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ORIGINAL ARTICLE

The role of the pediatrician in accurate recognition of autistic spectrum disorders and symptoms: literature review

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Abstract

Introduction: Autism spectrum disorder (ASD) is a neurodevelopmental disorder, diagnosed by disruption in the process of socialization, behavior, and communication. It is among the most common developmental disorders, affecting 1 in 100 children. Its subtle, symptomatic, and chronic profile has the involvement of essential biological mechanisms related to development. **Objectives:** Due to the low scope of the theme in the curriculum of the Medicine and specialization in Pediatrics course, which makes early diagnosis difficult, this review seeks to develop knowledge about history, clinical characteristics and diagnostic tools associated with ASD. **Methodology:** A systematic review used the databases: Medline (PubMed), SciELO, LILACS and Google Scholar, using "Autism and the pediatrician" and "Autism and the pediatrician", "Autism" as a search strategy. **Result and Discussion:** Through the CHAT (Checklist for Autism in Children), health professionals can make diagnostic suspicion, through parents reports on the childs behavior, as well as at the time of the consultation. **Conclusion:** The early introduction of therapies to rehabilitate children with ASD, through interdisciplinary intervention, is important for improving the behavior and quality of life of patients with autism spectrum.

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INTRODUCTION

The word "autism" is derived from the Greek word autos, which means the individual himself. There are several descriptions of autistics in the literature, and the first clinical descriptions were detailed by Leo Kanner, an American pediatrician, who published an article entitled "Autistic Disorders of Affective Contact" in 1943[1], based on the observation of 11 cases, which were initially diagnosed as an uncommon and early form of schizophrenia. What he noticed as unusual was the fact that a portion of the children had these symptoms from birth onwards[1-4].

Autism would be defined as a behavioral syndrome, present from the early stages of life, characterized by a deficiency in social interaction, language, communication, and activities. Currently called Autism Spectrum Disorder (ASD). The new classification of the Diagnostic and Statistical Manual of Mental Disorders (Diagnostic and Statistical Manual - DSM V) has two symptom domains: Restrictive social communication and repetitive behaviors. Considering the severity of symptoms and level of impairment, these abnormalities are usually present in early childhood and are likely to be seen in all different contexts[4-7].

The etiology of ASD remains uncertain. There are several hypotheses that involve environmental factors, genetic factors, virus involvement, exposure to chemical substances, and even obstetric complications. Regarding genetic factors, studies show that there is familial recurrence, with the highest incidence in monozygotic twins compared to dizygotic twins. It is possible that several genes with small effects may act through an epigenetic mechanism and environmental factors on phenotypic expression. In less than 10% of ASD cases, it is associated with a known medical condition, but causal mechanisms are not well understood[1].

Regarding epidemiology, ASD is not uncommon. The National Institute of Health and Clinical Excellence (NICE) states that the diagnosis is made in approximately 3% of the population, and new epidemiological studies suggest minimum prevalence rates of 1 per 100 inhabitants. Frequent in males, with a sex ratio of 3 to 4:1, associated with epilepsy and mental retardation indicate possible neurobiological bases of, not only a purely psychogenic cause, but as a neuropsychological developmental disorder, it is emphasized that when women are affected, the impairment is usually more severe[3,4,6].

In the present literature review, we sought to understand how pediatricians should pay attention to the diagnosis of ASD and its early signs. We propose to elucidate the knowledge about the early signs of the disease, understand the possible interventions to be indicated, discussing the importance of early diagnostic suspicion.

MATERIAL AND METHODS

An extensive search was carried out in the medical literature, extracting information from national and international literature. Medline databases (through PubMed), SciELO, LANCET, LILACS and Google Scholar were used to carry out this systematic review. Using the following search strategy: "Autism and the pediatrician", "Autism", "Autism and the pediatrician", 60 articles were found. Forty-three articles that did not meet the pre-established inclusion and exclusion criteria were excluded (Table 1).

RESULTS

The total number of studies that met the inclusion criteria, and that were included in the analysis of the results of this review, was 17. For didactic purposes, we divided the results through a table, summarizing the articles and their objectives (Table 2), clinical features, differential diagnosis, course and prognosis and medical management.

Clinical features

ASD is not a single disease, but a complex developmental disorder that is defined from a behavioral point of view with multiple etiologies and varying degrees of severity. The clinical picture has three symptomatic axes, they are in the social area (interpersonal relationship); in communication (verbal and non-verbal) and in behavior (restricted, repetitive, and stereotyped interests and activities). The onset of symptoms may become apparent in the first year of life, or normal development may occur until 12-18 months of age, and then regression of language and/or social skills ensues, which occurs in up to 30% of cases.

More typically, there is a developmental arrest after 6 months of age, such as a plateau, or there is a deceleration of development accompanied by some loss of social communication skills, such as joint attention, shared affection, and use of language. Children can show a range of behaviors, parents report that there is something "wrong" with their baby. Infants tend not to look at their parents when they want to show something, not point to an object, or not use their imagination in games, sometimes preferring to play alone.[8-10]

As for the social aspects, behaviors that can draw the attention of parents and educators are: showing resistance when they are hugged or kissed, crying alone in the crib instead of calling for someone, not raising their arms when someone wants to pick them up and they can not noticing when parents leave or return home.[9,10]

Regarding the aspects of communication, when they need something, they usually use the resource of "directing the hand" of a person to the desired object. Parents are used as tools to get what they want, unlike other children, who use verbalization. Two-thirds of infants with ASD do not acquire functional speech, from absolute silence to some dialogue, and if they do, it is delayed. They have great difficulty tolerating stimuli such as music, noises, different textures, new experiences or environments. They often appear to have a hearing loss, not reacting to loud noises or not responding

Table 1. Inclusion and exclusion criteria in the systematic review on autism and pediatrician management.

INCLUSION CRITERIA	EXCLUSION CRITERIA
1- Studies with ASD in infants, preschool and school children	1- Studies in adults
2- Articles on medical conduct in the face of ASD	2- Studies that include diseases other than TEA.
3- Studies on the pathophysiology and treatment of ASD	3- Articles on ASD behaviors of other professions besides doctors and nurses
4- Studies in Portuguese and English.	4-. Studies of other approaches to the patient with ASD, in addition to the health area.

Table 2. Results of the literature review.

AUTHOR/YEAR	GOALS	YEAR OF PUBLICATION
STEINER *et al.*[2]	It sought to expand the knowledge of Autism in terms of definition, history, and diagnosis.	2016
STEINER *et al.*[3]	Review the literature on autistic disorders, highlighting the clinical, genetic and neurological aspects, as well as the main differential diagnoses. To define, based on literature data, the most appropriate complementary exams for etiological investigation and the functional diagnosis of cases, as an initial diagnostic hypothesis of autism. Develop a protocol for systematic diagnostic investigation in cases with an initial diagnostic hypothesis of autism.	1998
LAI *et al.*[4]	Definitions of autism over the years. Define and highlight the role of society regarding the quality of life of autistic people. Prevalence of autism in the world.	2014
RAPIN *et al.*[5]	It aims to cover the early diagnosis of autism, in addition to citing treatments based on the literature, from pharmacological to multidisciplinary. Mention adjunct comorbidities to autism such as ADHD and seizures.	1991
YATES *et al.*[6]	It addresses the diagnosis of ASD and provides an assessment framework for professionals who encounter a child with suspected autism.	2012
KANNER *et al.*[1]	To describe the cases of eleven children with autism, from the behavior and education of each child, as well as the educational background of the children's parents. This is the first description of autism as a unique disease concept, distinct from schizophrenia, treating early childhood autism as a disorder resulting from abnormal brain development in autistic children.	1945
ZAMPIROLI *et al.*[8]	Discuss about the symptomatology in infants and preschool children with suspected autism and the treatment.	2012
MANUAL ON AUTISM[9]	Quick and objective reference for doctors and therapists regarding the diagnosis of children with the spectrum autistic.	2016
BUIE *et al.*[11]	Generate evidence-based recommendations for diagnostic evaluation and treatment of gastrointestinal problems in this patient population.	2010
KLIN *et al.*[12]	It focuses on the history, nosology and clinical and associated characteristics of the two most well-known pervasive developmental disorders – autism and Asperger syndrome.	2007

LORD *et al.*[13]	Review current knowledge about neurobiological aspects of autism and generalized developmental disorders, as well as provide pediatricians with information on autism diagnosis and treatment.	1994
LORD *et al.*[14]	Comparisons were made between male and female children with autism, 384 boys and 91 girls, ages 3 to 8 years, on nonverbal measures of intelligence, adaptive functioning, receptive vocabulary, hand-eye perception and integration, and affectation ratings. , game, relationship and human interest.	1982
KLIN *et al.*[15]	It focuses on the history, nosology and clinical and associated characteristics of the two most well-known pervasive developmental disorders – autism and Asperger syndrome.	2006.
FLORES *et al.*[16]	To understand how pediatricians and neuropediatricians from a city in the central region of Rio Grande do Sul notice autism and its early signs. In addition, there are proposals to elucidate physicians' knowledge of early signs of autism; to understand the possible interventions carried out by them when they identify premature signs or diagnose autism in children; to describe the proposal of the psychoanalytic theory regarding the possibilities of early intervention in the face of the risk of autism, as well as to discuss the benefits and harms of early intervention in babies.	2013
SERRA *et al.*[17]	Briefly present different educational programs that guide the attempt to include autistic students and investigate the feasibility of applying such programs in public schools.	2010
ALMEIDA *et al.*[10]	There has been an increase in the prevalence of ASD in recent years in several countries. Review the epidemiology, etiology, differential diagnosis and treatment of ASD in light of recent advances published in the scientific literature.	2018
Diagnostic and Statistical Manual of Mental Disorders: DSM-5[7]	Define how the diagnosis of mental disorders is made. Used by psychologists , doctors and occupational therapists . It has been one of the most used mental health diagnostic bases in the world.	2014

ADHD: Attention deficit hyperactivity disorder; ASD: Autism Spectrum Disorder

when called by name, but in other situations, their hearing appears to be normal.[9,16]

The higher the level of sensory exposure, the greater the chance of a behavioral change. The sensory aspects that distinguish them from other children are: extreme difficulty in cutting their hair, they may not like new experiences, such as birthday parties, so programming routine daily activities is usually difficult when you have a child with the Spectrum. autistic. The child may go for days without demonstrating the need for sleep, do not seem to notice the difference between day and night (behavior that usually appears at 4 months of age), has difficulty initiating sleep, sleeping for a short period of one or two hours.[5,9-11]

Stereotypes include swaying the hands or arms insistently or rocking the body back and forth. They can also look for ways to put pressure on the body, look for ways to cause strong impacts, such as jumping from a certain height.[9,12,13]

Regarding safety aspects, these children tend to have no sense of danger, being bitten by dogs, being burned on the stove, or being run over.[12.13]

In short, when should the pediatrician think about ASD? Pediatricians must remain attentive to the warning signs of impairment, development, and social communication in the first years of life:

1. Absence of vocalization at 6 months of age;
2. Absence of babbling syllables with consonants at 12 months of age;
3. Absence of gesture communication at 12 months of age; for example, the child does not point to a desired object or look at someone else's index

finger;

4. Speech does not include single words, other than "mama" and "dada", spoken spontaneously at 16 months of age;
5. Speech does not include sentences of two words at 24 months of age or of three or more words at 36 months of age;
6. Regression or stagnation of developmental milestones, from the loss of verbal and non-verbal communication skills [15,16].

Pathologies associated with ASD.

Gastrointestinal disorders are intensively studied by Dr. Tim Buie[11], Gastroenterologist at Harvard University and the LADDERS program, General Hospital, Boston, who has performed endoscopy on over 1000 children with ASD. In the first 400 children, autistic children were found to have more gastric changes than the control group. Being 20% - Esophagitis, 12% - Gastritis, 10% - Duodenitis, 12% - Colitis and 55% - Lactose deficiency. We can also mention toxoplasmosis, cytomegalovirus, microcephaly, hydrocephalus, West syndrome, tuberous sclerosis, celiac disease, adrenoleukodystrophy and Duchenne muscular dystrophy.[11]

Differential diagnosis

They make the differential diagnosis of ASD: Mental retardation (there is socialization in interpersonal forms, and it is important to note that 70 to 86% of patients with ASD are also mentally handicapped), schizophrenia (rare in childhood, especially before the age of five, does not present with echolalia or lack of speech), Landau-Kleffner syndrome (acquired aphasia) that exhibits on electroencephalogram, epileptiform activity with continuous slow-wave spike morphology during slow-wave sleep.[8,10,12-15]

Risk factors

Risk factors for ASD that still need further investigation are siblings with ASD; parental-type schizophrenia with psychosis or affective disorder; use of sodium valproate by the mother during pregnancy; gestational age less than 35 weeks; intellectual disability; birth defects associated with the central nervous system, including cerebral palsy; Down syndrome (trisomy 21); Fragile X Syndrome. As well as other medical conditions associated with ASD: Neurofibromatosis; untreated phenylketonuria; Fetal Alcohol Syndrome; Congenital rubella and iron deficiency anemia[6].

Course and prognosis

There may be several factors as predictors of course and long-term outcome, particularly the presence of some communication language, around age five or six, non-verbal intellectual level, severity of condition, and response to educational intervention. The intelligence of autistic people varies widely. In most cases, mental impairment is obvious, but some have superior ability for different skills such as calculation, puzzle-solving and verbal memory, about 75% or even more have an IQ of less than 70. There is not yet reliable evidence that the propensity to autism, once its risk is perceived, can be reversed, however, what has been seen is that numerous cases evolve in a favorable way.[2,13,12,16]

In relation to the autistic child's family, whatever the socioeconomic level of the family, there is always a search for a miraculous cure, and a belief in practices of dubious seriousness. From the news to the acceptance of reality, the path is long and tortuous and not all parents are able to overcome the moment of grief for a healthy child. Especially, due to the absence of affective exchange and communication, which is usually the biggest difficulty. Autistics have specific difficulties in understanding many of the human feelings. They appear to have no feelings, but in fact this behavior appears to be the result of cognitive inability. Prognoses about the future of the autistic child can become less obscure, and the idea that the child can accomplish nothing can be replaced by conscious hopes and investments in the child's development. The family, when well advised, becomes an important factor, as it can insert social rules, as well as establishing communication procedures with the child.[6,17]

ASD exhibit behaviors and presentation that vary over time, with a tendency to progress across domains, although there is great individual variation. Outcomes can be determined by severity of behaviors, cognitive skills, and useful expression. Many people need specific support, adults with a higher degree of ASD may be able to live independently and obtain a job, but at the present time few adults seem to reach their full potential. This may reflect the lack of appropriate social support in this group. Recent studies have reported some encouraging findings for individuals who received early intervention focused on skill development. However, it is uncertain to say that, with early diagnostic suspicion, there will be an impact on the long-term results for individuals with ASD. Thus, the clinical investigation begins with a complete anamnesis, with special attention to the risk factors described above, and a complete physical and neurological examination, with an emphasis on looking for dysmorphisms and signs of potentially implicated genetic syndromes. Screening for visual and hearing impairment should be included[7,16,17].

From the observation of one or more warning signs, the pediatrician can use a diagnostic screening instrument, such as the M-CHAT scale (*Modified Checklist for Autism in Toddlers*), which can be applied between 16 and 30 months of age or follow guidelines updated in 2013 by the *American College of Medical Genetics* (Table 2). There are two additional tools that strengthen the diagnosis: the Autism Diagnosis Interview (ADI-R, *Autism Diagnostic Interview)* and the Autism Diagnosis Observation Scale (ADOS, *Autism Diagnostic Observation Schedule*)[10].

If there is developmental regression, an electroencephalogram (EEG) prolonged for 6-8 hours, ideally at night, should be obtained to record deep sleep.[2,4,5]

Medical conducts

Some specific comorbidities, such as epilepsy or attention deficit disorder, seem to respond well to drug therapy, similarly to what occurs in non-autistic children. In any case, the only treatment available, so far, is multidisciplinary therapy, if possible, bringing together a Neurologist, Psychiatrist, Geneticist and Gastroenterologist, Speech-Language Pathologist, Occupational Therapist, Psychologist, Pedagogue, specialized educators and other specialties, depending on each case which despite determining some improvement, is still far from satisfactory in most cases. In general, there is no cure, however, some high-functioning autistics, if well managed, continue with a life very close to normality, and are highly functional, despite some impairment of socialization. In addition to these, family members play a fundamental role, participating as "co-authors" of the treatment, monitoring the child's progress day by day. The treatment can rely on contributions from language/communication therapies and school "experience"[4,8].

Early and intensive intervention brings great benefits to the child and family. Initial strategies aim to teach children to perceive what is happening around them, to be more attentive, to imitate acceptable behaviors, to develop communication skills, among others[9].

FINAL CONSIDERATIONS

Through this systematic review of the literature, we can observe autism spectrum disorder as a complex neuropsychological developmental disorder that affects children since early childhood. Its diagnosis is predominantly clinical, requiring full attention from health professionals, especially pediatricians, and early diagnostic suspicion is of great value for rapid diagnosis, and *a posteriori,* introduction of therapies to rehabilitate the autistic child, through therapy. multidisciplinary approach, it is important to improve the behavior and quality of life of these patients.

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