International Journal of Health Science

AUTISM SPECTRUM DISORDER - A REVIEW ON EARLY DIAGNOSIS IN PRIMARY CARE AND WARNING SIGNS FOR THE FAMILY

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Abstract: Autism Spectrum Disorder (ASD) is a neurodevelopmental disorder with cognitive-behavioural manifestations characterized by atypical development, deficits in communication and social interaction, repetitive and stereotyped patterns of behaviour and a restricted repertoire of interests and activities. Many children on the Autistic Spectrum show warning signs in the first few months of life and the diagnosis is established at around 2 to 3 years of age. Its etiology is still unknown, but scientific evidence indicates that there is no single cause, but rather the interaction of genetic and environmental factors. Environmental factors can increase or decrease the risk of ASD in genetically predisposed people. As this is a genetic disorder that manifests itself in childhood, early diagnosis is essential in order to guide specific treatment interventions and thus help minimize social problems and barriers, enabling a better quality of life for the child and their family. In this sense, primary care and parents play an important role in the prognosis because the first manifestations are usually noticed in the family with clinical signs that vary, such as impaired eye contact, lack of imitation skills, preference for objects and repetitive activities, as well as difficulty or delay in speech. These signs are evident during consultations and are usually investigated by the primary care doctor who, when identifying the suspicion, must refer the patient to specialized rehabilitation care, which will confirm the diagnosis and outline the appropriate therapy. It is therefore up to the professionals involved to have a sensitive eye for observing development and noticing clinical characteristics associated with ASD in the child who arrives for diagnostic confirmation. The aim of this paper is to help with the early diagnosis of ASD, by reviewing the literature and highlighting the main forms of clinical diagnosis, as well as the warning signs that can be identified by parents through family
relationships in the first few months of life.

METHODOLOGY

An integrative literature review with a qualitative approach was carried out, with searches in the Virtual Health Library (Ministry of Health), Scientific Electronic Library Online, Google Scholar and Scielo Brazil. The study includes information collected from 12 scientific publications that deal with autism spectrum disorder and its diagnosis. The main descriptors used were: autism, early diagnosis and family diagnosis. To select the articles, the inclusion criteria were articles published between 2013 and 2023, except for the basic references.

LITERATURE REVIEW

Autism Spectrum Disorder is a neuropsychiatric syndrome that was first described by Kanner, in 1943, as Autistic Disorder of Affective Contact, when he observed 11 children who differed in their behavioral patterns, referring to characteristics that showed a possible disorder. During this period, the dominant idea in international medical-psychological literature emphasized affective and relational factors, justifying that autism resulted from disturbances in early interactions between parents and babies. The cognitivist bias emerged only 40 years after the initial description of the disease, at which time the condition later known as "Asperger's Syndrome" began to spread. Articles report that the conceptualization of autism between 1943 and 1983 went through a transition from the "affective-relational" view to the "cognitive-cerebral" conception of autism in the psychiatric field.

Currently, research into autism continues to gain prominence in medicine and has undergone important changes, culminating in new and earlier diagnoses, as well as the breaking down of prejudiced stigmas. Today the disease is defined, both in the 10th revision of the International Classification of Mental Disorders

(ICD-10) and in the Diagnostic and Statistical Manual of the American Psychiatric Association (DSM-V) as a complex developmental disorder, presenting impairments in social interaction, communication as well as patterns of repetitive and stereotyped interests and behaviors. Its definition is well elucidated in current literature, but diagnosis is still a challenge. According to the American Academy of Pediatrics, there is no pathognomonic sign or laboratory test that differentiates ASD, so in order to establish the diagnosis, doctors must first get to know the clinical symptoms well, evaluate the child's clinical characteristics and listen carefully to the family.

However, studies using genetic markers are increasingly advanced in the search for a diagnosis. A study carried out by Herault et al. with the aim of confirming an association between autism and markers of the C, Harvey-ras (HRAS) oncogene, studied the genotype of a group of clinically and genetically well-characterized autistic children (n = 55), compared to a control group of normal children (n = 55). They found significant differences in the allele frequencies between the two populations of two HRAS gene markers located on the short arm of chromosome 11, suggesting that this region of DNA on chromosome 11 confers susceptibility to childhood autism. This finding suggests that genetic markers could be used in the clinical assessment of the disease in the near future.

In Brazil, it is estimated that one in every 160 children has ASD, so 2 million people are considered to be on the autistic spectrum, with an incidence four times higher in boys than in girls. In general, the number of people diagnosed worldwide with the disorder is increasing, which does not necessarily indicate an increase in its prevalence. This can be explained by the expansion of diagnostic criteria, the increase in health services related to the disorder and the change in the age of diagnosis, among other factors. To help with the diagnosis, as well as offering appropriate therapy for those with the spectrum, the National Policy for the Protection of the Rights of People with Autism Spectrum Disorder was implemented in 2012 in order to guarantee comprehensive care and stimulate discussion and scientific production. This is important because most of the care given to people with ASD in Brazil is provided by the Unified Health System (SUS).

The care of patients with ASD takes place mainly at the Primary Care and Specialized Care levels. Until now, there have been no biological markers or tests to confirm the full spectrum of ASD. It is therefore up to the professionals involved to have a sensitive eye for observing development and noticing clinical characteristics associated with ASD in the person who arrives for diagnostic confirmation. In Primary Care, it is possible to identify symptoms and signs suggestive of Autism Spectrum Disorder (ASD) by monitoring the child's development in their first years of life. The initial alterations of ASD are often present before 18 months of age. There is no pathognomonic symptom of ASD, so you should avoid jumping to conclusions, such as excluding the possibility of the condition because the patient doesn't show any changes in speech or because they have already made eye contact at some point, for example. Early signs that indicate the possibility of the condition include: concerns reported by parents or caregivers about deficits in social skills, language deficits or difficulties tolerating change; delay in the acquisition of social and communication skills, such as not pointing to an object of interest at around 14 months, not playing 'make-believe' at 18 months (e.g. avoiding eye contact or wanting to be left alone; difficulty understanding other people's emotions or talking about their own emotions; frequently repeating phrases or words

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(echolalia); answering questions with unrelated or "random" answers; getting irritated at the slightest changes; showing restricted and obsessive interests; showing movements such as flapping their hands, spinning in circles or rocking their body; showing unusual reactions to noise, smell, taste or touch. Patients who show any of the above signs can be assessed using the M-CHAT scale (The Modified Checklist for Autism in Toddlers) in primary care. The M-CHAT scale helps to identify patients aged between 16 and 30 months with possible ASD. It is a quick questionnaire to be answered by parents or caregivers during the consultation. Recently, the Ministry of Health included the M-CHAT-R/F.A Checklist and the M-CHART-R scale in the child's booklet in order to make it easier for health professionals to detect the initial signs.

The M-CHAT-R/F scale (FIGURE 1) is an instrument used to help identify patients with a possible Autism Spectrum Disorder . The instrument can be used by any health professional and is quick to apply. It can also be answered by the child's caregivers and parents during the consultation. According to Law No. 13.438, of April 26, 2017, the M-CHAT-R assessment is mandatory for children in follow-up pediatric consultations carried out by the Unified Health System (SUS). It consists of a test with yes/no questions to be answered by parents considering the behavior they observe in their children. At the end, the points are added up and the result indicates the presence of autism symptoms, but does not confirm the diagnosis. When the score is very high, the pediatrician refers the child to a specialist or a multidisciplinary team for evaluation.

- **Score of 0 to 2 - Low risk. With** this score, the chances of confirming a diagnosis of ASD are low, so it is not necessary to investigate further, although it is recommended to repeat the test if the child is under 2 years old.

- **Score from 3 to 7 - Moderate risk.** With this score, the pediatrician moves on to the second stage of the M-chat, the follow-up interview. This interview gathers more information about the signs of ASD. If the score is greater than or equal to 2 at this stage, it may indicate the presence of autism and the child should be referred to a specialist. If the score is between 0 and 1, it does not indicate autism, although it is recommended to repeat the test later.

- **Score of 8 to 20 - High risk.** With this score, parents should go directly to a specialist to confirm the diagnosis, without the need for a follow-up interview.

In addition to the M-CHAT-R/F scale, the literature cites other scales for autism screening, the Childhood Autism Rating Scale (CARS), a scale made up of 15 items ranging from 15 to 60 points, in which autism is classified by the score of 30 points. Autistic Traits Scale - ATA, its score ranges from 0 to 15, where 0 represents no symptom, and 2 if there is more than one symptom in each of the 36 items. Autism Treatment Evaluation - ATEC, a method used to evaluate the effectiveness of autism treatments, containing 77 questions, but which is not recognized by the Federal Council of Psychology, as it is not standardized in Brazil. Autistic Behavior Checklist - ABC or ICA, a 57-item questionnaire based on the behavioral analysis of individuals, which is a method capable of identifying individuals with a high autistic profile.

Regardless of the scale used, the diagnosis of ASD requires a great deal of observation, so that it can be treated correctly and early on. Due to daily life, involving different contexts and occasions, it is recognized that, most of the time, it is the parents, and not the professionals, who are the first to suspect problems in the child's development. Coonrod and Stone (2004) point out that caregivers' initial

M-CHAT-R™

Please answer these questions about your child Remember how your child usually behaves. If the behavior has been observed a few times, but they don't usually do it, then please answer no. Please answer **yes or no to** each question. Thank you very much.

1. If you point at something on the other side of the room, does the child look at what you're pointing at? (For example, if you point to a toy or an animal, does the child look at the toy or animal?)	Yes	No
2. Have you ever wondered if your child might be deaf?	Yes	No
3. Does your child play make-believe? (For example, pretending to drink from an empty glass, talking on the phone or pretending to feed a doll or teddy?)	Yes	No
4. Does your child like to climb on things? (For example: furniture, slides in the playground or stairs?	Yes	No
5. Does your child make unusual movements with their fingers near their eyes? (For example, wiggle your fingers near your eyes)	Yes	No
6. Does your child point with a finger to ask for something or to get help? (For example, pointing to a food or toy that is out of reach)	Yes	No
7. Does your child point with a finger to show you something interesting? (For example, pointing at an airplane in the sky or a big truck on the road)	Yes	No
8. Is your child interested in other children? (For example, does your child watch other children, smile at them or approach them?	Yes	No
9. Does your child show you things by bringing them to you or lifting them up for you to see - not to get help, but just to share? (For example, show a flower, a soft toy or a toy truck)	Yes	No
10. Does your child respond when you call them by name? (For example, she looks, speaks or babbles, or stops what you're doing when you call her name)	Yes	No
11. When you smile at your child, do they smile back at you?	Yes	No
12. Is your child bothered by everyday noises? (For example, does your child scream or cry at noises like vacuum cleaners or loud music?)	Yes	No
13. Is your child walking?	Yes	No
14. Does your child look you in the eye when you talk to them, play with them or dress them up?	Yes	No
15. Does your child try to imitate what you do? (For example, does it wave goodbye, clap its hands or make funny sounds when you make them?)	Yes	No
16. If you turn your head to look at something, does your child look around to see what you're looking at?	Yes	No
17. Does your child try to get you to look at them? (For example, does your child look at you for a compliment or say "look" or "look at me(")?)	Yes	No
18. Does your child understand when you tell them to do something?	Yes	No
19. When something new happens, does your child look at your face to see your reaction?	Yes	No
20. Does your child enjoy activities with movement? (For example: being rocked or bounced on your knee?)	Yes	No

M-CHAT-Revised (Robins, Fein, & Barton, 2009)

Translated and Adapted by Carla Cintrão Almeida

	Social Interaction		Language		Jokes	
From 0 to 6 months	Children with ASD do not look for their caregiver	Pay more attention to objects than people	Ignore or do not recognize the speech of their caregivers	Tend to silence or random screams	Long-lasting crying with no apparent connection to events or people	Do not explore objects and its forms (shake, hit and throw)
From 6 to 12 months	Children with ASD have difficulty reproducing/imitation behaviors	They do not respond by name, they only react after insistence or touch	Do not show meaningful facial expressions	They don't respond as if they were talking with screams and noises	They do not repeat manual or body gestures when requested (kiss) but may repeat the gesture randomly out of context	They need a lot Insistence from adults to engage in pranks.
From 12 to 18 months	Does not point out objects, does not show which objects arouse curiosity	Difficulty understanding new situations outside of everyday life	They present fewer facial variations when communicating. Express joy, anger, frustration, but not surprise or shame	May not present their first words in this age group	They don't play pretend games (starts at 15 months)	Explore less objects than others children and tend to fix on an action repetitive than explore the functions of objects
From 18 to 24 months	They do not follow the other person's gaze or point to an object. They can look at the finger, but they don't make the connection to something being shown	They are not interested in taking objects offered by familiar people	They gesture less than other children or use gestures randomly. They may also not know how to signal "yes" and "no" with their head	Language does not develop, they do not explore speech and tend to repeat what listen. Repeated speech and without autonomy	They do not imitate the actions of adults, they are not interested in playing house or role-playing	Don't play with that the object represents and may only be interested in one aspect such as rotating the wheels of a cart
From 24 to 36 months	Gestures and comments in response to adults tend to be isolated. Rare initiatives to point out, show or give objects	Speech tends to be the repetition of the other person's speech	Lack of interest in everyday narratives and dialogue with parents	They do not distinguish between gender, number and verbal tense of speech. Tend to repeat randomly, not in dialogue with the adult	Tend to stay away from other children or limit themselves to observing them from a distance	When they accept play with others children, have difficulty in understand them

Source: Adapted from Diretrizes de Atenção à Reabilitação da Pessoa com Transtorno do Espectro Autista. Brasília: Ministry of Health, 2013.

concerns are generally accurate and legitimate. Therefore, interviewing parents is an important source of information when it comes to diagnosis or research involving children with ASD (Lord, Storoschuk, Rutter & Pickles, 1993), although there are limitations to this procedure. Among the obstacles are the difficulty and lack of knowledge on the part of parents of the aspects of development that are expected for a given age. This last aspect points to the need for studies and the dissemination of results beyond the scientific community.

Specialized care includes the Psychosocial Care Centers (CAPS), which are open and community-based services that must provide daily care, offer efficient and personalized clinical care, promote the user's social inclusion, and support and supervise mental health care in the basic network. The Child and Adolescent Psychosocial Care Centers (CAPSI) were created to care for children and adolescents. This center provides well-dimensioned care because it is made up of multi-professional teams, operates in the territory and respects the need/demand of each case.

PARENTS IDENTIFYING THE FIRST SYMPTOMS OF AUTISM

Over the last two decades, studies have sought to identify the age at which the first symptoms of ASD are recognized. To this end, the Ministry of Health has developed a table with indicators of child development and warning signs of ASD (FIGURE 2). Parents should be aware of the signs because early diagnosis between two and six months of age is important, due to the period of greatest neural plasticity. During this period, the help of a multidisciplinary team, combined with appropriate guidance for parents, significantly improves the communication and social skills of these children. Signs can be observed by parents even before 6 months of age. Infants can show signs of ASD from the first months of life, presenting early signs that indicate special attention in the routine assessment of neuropsychomotor development. Signs that may suggest atypical development and should be assessed at the childcare appointment: delay in acquiring a social smile, lack of interest

or little interest in the human face, unsupported or absent gaze, preference for sleeping alone in the crib, irritability when swaddled, absence of separation anxiety and indifference when parents are absent.

After 18 months, ASD traits become more evident. Any verbal or non-verbal language delay, deficits in social contact and interest in others, prominent repetitive interests and stereotypes should be investigated. Regarding the nature of the first symptoms observed by parents, delayed communication and language development is the most frequently reported symptom. On the other hand, research shows that impairments in social development are the first symptoms to emerge, although they are only recognized by a small proportion of parents. In addition to delays in communication and social development, a study of 36 children with ASD and 20 controls with typical development and their parents showed that 20 to 30% of caregivers described patterns of language regression, involving the loss of previously acquired words (Werner & Dawson, 2005). In fact, recent research on the subject has shown that regression is a reliable phenomenon that affects a significant proportion of children with ASD

FINAL CONCLUSIONS

Autism spectrum disorder is a highly complex disease that must be approached in a multi-centered way, with the aim of improving the patient as a whole. Therefore, early diagnosis of ASD is a watershed between children who will achieve greater autonomy in the future and those who will always be dependent on someone. The earlier this identification is made, the more effective the intervention actions will be, since the later the perception of autism, the more consolidated the symptoms will be.

Among the most important causes of early diagnosis are the lack of professionals with knowledge of the subject, the lack of a gold standard diagnostic tool and insecurity on the part of family members. There are various methods that assess the patient and can suggest a diagnosis, but it is up to the professional to assimilate the patient's clinical characteristics, the family's reports and the results of the tests and questionnaires applied in order to make a more reliable diagnosis

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