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# AUTISM SPECTRUM DISORDER AND EARLY DIAGNOSIS: A NARRATIVE REVIEW OF THE LITERATURE

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## ABSTRACT

Objective: To analyze autism spectrum disorder and early diagnosis: Literature Review: Autism is defined as a complex developmental disorder, from a behavioral point of view, with different etiologies that manifests itself in varying degrees of severity (GADIA, 2006). The characteristics of the spectrum are persistent impairments in communication and social interaction, as well as in behaviors that may include interests and activity patterns, symptoms that are present since childhood and limit or impair the individual's daily functioning (APA, 2014). The subcategories are part of ASD, and impairment can occur at three levels of severity. Final considerations: The importance of early diagnosis of ASD is highlighted, which tends to seek and collect more information about the established diagnosis. It is understood that the earlier the child is diagnosed and starts treatment, the greater the possibilities of development within their physical and mental capacities. The choice of appropriate treatment is extremely important, as ASD accompanies the individual throughout his or her life.

**Keywords:** ASD. Diagnosis. Mental Disorder.

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## INTRODUCTION

Autism is defined as a complex developmental disorder, from a behavioral point of view, with different etiologies that manifests itself in varying degrees of severity (GADIA, 2006). According to Oliveira (2009), "autos" means "one's own" and "ism" translates a state or an orientation, that is, a closed person, reclusive in himself. Thus, autism is understood as a state or a condition, which seems to be secluded in itself. The term "autism" has undergone several changes over time, and is currently called Autism Spectrum Disorder (ASD) by the Diagnostic and Statistical Manual of Mental Disorders (DSM-V) (APA, 2014).

The characteristics of the spectrum are persistent impairments in communication and social interaction, as well as in behaviors that may include interests and activity patterns, symptoms that are present since childhood and limit or impair the individual's daily functioning (APA, 2014). The subcategories are part of ASD, and impairment can occur at three levels of severity. At level one, the individual demands support; at level two, it requires substantial support; and at level three it requires a lot of substantial support (APA, 2014).

The individual with ASD, according to the DSM-5 (APA, 2013), is characterized by presenting a compromised or markedly abnormal development of social insertion and communication and a very restricted repertoire of activities and interests. The manifestations of the disorder vary immensely, depending on the level of development and chronological age. Delay can occur in at least one of the following areas: social interaction, communicative language, symbolic or imaginary games (BAGAROLLO; BROOK; PANHOCA, 2013).

Although it is believed that environmental factors, such as infections or the use of certain medications during pregnancy, play a role in the development of the disorder, it is estimated that ASD is hereditary in about 50 to 90% of cases, which demonstrates the importance of genetic factors in the pathogenesis of the disease. Understanding the genetic aspects involved in a disease provides valuable information about the risk of recurrence, prognosis, and possible therapeutic interventions. Thus, all the work undertaken in recent decades to better understand the genetic factors associated with ASD has greatly improved diagnostic accuracy and genetic counseling for the disorder. (BAGAROLLO; BROOK; PANHOCA, 2013).

## LITERATURE REVIEW

ASD is considered a genetically heterogeneous and complex disease, as it has different inheritance patterns and causal genetic variants. To understand the currently defined genetic architecture of ASD, it is important to consider epidemiological and evolutionary aspects, as well as all available knowledge about the molecular alterations related to the disease. First, we must consider a primordial evolutionary rule that influences the frequency of genetic variants present in the population: if a certain genetic variant has a harmful effect on the organism and negatively affects the reproductive chance of individuals (their reproductive potential), this variant tends to have a low frequency in the population, since it will not be transmitted to the next generations.

In fact, this is what happens in most monogenic diseases: they are generally rare in the population due to the low frequency of their causal alleles. According to this assumption, if a disease that reduces adaptability is common in the population, it is unlikely to be caused by a single variant with an extremely deleterious functional effect. For this reason, it is assumed that common diseases with genetic components have a polygenic or multifactorial inheritance model (genes combined with environmental factors) and are therefore caused by the inheritance of a combination of genetic variants, each associated with a low risk of developing the disease.

Parents of individuals with ASD are usually the first to notice that something different is happening to their child. At this moment, the search for help begins, and a period of uncertainty precedes the process of elaboration and formation of the diagnosis. However, it is worth noting the importance of the way this diagnosis is made by the parents of children with ASD. Schulman (2002) states that, as soon as the diagnosis is communicated, it is important to direct parents to useful resources, which helps in the feeling that there is something to do. Among the resources referred to by this author for understanding the disorder are readings about the diagnosis, in order to understand the symptoms of ASD, which vary greatly depending on the case. It is known that, in autism, "not everyone is the same and not everyone has the same characteristics. Some can be more attentive, some more intellectual and others more sociable, and so on" (FERREIRA, 2009, p. 15)

. It is known that there are few instrumental resources for diagnosing individuals with suspected autism and, even with many studies in the area, there is no biological

marker that allows an accurate test to confirm or not this diagnosis. Another important factor to be highlighted is the way in which the communication of the diagnosis of autism to parents will occur. It is a delicate process, which provides a unique opportunity for professionals to establish an alliance of trust with them, and so that they can make the diagnosis in the most coherent and less stressful way (BOSA; SEMENSATO, 2013).

It is up to parents to understand, therefore, the real needs of their child and accept their differences, so that they can put aside the fear of being inadequate, seeking help and information about their condition. Thus, the earlier the child is treated and diagnosed, the greater the chances of their development happening in the best possible way. However, sometimes in practice it is known that it does not occur properly

In 2013, the Brazilian Ministry of Health, in partnership with the Unified Health System (SUS), created a booklet called "Guidelines for Attention to the Rehabilitation of People with Autism Spectrum Disorders (ASD)". The main objective of these guidelines is to highlight the guidelines for multiprofessional teams for the health care of individuals with ASD and their families, at the different points of care in the care network for people with disabilities. For its elaboration, the International Code of Functionality and Disability (ICF) and the international systems of the International Statistical Classification of Diseases and Related Health Problems (ICD-10) (MINISTRY OF HEALTH, 2013) were used.

As ASD has no cure, the search for specific treatment carries with it, however, the importance of mitigating the deficits presented, as some treatments may be more effective for some and less for others, due to each autistic person presenting a different level of development from the other. However, with regard to treatment, behavioral psychotherapy is still the most recommended, along with the conditioning process that facilitates the care of the autistic person, making him better emotionally structured and organized (SANTOS, 2008).

Psychotherapy aims to help interpret body language, non-verbal communication, learning, and also emotions and social interactions (BARROS; SENRA; ZAUZA, 2015). Cognitive behavioral therapy (CBT) contributes to teaching autistic people about different ways of using, remembering and processing information, such as self-instruction training (WHITMAN, 2015).

## FINAL CONSIDERATIONS

The importance of early diagnosis of ASD is highlighted, which tends to seek and collect more information about the established diagnosis. It is understood that the earlier the child is diagnosed and starts treatment, the greater the possibilities of development within their physical and mental capacities. The choice of appropriate treatment is extremely important, as ASD accompanies the individual throughout his or her life. Like any individual, the autistic person is unique within his uniqueness, and the results of this treatment will be variable.

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