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ORIGINAL ARTICLE

The challenges of autism spectrum disorder: from suspicion to diagnosis

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Abstract

Autistic spectrum disorder (ASD) is a neurodevelopmental disorder whose diagnosis requires trained and informed professionals to perform it properly. Knowing that the prognosis of children with the disorder is directly related to early diagnosis, this study aimed to verify the inclusion of the active search for signs of risk for autism in routine pediatric consultations. The target audience of the study was formed by parents or guardians of children already diagnosed with ASD, and the analysis took place through a questionnaire applied to the parents, as well as interviews with them. The data in this study showed that in 62.5% of cases, parents were the first to identify early signs of ASD, and in only 3.5%, pediatricians identified them. The average age of the children at the time the warning signs were perceived was 24.5 months, with two years of age in 53.5% of cases. Was also found that 42.8% of parents had their complaints not valued by pediatricians, and that in 78.5% of children, delayed speech was the early sign that first caught the attention of parents. Other early signs such as not answering by name were present in 51.8% of children, and also, 51.8% had repetitive movements. It was concluded that there is still a lack of information about autism both at a professional and social level, since most families had never heard of autism and that prejudice is still a difficulty related in the disorder.

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INTRODUCTION

The expansion of the Autism Spectrum Disorder (ASD) concept after the publication of the fifth edition of the Diagnostic and Statistical Manual of Mental Disorders[1] (DSM-V), led to an increase in the number of autism cases. ASD encompasses disorders formerly called early childhood autism, childhood autism, Kanner's autism, self-functioning autism, atypical autism, pervasive developmental disorder not otherwise specified, childhood disintegrative disorder, and Asperger's disorder[2].

Its diagnosis is essentially clinical and considers the criteria established by the DSM-V, which are: persistent deficits in reciprocal social communication and social interaction, restricted and repetitive patterns of behavior, interests or activities. Symptoms must be present early in the developmental period, and cause clinically significant impairment in social, professional, or other important areas of the individual's present life.

Among the early signs that can be noticed in children with Autistic Disorder, the main reason for seeking a medical evaluation is speech delay, but there are other signs that can be present even before the perception of the speech disorder, such as poverty in social interaction, which can be perceived even during the breastfeeding phase, when there is no shared gaze with the mother during the breastfeeding period[3]. Other signs that can be seen early in children with autism are stereotyped or repetitive behavior, including excessive use of objects, sleep disturbances, lack of make-believe play, as well as little or no ability to initiate social interactions and sharing emotions[4].

Although the average age at diagnosis is currently approximately 3 years[5], recent studies show that the early diagnosis of ASD can be made even in the first months of a child's life, as they can manifest signs even before 18 months of age. Delay in global developmental acquisitions and absence of expected behaviors for the chronological age are perceptions that can be made by both parents and pediatricians during children's developmental assessments; however, early identification comes up against the lack of preparation and insecurity of the pediatricians for the identification of these signs, delaying the diagnosis, generally, for periods between 3 and 5 years of age.

Today we know that the origin of ASD is at the intersection between environmental factors and multiple mutations in different genes, with variability in the set of mutations among carriers of the disorder, which makes genetic screening for the disorder difficult[6]. Through the elucidation of the genetic basis of ASD and the observation that younger siblings of children with ASD have a recurrence rate for the disorder of approximately 18.7%[7], the importance of early screening in children who have siblings with the disorder was established, in addition to those who have other risk factors, due to environmental influence, such as low birth weight, premature birth, high parental age at conception, maternal hypothyroidism during pregnancy and other factors that may be associated with the development of ASD[8].

Due to the daily contact with their children, it is the parents who usually notice the first signs; however, the meaning of these signs is not always clear and they end up being considered normal. It is then up to the pediatrician, a person constantly present in the daily lives of children, the curiosity to actively seek or question the child's development and behavior in order to identify the pathological milestones of development.

In times where the media have fast and free information, parents are more aware about ASD and, therefore, more likely to develop concerns about the possibility of autism. In this context, it is important that pediatricians accept the questions raised by the family and actively search for signs of ASD. To facilitate early autism screening, screening questionnaires have been developed, which offer a quick and inexpensive method of early identification of children with autism[9]. One of the most used is the Modified Checklist for Autism in Toddlers (M-CHAT). This scale presents an excellent instrument to be included in the pediatric routine, being recommended by the Brazilian Society of Pediatrics (SBP)[10], as it can be applied to all children during pediatric visits, including those who do not have symptoms, but have a risk factor for the disorder[11].

In the knowledge that the early diagnosis of ASD is the main tool for an effective prognosis, and that for this it is necessary that professionals in contact with children have the information and the ability to perform it properly, the present study aims to assess the inclusion of the active search for signs of risk for autism in the routine consultations of pediatricians, analyzing the perception of parents of children diagnosed with ASD, regarding the performance of pediatricians in the lookout and diagnosis of autism.

MATERIALS AND METHODS

This is a quantitative and cross-sectional study carried out in the city of Mossoró-RN and region. The study group consisted of fathers and mothers of children diagnosed with Autism Spectrum Disorder and residents of Mossoró-RN and region. The approach of these participants took place in associations for parents of children with autism in these regions, and also in the Ação Pró-TEA outreach project from the University of the State of Rio Grande do Norte. The study instrument was an objective questionnaire, made up of ten questions encompassing issues related to the diagnosis of ASD and its challenges from their perspective. The parents were approached individually, and then presented with the Informed Consent Form (ICF), where they were able to learn about the study objectives and its ethical issues. Those who agreed to participate answered the questionnaire individually, allowing data privacy and confidentiality. A questionnaire was designed for each child diagnosed with ASD, since some families have more than one child with the disorder, and the diversity in the diagnostic process

of each one. Along with the questionnaire, a previously structured interview was carried out, in which they were asked how the child's ASD suspicion process was, if the pediatrician who regularly followed the child noticed the early signs of ASD, if he/she was sought and considered the suspicion; and what was his/her behavior in the face of the complaint.

The children represented in this study had their diagnoses performed following the DSM-V criteria, and by various professionals, such as pediatric neurologists, neurologists, psychiatrists and neuropsychologists.

Data related to the questionnaire were tabulated in the Excel version 2016 software, and then we employed an analysis of variance (ANOVA), while the analysis of data referring to the interview was performed using the "Prose analysis" method[12]. The notes produced during the interviews were then read, seeking the meaning of the qualitative data and generating, at the same time, topics contextualized with the study.

Respecting ethical standards in research, the present study was submitted to the Research Ethics Committee of the University of the State of Rio Grande do Norte, being approved as stated in registration 02454818.8.0000.522 and opinion number 3,101,712.

RESULTS

We had 56 parents or guardians of children diagnosed with Autism Spectrum Disorder residents of Mossoró-RN and region participating in the study. Of these, 62.5% reported that they were the first to notice the first atypical signs in the child, while in 8.9% of the cases it was the grandparents, 5.4% the neighbors, 16% the teachers and only 3.5% stated that the pediatrician was the first to notice the early signs (Table 1). Still, 17.8% of parents pointed out that other people such as friends, aunts, psychologists, speech therapists and on-call clinicians in the emergency department were the first to notice the early signs in the children.

As for the age of the children when the first signs of ASD were noticed, the average was 24.5 months, with a minimum of 6 months and a maximum of 60 months. 53.5% of parents stated that the age of two was the starting point, while 16% indicated that the first signs were noticed at three years of age, and 8.9% when the child was less than one year old (Minimum of six months and maximum of 10 months). 8.9% of parents stated that the child was over three years old when they noticed the first signs of ASD.

When asked about the pediatrician's role in the children's diagnostic process, 42.8% of the parents stated that the pediatricians did not value their complaints, 23.2% indicated that the professional accepted the complaint and referred the child to a neuropediatrician, while 12,5% accepted the complaint and did not diagnose autism in the child. 21.4% of the parents stated that they sought out the pediatric neurologist directly, either on the recommendation of the school or other professionals, such as psychologists and speech therapists.

Table 1. Authorship of the perception of the first signs of Autism Spectrum Disorder and the most prevalent early signs reported by parents of children diagnosed with autism in Mossoró-RN and region.

ITEMS	(%)
Who perceived the first signs?	
Parents	62,5%
Grandparents	8,9%
Teacher	16%
Neighbor	5,4%
Pediatrician	3,5%
Other	17,8%
Which signs made you suspicious and go for a medical evaluation?	
Speech delay	78,5%
Child did not respond to name	51,8%
Repetitive behavior	51,8%
No eye contact	42,8%
Sensitivity to light or sound	34%
Social isolation/Poor interaction	50%
Other	17,8%

Source: Developed by the author (2019).

"*Does this boy have autism?*", "*You are looking for a problem with your child*" "*That is normal, mother, each child has their own time*", "*Your child will speak, he has not yet spoken because he is lazy*". These are some of the phrases that the participants of this study revealed to have heard from pediatricians when they presented them with complaints regarding ASD in their children.

A mother reported a negative experience when she went to the pediatrician because she suspected that her youngest child was on the spectrum, as he had some similar behaviors to her oldest son already diagnosed with ASD: "*Do you really want to treat them as autistic? Do you really want to tell everyone they are autistic? Aren't you afraid of what they're going to suffer, the prejudices they might suffer?*" The prejudiced tone in the doctor's speech was perceived and described by her, accentuating her anxiety in the face of doubt about the diagnosis and the future of her children.

With regards to the early signs manifested by the child, in 78.5% of the cases, speech delay was the first sign that drew attention. Among the other perceived signs, 51.8% of the parents stated that the children did not answer by name, 51.8% had repetitive behavior, 42.8% did not make eye contact, 34% were sensitive to light or sound and 50% had social isolation (interaction poverty). Other signs, such as echolalia, hyperactivity, excessive crying, walking on tiptoe, graphics and fixation on letters and numbers were present in 17.8% of the children (Table 1).

Of the children diagnosed with ASD who were represented in this study, 78.5% of the diagnoses were made by a pediatric neurologist, 14.3% by a psychiatrist and 5.3% by other professionals such as a neurologist and neuropsychologist.

The time elapsed between the presentation of the suspicion to a professional and the diagnosis was less than one year in 39.2% of the cases, one year in 19.6% of the cases, two years in 19.6%, three years in 8 .9%, and 12.5% took more than three years to complete the diagnosis (Table 2).

When asked if the child received any other diagnosis before ASD, 66% of parents said no, while 10.7% reported that the child was diagnosed with developmental delay, 7.1% was diagnosed with ADHD, 10.7% with hyperactivity, 3.5% with primary language delay and another 3.5% received other diagnoses such as "tics" and Dandy Walker Syndrome (Table 2). In this study, we found that the children went through an average of two professionals for the conclusion of the ASD diagnosis, with 23.2% of the children going through only one professional, 46.4% by two professionals, 16% by three professionals and only 12.5% went through more than three professionals to then receive the diagnosis of ASD.

Table 2. Determining topics for the chronology of the autism diagnosis: time elapsed between suspicion and diagnosis, number of professionals the family went through before completing the diagnosis and other diagnoses received by the child.

Items	(%)
How long did it take from the suspicion to the final diagnosis?	
Less than one year	39.2%
01 year	19.6%
02 years	19.6%
03 years	8.9%
More than 03 years	12.5%
How many healthcare professionals saw the child before the diagnosis?	
Only 1	23.2%
Two	46.4%
Three	16%
Four	12.5%
Did the child receive another diagnosis?	
No	66%
Development delay	10.7%
ADHD	7.1%
Hyperactivity	10.7%
Primary language delay	3.5%
Other	3.5%

Source: Developed by the author (2019).

DISCUSSION

The data obtained in the present study showed that parents were the main ones responsible for the perception of the first atypical signs of ASD in their children (62.5%). In a study carried out in Mexico in 2013, Sampedro-Tobón, González-Gonzáles, Véles-Vieira and Lemos-Hoyos[13] found results similar to those presented in this study, with 59.5% of the cases being the first signs perceived by the parents, 21,4 % by teachers and only 2.4% by pediatricians, confirming that parents are still the main source in the process of identifying signs of risk for ASD.

Teachers appear in second place as identifiers of early signs in both studies, suggesting that they are looking for information and training to work on inclusion in their classrooms, as well as to identify warning signs, thus constituting important allies in the autism diagnosis process.

Zalaquett, Schönstedt, Angeli, Herrera, and Moyano[14] point out that early intervention can be used to modify the course of autism at the brain level, by directing development towards a more adaptive trajectory, optimizing the prognosis, so it is important that the diagnosis be made as early as possible, making it necessary for both parents and professionals who are in contact with the child, such as pediatricians, for example, to be alert to the signs that children may be showing, in addition to having the training to adequately identify these developmental delays.

That said, it is important that pediatricians know how to identify the signs of ASD, diagnose and also know how to recommend the appropriate treatment, so that little time is spent until its beginning, favoring the evolution of patients. Flores and Smeha[15] in 2009 verified in their study that pediatricians, although they were aware of the most classic early signs of ASD, were unaware of signs such as hypotonia, refusal to look, sleep disturbance and food refusal. Ferreira and Smeha[16], in 2017, pointed to an evolution in the knowledge of pediatricians, although still precarious, showing that this group still lacks information about the disorder, putting at risk, through misinformation, the good prognosis.

Analyzing the data from this study, we can say that in Mossoró pediatricians still lack training on autism. It is worrying that 42.8% of the parents have not had their concerns addressed and investigated by these professionals, and that 12.5% of the families who had their complaints dealt with have not received an adequate diagnosis, generating anguish and clearly delaying the children's diagnosis.

According to the parents' perception reported during the interview, pediatricians are resistant to diagnosing mainly milder cases of autism, given the social stigma that autistic children are those stereotypical, agitated, aggressive children who are unable to develop bonds. For many, especially those with longer training in pediatrics, the diagnosis of autism is used only in the most severe cases, the "typical" cases, and this conduct is the result of the lack of information and professional training.

Prejudice was a topic frequently mentioned by the participants in this study, and it is possible to infer that the origin of this problem lies in the lack of information about

the disorder. The association of autism with madness and disability is common, causing discomfort to the closest people when the diagnosis is suggested[17]. The most striking, in fact, was the finding that prejudice is present even in the doctor's office, being the most frequent complaint of parents during the interview of this study.

Language delay from the second year of life onwards was the sign most cited by parents when asked about the signs perceived prior to the diagnosis, but we know that in fact other signs are already present earlier and are considered only when questioned retrogradely. by professionals, especially the commitment to social development, which Zanon, Backes and Bosa18 point out as one of the first signs to manifest, but which is little noticed by parents. In children under one year of age, this impairment is mainly manifested by the non-correspondence to the mother's gaze during breastfeeding, rejection of the mother's breast and changes in sleep quality. It is important to point out that these isolated signs do not constitute a diagnosis of ASD but should raise an alert for the search for a professional evaluation and vigilance of this child for the presence of other signs.

Most parents reported that they perceived delays and "different" behaviors in their children, but they did not attribute a meaning to such atypia because they did not know about autism, some said they had never heard of the disorder. Zanon, Backer and Bosa18 also showed in their study that the parents' belief in the nature of the first signs manifested by the child alters the chronology of the diagnosis, since those parents who attributed them to tantrums, disobedience and difficult personality, that is, reasons disorders not specific to the disorder, received the diagnosis in a delayed manner.

We also found that the children went through an average of two professionals for the conclusion of the ASD diagnosis, which is different from the results found by Sampedro-Tobón, González-Gonzáles, Véles-Vieira and Lemos-Hoyos[13], who revealed an average of five professionals sought before the diagnosis (minimum of two and maximum of ten), and that up to 33% of the children received some diagnosis before the ASD, being possible to conclude that there has been progress with regards to the flexibility of the diagnostic process, this being performed in a more direct and precise way and in less than a year in 39.2% of cases, generally by pediatric neurologists and psychiatrists, as this study has already pointed out.

In general, the data from this study regarding the diagnosis corroborate the perception frequently reported by parents that pediatricians are absent from the responsibility of investigation and diagnosis, immediately referring the child to a neuropediatrician or psychiatrist, without even performing an initial evaluation in your office or offer information about the disorder.

Jendreieck[19] characterizes in her study what are the difficulties pointed out by pediatricians in the early diagnosis of ASD, namely: the difficulty of getting to know the child and the family better, given the short time of consultations, in addition to the different environment; the family's lack of knowledge about autism; need to accompany the child for longer; the existence of different degrees of behavior within the spectrum; the need to inform the family about the diagnosis. In order to overcome these difficulties, pediatricians can suggest to parents the behavioral observation of the child, one of the resources being that suggested by Garcia and Lampréia[20] and Mansur, Colares and Mansur[3], who suggest the use of retrospective family videos in the search for early signs, mainly in the first year of life, as they claim to help in the identification, mainly, of the more subtle signs, such as the absence of a shared look, smiles and irritability.

This study showed that the municipality of Mossoró-RN and region still lacks pediatricians prepared to identify the early signs of autism and diagnose it properly, characterizing that the signs of risk for ASD still go unnoticed by professionals, delaying diagnosis and culminating in late interventions. In addition, it showed that when compared to previous studies, there was progress related to the time elapsed between suspicion and diagnosis, as well as the number of professionals sought by the family before having a diagnosis.

Active listening to complaints reported by parents is vehemently important, as is effective communication, explaining the disorder to them, applying rapid tests, when appropriate, and also referring them to other professionals when deemed appropriate. It is necessary that pediatric professionals seek information and training so that the identification of signs of risk for autism, diagnosis and clarification to the family are carried out properly and early.

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