

Contribuição dos autores: SSR concepção, planejamento do projeto de pesquisa, obtenção, análise/interpretação dos dados, elaboração do manuscrito. ALBM interpretação dos dados, discussão dos achados, elaboração do manuscrito e revisão crítica. VFC análise/interpretação dos dados. ACCC obtenção, análise/interpretação dos dados e redação e revisão crítica do manuscrito. MCO obtenção, análise/interpretação dos dados e redação e revisão crítica do manuscrito. ACFC concepção, planejamento do projeto de pesquisa e redação e revisão crítica do manuscrito.

Agradecimento: Aos pacientes e famílias que participaram da pesquisa.

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Conflito de interesses: Não

Financiamento: Não há

Recebido: 03/01/21
Aprovado: 14/01/23



O contexto diagnóstico de indivíduos com autismo leve

The diagnostic context of individuals with mild autism

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RESUMO

Introdução: A Síndrome de Asperger (AS) é um distúrbio do neuro desenvolvimento; parte do diagnóstico definido como Transtorno do Espectro Autista (TEA), também considerado como autismo leve, segundo a nova classificação do Manual Estatístico e Diagnóstico de Doenças Mentais (DSM-5). Indivíduos com AS apresentam déficits sociais e comportamentos estereotipados. **Objetivo:** Investigar o contexto e trajetória do diagnóstico de AS, desde a idade de percepção dos sintomas, idade da primeira avaliação clínica, a hipótese diagnóstica que precedeu o diagnóstico conclusivo, período de tempo entre a percepção dos sintomas e o diagnóstico conclusivo, a especialidade do profissional que diagnosticou e as principais recomendações recebidas no diagnóstico. **Casística e Métodos:** Quarenta e quatro indivíduos; 38 homens e seis mulheres foram investigados por meio de avaliação clínica e entrevista semiestruturada. **Resultados:** A idade da percepção de que havia algo de diferente variou de um a nove anos (média=3.2 anos), e os primeiros a percebê-lo foram os pais (57%), e os últimos foram os profissionais da saúde (7%). A primeira avaliação diagnóstica foi realizada entre um e 15 anos de idade. O diagnóstico inicial mais comum foi de Transtorno de Déficit de Atenção e Hiperatividade (34,1%) realizado na maioria parte pelo neurologista (54,5%). O tempo médio entre a percepção de que havia algo de diferente no desenvolvimento da criança e o diagnóstico de SA foi de 7,8 anos, realizado por psiquiatras na maioria dos casos (61,3%). As principais recomendações foram frequentar escola regular (68,1%), realizar psicoterapia (59%), usar medicação (56,8%), realizar Fonoterapia (38,6%) e avaliação genética (36,3%). **Conclusão:** Os resultados mostram que o contexto do diagnóstico de AS faz-se tardio; mesmo do fenótipo comportamental considerado mais brando é complexo. A maioria dos profissionais da saúde não está preparada o suficiente para detecção precoce de SA, o que também compromete a intervenção imediata, isto é, essencial nestes casos.

Palavras-chave: Síndrome de Asperger; Diagnóstico Precoce, Transtorno do Espectro Autista.

ABSTRACT

Introduction: Asperger's Syndrome (AS) is a neurodevelopmental disorder; part of the diagnosis defined as Autistic Spectrum Disorder (ASD), also considered as mild autism, according to the new classification of the Statistical Manual and Diagnosis of Mental Diseases (DSM- 5). Individuals with AS have social deficits and stereotyped behaviors. **Objective:** To Investigate the context and trajectory of AS diagnosis, from the age of symptom perception, age of the first clinical evaluation, diagnostic hypotheses that preceded the conclusive diagnosis, time between symptom perception and conclusive diagnosis, = mainly the professional who performed the diagnosis and main recommendations received at diagnosis. **Casistics and Methods:** Forty-four individuals, 38 men and six women were investigated through clinical evaluation and semi-structured interview. **Results:** The age of a behavioral discrepancy perception ranged from one to nine years old (mean=3.2 years) and the first to notice this were the parents (57%), and health professionals were the last (7%). The first diagnostic evaluation was carried out between one and 15 years of age with the most common initial diagnosis as Attention Deficit Hyperactivity Disorder (34.1%) performed mostly by the neurologist (54.5%). The average time between the perception that there was something different in the child's development and the diagnosis of AS was 7.8 years performed by a psychiatrist in most cases (61.3%). The main recommendations were attending regular school (68.1%), undergoing Psychotherapy (59%), using medication (56.8%), undergoing Speech Therapy (38.6%) and genetic evaluation (36.3%). **Conclusion:** The results have showed that the context of AS diagnosis is late; even the behavioral phenotype considered milder is complex. Most health professionals are not enough prepared for early detection of AS, consequently, compromising immediate intervention, that is essential in these cases.

Keywords: Asperger Syndrome, Early Diagnosis, Autism Spectrum Disorder.

INTRODUCTION

Asperger's syndrome, first described by the Russian psychiatrist GrouniaEfimovna Soukhareva¹ and the German psychiatrist Asperger (1943)², is a neurodevelopmental disorder that is currently part of the diagnosis of Autistic Spectrum Disorder (ASD), according to the latest edition of DSM-5³.

Individuals with Asperger's syndrome have difficulties in social interactions, verbal and non-verbal communication, and may exhibit narrow stereotypes and interests⁴.

Some patients with Asperger's syndrome do not meet the new ASD criteria in the DSM-5⁵; therefore, they do not receive proper treatment, while others have claimed that the new inclusion

of Asperger's syndrome in the category of autism spectrum disorders, without any distinction, negatively affects=impaired the identity of these individuals⁶.

However, Asperger's syndrome has a well-defined clinic, with a rich semiology and behavioral presentations; that is, distinctive⁷. Worldwide, teams working with Asperger's syndrome patients have been continuing to use this diagnosis as a subtype of ASD without delay of language and a normal or higher IQ⁸. The prevalence of Asperger's syndrome is estimated at 0.06% in the general population⁹. Some psychiatric comorbidities are frequent, such as schizophrenia, obsessive-compulsive disorder, attention deficit hyperactivity disorder, schizoid personality disorder, intellectual disability, anxiety disorder, social phobia, learning disorder and mood bipolar disorder should be considered in the differential diagnoses¹⁰.

This study has considered the challenging obstacles to the diagnosis of ASD, in which SA can be adequate. In general, ASD can be identified as early as 16-24 months, a significant proportion of children are not diagnosed or treated until the school years¹¹⁻¹³. However, mild ASD conditions, such as AS, can present more difficulty on early diagnosis; causing professionals to be insecure about closing the diagnosis quickly. Mild conditions of ASD can provide concern among professionals in giving "false positives", that is, including children with different mental disorders as autism and, thus, leading to a significant increase in diagnoses¹⁴.

A delay in diagnosis is very common in low and middle-income countries as shown in a study conducted in Venezuela, and recently in Brazil. In both countries, the perception of the first symptoms was similar, at around two years of age, but the diagnostic delay was about 2.2 years in Venezuela and three years in Brazil¹⁵⁻¹⁶. As in countries such as Colombia, India, Jamaica, Jordan and Mexico, the average age of the diagnosis was late from 3.7 to 4.7 years old¹⁷. In high-income countries, such as the USA, for children diagnosed with ASD at age four, the average age of the first assessment was 2.2 years old, as compared to 2.6 years old for children aged eight¹³. In the United Kingdom, the most common age ranges from two to five years old for diagnosis of children with ASD.¹⁸

Accordingly, the high frequency of comorbid or associated disorders; in most cases, the unknown etiology as well as the lack of biomarkers further complicate the diagnostic process, which usually is not reached until the child starts school¹⁹. In individuals with better social and cognitive performance, the diagnosis may delay until early adolescence, when the pressures of social groups can cause anxious and depressive or even psychotic states that can result in appointments with mental health professionals²⁰.

When the SA diagnosis as well as the start of interventions are delayed; the results are likely to be less promising. Hence, the delay of correct diagnosis can create a situation that affects not only the mental health of the individual with SA, but also the family members, causing direct harm, compromising the prognosis²¹. Taking into account these difficulties; this study has investigated some aspects of the context of diagnosis in which families and their children with SA have undergone, since the perception of symptoms until the diagnosis, followed by some of the consequences.

METHODS

This study was performed after approval of the Research Ethics Committee of FAMERP (001-005530/2009) and after obtaining informed consent, with a convenience sample of 44 individuals diagnosed with Asperger's syndrome at interview time with ages that ranged from six to 31 years old (standard deviation - SD: 6.35 years).

Thirty eight male (82%) and six female (18%) individuals were studied, all from the city of São José do Rio Preto, SP, and region.

ASD diagnosis was ascertained by a team of specialists, based on DSM-IV criteria²² and ICD 10²³. Only individuals without linguistic impairment, who had already received diagnosis of ASD, were studied, regardless of gender, age, ethnicity, origin, social status and personal or family history. Subjects whose diagnosis of SA was not confirmed, who presented a different birth defects (syndromic), whose parents or caregivers were unaware of the details of the diagnostic process or who did not have accurate information were excluded from the study. The use of any category of medicinal drugs, therapeutic stimulation or rehabilitation was not considered in the exclusion criteria.

After the diagnosis of the SA was confirmed; the parents were interviewed by psychiatrists (semi-structured interviews). A questionnaire designed by the researchers was used to obtain sociocultural data, information about neuropsychomotor development, personal and family history and about the whole process of establishing the SA diagnosis. The age when the symptoms were noticed; who initially noticed the symptoms; the age of the first clinical evaluation; diagnostic hypotheses that preceded the diagnosis interval between the perception of symptoms and the diagnosis were investigated. Also, the specialty of the professional who suspected SA, and the one who performed the diagnosis and the main recommendations received at the SA diagnosis were observed.

In all cases, the interview was conducted in the presence of the parents or caregivers of the individual with SA.

RESULTS

The age at which something different about the child was perceived (age, the symptoms were noticed) ranged from one to nine years old. The highest frequency of perception being at three years old (27.2%); the accumulated rate by three years of age was 65.9% and the mean age was 3.2 years old (SD = 1.88). The average age of suspicion of Asperger's Syndrome symptoms was 10 years old and the average age of diagnosis was 10 years and six months old.

The person or people who first realized that something was different in the child's development (who noticed the first symptoms) were both parents or just the mother (56.9%), followed by school staff (22.7%), other relatives (13.6%) and pediatricians (6.8%). For children, whose parents were the first to notice the child's atypical development; 80% had siblings, either older (56%) or younger (24%) and 20% had no siblings.

The age of the first clinical evaluation was on average 4.4 years old, i.e. approximately 1.2 years after noticing symptoms. At this time, the subjects were evaluated by professionals of one or more specialties. The patient has been examined by more than one professional or a multidisciplinary team in 59.1% of the cases. In the evaluation process, the most consulted specialist was the neurologist (28%), followed by psychologist (19.6%), psychiatrist (16.3%), speech therapist (15%), pediatrician (12.8%), otorhinolaryngologist, orthopedic doctor, ophthalmologist, physical therapist and multidisciplinary team (1.2%).

Before the diagnosis of SA, the majority of patients has not received any diagnostic hypothesis or received diagnoses which sometimes were wrong or partial, and that after were considered associations or comorbidities. In the first evaluation; (13) 29.5% of the individuals have not received any diagnosis or diagnostic hypothesis. Thirty-one (70.4%) of the individuals received a diagnostic or diagnostic hypothesis. The most frequent initial diagnosis (34.1%) of the individuals was ADHD, alone or combined with up to four other diagnoses. Only seven (15.6%) have been diagnosed with AS in the first assessment.

The suspicion of SA was most frequently pointed out among psychiatrists (18.2%) who were the professionals that most reached the diagnosis (61.3%). The specialists who suspected and those who made the diagnosis of AS are described in Table 1. The age at which the diagnosis of SA was made ranged from two to 26 years old, with an average of 11 years old (SD = 6.67). The mean time between noticing something different in development and diagnosis of the SA was 7.8 years. Figure 1 shows Graph with the comparison between the ages at which the signs were perceived; the first clinical evaluation; the initial diagnostic suspicion and the SA diagnosis.

Table 1. Individuals who suspected and concluded the ASD diagnosis

Who suspected ASD	n	%	Professionals who concluded the diagnosis of ASD	n	%
Child psychiatrist	9	20.4	Psychiatrist	27	61.3
Psychiatrist	8	18.2	Pediatric psychiatrist	7	16
Psychologist	5	11.3	Psychologist	3	6.8
Speech therapist	5	11.3	---	-	-
Parents	4	9.1	---	-	-
School staff	4	9.1	---	-	-
Neurologist	4	9.1	Neurologist	3	6.8
Pediatrician	3	6.9	---	-	-
Pediatric neurologist	1	2.3	Child neurologist	1	2.3
Geneticist	1	2.3	Geneticist	1	2.3
-----	-	-	Multidisciplinary Team	2	4.5

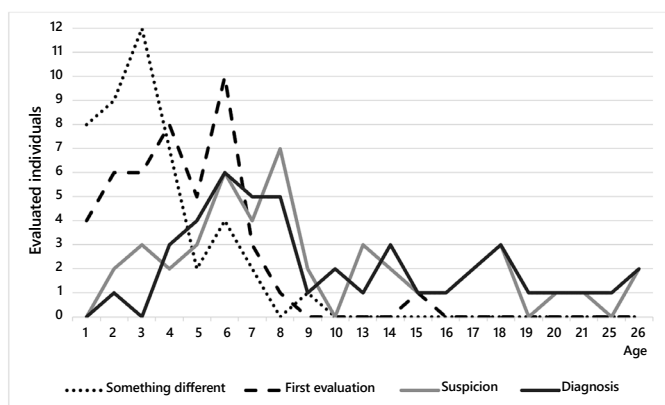


Figure 1. Comparison between the ages at which "something different" in the child's development was observed, the "first clinical evaluation", "suspicion" and "ASD diagnosis".

Main recommendations received at diagnosis are described in Table 2. The three most common were that the child should be sent to or continue attending regular school (68.2%), followed by a referral to a psychologist (59%), and prescription of medications (56.8%).

Table 2. Main recommendations received at ASD diagnosis

Guidelines	Nº	%
Mainstream school	30	68.2
Psychologist	26	59.0
Medication	25	56.8
Speech therapist	17	38.6
Genetic evaluation	16	36.3
Psychiatrist	4	9.1
Neurologist	3	6.8
Complementary exams	3	6.8
Multidisciplinary team	3	6.8
Professionalization	3	6.8
Audiometry	1	2.3
Specialized school	1	2.3
None	1	2.3

DISCUSSION

In the United States, the diagnosis is made at 74 months old on average¹³. Before the age of three, the child with ASD already has at least some characteristics that signal delay in social development or other disorders that could concern the parents²⁴. But, although ASD can be identified between 16 and 24 months old; a significant proportion of children are not diagnosed or treated until school age¹¹. Data of the present study have showed that the perception that there was something different about the child's development occurred around the age of three in most families. In addition, the parents were the first to notice, followed by the teachers. Similar results related to parents have been reported²⁵. Teachers observed signs of ASD in only 22.2% of cases and this reveals that they have an inadequate understanding of autism due to various misconceptions, as previously reported in other developing countries²⁶. Parents and teachers are expected to notice developmental delays earlier than other people as they spend more time next to the child.

It is important to highlight that in most cases in which parents were the first to notice something different with their children's development; they had older or younger siblings. The presence of older siblings without ASD was the most common finding in this study. This suggests that parents, when having a second child with atypical development, are able to notice differences when comparing behaviors between siblings; this facilitates the search for the diagnosis.

The medical professional responsible for the child's health is the pediatrician, who is also able to assess neuro typical development and recognize when early signs of Asperger's Syndrome may be present²⁷. The perception of health professionals if the child had something different, including pediatricians, was also unsatisfactory. This requires training to recognize and help children with autism and may be related to difficulties in diagnosis; the lack of knowledge; reluctance of professionals to label; or the lack of use of ASD screening tools by pediatricians²⁸.

In addition to the pediatrician, all primary health care professionals must be trained to suspect atypical behavior and refer the child for further investigation, in addition, to providing the necessary guidance to the family and the school as soon as possible.

In this study, during the evaluation process until diagnosis, the most sought after medical professional was the neurologist, while the pediatrician were in the fifth place in the search for diagnosis by parents. Data from South Africa have showed that the most sought after professional was the pediatrician followed by the neuropediatrician²⁹. The search for neurologists by parents may be associated with the lack of preparation and training of pediatricians in Brazil in recognizing the early signs of ASD-AS. Since the most frequent complaint of the individuals evaluated was the speech delay. This could have been understood as an isolated signal and not as part of a set of clinical signs; leading to a greater demand by the neurologist, due to the suspicion of some neurological problem.

In the first assessment with a mean age of 4.4 years old; 29.5% did not receive a diagnostic hypothesis; 70.4% received a comorbid diagnosis of AS. Among them the most frequent was ADHD, 34.1% of the cases, and only half of these received a correct diagnosis of AS. Several symptoms overlap with ADHD and Asperger's syndrome. Studies have long demonstrated that these phenomena are co-occurring and based on these findings; DSM-5 has integrated this diagnostic possibility. Both disorders can lead to a decrease in the level of attention, for example, high distraction for those with ADHD and lack of mental flexibility for individuals with ASD-AS. Likewise,

for relational difficulties, explanations vary according to the main diagnosis. Individuals with ADHD are impulsive, impatient, have trouble waiting for their turn or listening to others and, as a result, adjusting their behavior becomes difficult. Individuals with ASD-AS have difficulty processing emotional information, that is, they may not respond as expected, which can lead to complications in social interactions. Patients with any of these disorders usually demonstrate a special ability to engage their interests deeply: it is typical for individuals with ASD to develop "special interests", while ADHD patients tend to "hyper-focus" on the subjects that interest them. In the end, the two types of situations ("overlapping symptoms" or "association of the two disorders") mean that, when diagnosing ADHD with serious difficulties; ASD-AS should be sought³⁰. In addition, other diagnoses are given, due to an overlap of symptoms with other disorders, or comorbidities such as sleep disorders, intestinal problems, self-mutilation, eating disorders, obsessive symptoms, oppositional defiant disorder and seizures, which directly interfere with diagnosis and prognosis⁹.

Between the suspicion and the diagnosis of ASD; the psychiatrist was the medical professional who performed most of the diagnoses; 61.3% as compared to pediatricians; the children's average age was 11 years old. Similar data were described in a review in which Asperger's Syndrome is often diagnosed late. The average age of 11 years was also found, or even in adulthood in some cases³⁰.

After the diagnosis of AS, the most common recommendation for the families of patients in this study was to send or continue to send their child to a regular school. followed by the need of an evaluation from a psychologist, medication use, speech therapy and a genetic evaluation. These are important recommendations, but the fact that when receiving the diagnosis, the family may be confused, shocked and emotionally disturbed, with no ability to retain much information, should also be considered. It is extremely important that information on possible etiologies, risk of recurrence, associations and therapeutic strategies is offered, among other resources, according to the needs and capacity of retaining information in each case. With the lack of adequate information, parents can seek answers to their questions by consulting the literature or non-scientific websites on the Internet, which may contain inaccurate information, consequently increasing anxiety and even creating false beliefs; this must be clearly addressed by the professional.

Delay in diagnosis is extremely harmful. Some studies point out the beginning of therapeutic interventions and family support as the only factor that directly affects the improvement of these patients^{32,33}. Although the diagnosis of AS in children at an early age is more possible when performed by specialized and experienced professionals, even when ASD-AS is not diagnosed; multidisciplinary work will help to adjust developmental delay. An early and accurate diagnosis also allows families to prepare themselves to face the daily challenges in parenting their children, learn to deal with the demands of care, seek genetic counseling before having other children and obtain appropriate services in the early stages of development³¹.

In addition to the limitations of this study, since the sample size was small, this may have caused a selection bias and inaccuracy in the results, however, the findings were similar to other countries similar to Brazil.

CONCLUSION

The context of the ASD diagnosis, specifically when the behavioral phenotype has the peculiarities of AS, is complex. There is a tendency to fail to recognize ASD in diagnostic practices. A series of difficulties,

from the lack of preparation observe the signs to the therapeutic indications that result in delayed diagnosis and treatment, are prejudicial to the patients and their families. Continued efforts are needed to promote clinical and training programs to provide benefits in the lives of people with ASD.

ACKNOWLEDGEMENT

We are grateful to the families participating in this study.

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