et al*., 2014).

On the other hand, population and twin studies show that ASD has a strong genetic basis, with an estimated heritability of 83% to 87%, indicating that most of the variation in the phenotype is explained by additive genetic factors. Non-shared environmental influences contributed about 17% of the variation, while the effect of shared household environmental factors was small or negligible (Sandin *et al*., 2017).

De novo *mutations*, formed spontaneously in the individual and absent in the parental genome, represent an important source of genetic variability associated with Autism Spectrum Disorder (ASD). These mutations, as well as those inherited from affected parents, are more likely to exert a pathogenic effect. However, the contribution of variants inherited from phenotypically healthy parents to the manifestation of ASD cannot be excluded, since such alterations may present incomplete penetrance and variable expressivity (Ribeiro, 2013; Coutinho, Bosso, 2015).

The occurrence of point mutations in the germline has been associated with advanced parental age, especially paternal age. With aging, there is a progressive accumulation of mutations in germ cells, which can be transmitted to the embryo during conception. This process may also favor the emergence of new Copy *Number Variations (CNVs*), so that older

parents constitute a potential reservoir for such mutational events. In addition, mutations originating in the male germline tend to have greater penetrance (Coutinho, Bosso, 2015).

Several studies have identified the participation of multiple genes associated with the risk of developing ASD, evidencing the polygenic and heterogeneous nature of the disorder. Among the genes described in the literature, MDGA2, FHIT, HTR2A, SHANK2, GRIA3, ZNF778, PRKCA, CDH15, DIAPH3, GCH1, GRM5, MARK1, SLC17A6, IMMP2L, BZRAP1, SYNGAP1, ANK3, MAP1A, GABRR2, LAMC3, LRRC7, LRRIQ3, CADPS1, NUFIP, SEMA3A, SNAP29, MBD2, GAD2, DGKH, and PARD3 stand out, which participate in fundamental processes for the functioning of the nervous system, including synaptic organization, neurotransmission, neuronal plasticity, and brain development (Ribeiro, 2013).

In addition to changes in the DNA sequence, epigenetic mechanisms also play a relevant role in the pathophysiology of ASD. DNA methylation (mDNA) consists of an epigenetic modification characterized by the addition of a methyl group ($-CH_3$) to position 5 of the cytosine base, resulting in the formation of 5-methylcytosine (5mC). This process regulates gene expression without altering the nucleotide sequence and allows genetic and environmental factors to act together in modulating the phenotype. Thus, changes in methylation patterns can influence the expression of genes involved in neuronal development and function, contributing to the clinical manifestation of ASD (Martin, Fry, 2018; Smith *et al*., 2014).

A Genetic Ontology analysis of ASD risk genes revealed enrichment in neuronal signals, neurogenesis, chromatin remodeling, and transcriptional regulation, indicating that these genes are clustered in specific biological processes (Lin *et al*., 2020). These findings complement the results of protein–protein interaction networks, which show that different but interconnected genes act on common functional pathways related to brain development and function.

Recent studies suggest that mosaic somatic mutations, which affect only part of the body's cells, contribute modestly to autism risk, accounting for about 5–7.5% of *de novo mutations*. These mutations can occur in genes that are different from those identified in germline studies and influence the phenotype differently. Postmortem prefrontal cortex sequencing has identified potentially harmful somatic mutations, including changes in regulators of gene expression, indicating that these mutations may affect the underlying neurobiology of ASD (Dias *et al*., 2020). Future studies with brain DNA and analyses of non-coding regions are needed to better understand the contribution of somatic mutations to the pathogenesis of autism.

5 FINAL CONSIDERATIONS

Autism was long considered to be of low prevalence (1/1000 children) in the population. Today this rate is 1/160, and it is quite likely that it will rise in the coming years (Masini, 2020). Autism is a very heterogeneous condition, with atypical social characteristics with restricted and stereotyped interests (Bourgeron, 2015). Population studies of twins have shown that some autism traits have a strong correlation with genetics and neurodevelopmental diagnosis. These studies reveal that rare variants with a large effect, as well as common variants with a small effect, contribute to the increased risk of autism in the population, challenging traditional diagnosis, as there is a large clinical heterogeneity (Thapar, Rutter, 2021). Clinical heterogeneity is related to a great complexity that involves point mutations that have been inherited or spontaneous. More than 100 risk genes are involved from rare or frequent mutations that potentiate symptoms. Changes in these genes are responsible for a substantial risk to the individual and a small proportion to the population.

Most of the risk is attributable to common variants that are inherited and that act together, bringing a summation effect. Alterations in these genes normally converge to the same mechanisms, involving gene regulation events and synaptic connectivity. These mechanisms could be involved in epigenetic lack of control (Havdahl *et al*, 2021). The identification of the regions of the genome or these genes contributes to understanding a little more about ASD. However, despite many studies and an extensive list of risk genes, we have not yet observed a transformative impact for understanding the nature of genetic risk for a variety of psychiatric disorders (Devanand, *et al*, 2020).

The expansion of scientific knowledge about Autism Spectrum Disorder has highlighted the need for interdisciplinary approaches that integrate different areas of knowledge, such as genetics, neuroscience and psychology. Recent studies have shown that ASD has a strong hereditary component and great genetic heterogeneity, involving both common and rare variants that affect processes such as synaptic plasticity and neuronal development (Bourgeron, 2015; de Rubeis; Buxbaum, 2015). In this context, the advancement of genetic research has contributed to a broader understanding of the etiology of the disorder and to the development of more accurate diagnostic and therapeutic strategies.

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