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Autistic Spectrum Disorder - a New Perspective About the Early Diagnostic

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ABSTRACT

The term “autism spectrum disorder” (ASD) describes today a heterogeneous group of neurodevelopmental disorders with diverse etiologies. Autism spectrum disorder is obviously a neurodevelopmental disorder that seems to be a big challenge today for both: the family doctor and the pediatrician. The core of this disorder is mainly integrated by the patient’s communication and social interaction difficulties and by the presence of repetitive or restricted behaviors and / or interests. (AUGUSTYN, PATTERSON, TORCHIA, 2019 p. 1)¹⁻¹⁰ Autistic Spectrum Disorder is a pervasive and permanent disorder. It has no cure, no specific treatment, and this must be clarified from the beginning, however, early intervention can drastically alter prognosis and soften symptoms (SOCIEDADE BRASILEIRA DE PEDIATRIA, 2019)¹⁻¹⁰.

To benefit from early intervention, the patient with autistic spectrum disorder needs an early diagnosis. The key to their better social integration is the time.

It is obvious that children identified with risk for autism spectrum disorder should be referred to a specialist with the purpose to establishing the diagnosis. However, it is primarily up to the primary-care physician to identify children at risk through developmental follow-up, behavioral follow-up and eventually through a valid screening and clinical judgment. In fact, early, accurate and appropriate diagnosis usually requires a clinician with experience in diagnosis and treatment. However, the contribution of a multiprofessional team to assess key symptoms, functional impairment, severity, and comorbid conditions is very important. The management of this patient should be individualized according to the child’s age and specific needs. The primary care provider can refer the child to local consultants or the public school system for ancillary evaluations (speech language, cognitive and adaptive testing, psychoeducational testing) (AUGUSTYN, PATTERSON, TORCHIA, 2019 p. 2)¹⁻¹⁰ The key to our attitude as professionals is continuous follow-up. And it needs to be done together with an expert - or rather a team of professionals who can monitor both the process and the progress.

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OVERVIEW

The concept of "early intervention" was really the biggest success for the management of autistic patient. Despite the common goal of all strategies used - which is the earliest possible diagnosis and intervention - there is a consensus that the entire treatment of autistic spectrum disorder needs to be individualized, considering the weaknesses and the strengths of the respective patient and the needs and perceptions of the child and family. Precisely, we can say that managing Autistic Spectrum Disorder requires a multidisciplinary approach that utilizes these child's strengths to defeat the weaknesses.

But, in fact what is the benefit of early intervention? Early diagnosis and early treatment have the ability to positively influence the development of patients with autistic disorder, particularly their behavior, functional skills and communication. Even more, we can expect as a result the attenuation of symptoms over time or the minimization of symptoms so that they no longer cause disability or failure of social integration.

What's new about the concept of early intervention?

The Guideline of the Scientific Department of Pediatrics of the Development and Behavior of the Brazilian Society of Pediatrics, states that although TEA symptoms are consistent and identifiable between 12 and 24 months of age (for OZONOFF et al. 2016 sometimes we can find suggestive symptoms even from 6 months of age), unfortunately, the diagnosis of TEA occurs, on average, at 4 or 5 years of age. For the Brazilian Society of Pediatrics (2019)¹⁻¹⁰ this would be a "lamentable situation", because, according to some researchers, the early intervention even prevents the complete manifestation of the autism spectrum disorder if therapy start with 12-24 month of age, which is considered the best time of development when the brain is highly plastic and malleable.

In the first place, the new perspective on autism diagnosis means the search for early signs described below and remains an area of intense scientific research:

- abnormalities in motor control;
- delayed motor development;
- decreased sensitivity to social rewards;
- negative affect;
- difficulty in controlling attention;

For Augustyn (2019)¹⁻¹⁰, in most cases, parents can be the best source of information and each of their concerns must be investigated. The first issue that matters is the history of development, especially the age-appropriate early socio-emotional and linguistic habilities.

Early signs of autism may appear, sometimes, from the first months of life. However, changes in the media and language domains and repetitive behaviors between 12 and 24 months have been proposed as early identification markers for autism (Sociedade Brasileira de Pediatria, 2019)¹⁻¹⁰:

- lose acquired skills, such as babbling or deictic gesture of reaching
- absence of interest for sounds, noises and voices
- low eye contact and sustained eye deficiency;
- low attention to the human face (preference for objects);
- show greater interest in objects than in people;
- do not follow moving objects and close people;
- have few or no vocalization;
- do not accept the touch ou having abnormally reaction;
- do not respond to the name;
- poor imitation;
- low smile frequency and social reciprocity, as well as restricted social engagement (low initiative and low response availability)

- unusual interests, such as fixation on details;
- uncommon annoyance with loud sounds;
- moderate or severe sleep disorder;
- poor responsiveness at breastfeeding;

All of these markers are extremely valuable to those professionals who get the attention they need in childcare consultations. Generally, parents themselves draw attention to some characteristics that they find atypical. Here makes a difference that professional that listen and examines with all the attention.

In all cases, until the diagnosis is reached, must be used the screening, which is a brief, formal and standardized assessment, with the objective of early identification of patients with unexpected deviations from the norm (AUGUSTIN, 2019)¹⁻¹⁰.

Screening is defined as the instrument that allows detection of conditions / concerns that may not be apparent without the application of this tool. (AUGUSTYN, DURYEA, TORCHIA, 2019, p. 5)¹⁻¹⁰

In most cases screening does not provide a diagnosis; however, it helps to select candidates for diagnostic criteria. And this may be the first and the most important step. The participation of physicians and other professionals, usually organized in trained teams with special experience in autism spectrum disorder, is the "golden key" of this process.

Effective screening requires that standardized screening test results be considered in conjunction with clinical judgment. The choice of screening test depends on the age of the child, whether the child is being screened for the first time or has been identified through developmental surveillance or screening to be at risk for developmental problems.

EARLY SCREENING TOOLS FOR AUTISM

Many screening tools have been developed for use in children under three years of age. In this category are included: MCHAT R/F (Modified Checklist for Autism in Toddlers), ESAT (Early

Screening of Autistic Traits), STAT (Screening Tool for Autism in Toddlers and Young Children), ITC (Infant-Toddler Checklist), POSI (The Parent's Observations of Social Interaction).

1) MCHAT R/F

The Modified Questionnaire for Screening Autism in Children 16-30 Months Revised with Follow-up Interview (M-CHAT-R / F) is the recommended instrument by the Brazilian Society of Pediatrics. Must be clear from the beginning that, in fact, M-CHAT-R / F is a screening rather than a diagnostic test and is unique to early signs of autism and not to an overall neurodevelopmental analysis. (SOCIEDADE BRASILEIRA DE PEDIATRIA, 2019, p. 5)¹⁻¹⁰.

To improve the sensitivity and diagnostic specificity of ASD this tool has two stages to assess the risk of ASD in children between 16 and 30 months of age.

The first stage is a 20-item yes / no questionnaire for the parent attention. Parents are asked to remember their child's behavior in recent months and answer the questions on the list, which will generate a score.

The second stage consists of a follow-up questionnaire administered by a healthcare professional. It consists of the same questions as the first step, but investigates other information for items that failed the first stage.

This screening scale was validated through a study of more than 16,000 children aged 18 to 24 months during visits (routine appointments). Clinical reasoning was extrapolated from a psychologist or pediatrician, including tools for the diagnosis of ASD (Autism Diagnostic Observation Program (ADOS), the Childhood Autism Rating Scale-2, and the Child Interview. symptom of child autism. (AUGUSTYN, DURYEA, TORCHIA, 2019, p. 5-7)¹⁻¹⁰

Advantages of MCHAT - R / F:

1. It is available in many languages and translations into other languages are in progress.

2. Although copyrighted, it can be downloaded for free through the official M-CHAT website.
3. Revised version includes fewer questions, a different order of questions,
4. takes between 5 and 10 minutes to apply

SCORE	MEANING	ATTITUDE
0 – 2	LOW RISK	Repeat M-CHAT-R in 24 months
3 – 7	MEDIUM RISK	Administer the Follow-up Interview (second step of M-CHAT-R / F), if its score was more than 2 the child scores positive, if 0-1 scores negative.
8 – 20	HIGH RISK	Follow-up interview and the child should be referred for diagnostic evaluation and also for the need for intervention.

Source: Transtorno do Espectro do Autismo, Sociedade Brasileira de Pediatria p 11

2) ESAT (Early Screening of Autistic Traits)

Early Screening of Autistic Traits: The Dutch developed their own early autism screening system, we talk about the ESAT scale which is a 14-item yes / no questionnaire that targets early signs and symptoms of Autistic Spectrum Disorder (eye contact, joint attention) and no include skills that develop after age 15 months (for example, simulate play). One point is given for each "no" answer. ESAT takes approximately 10 minutes to be apply. It is available for free at DisabilityMeasures.org.

3) STAT (sensitivity and specificity of 92 and 85%) - Screening Tool for Autism in Toddlers and Young Children

Screening Tool for Autism in Young and Young Children (STAT) is an interactive scale that can be used for screening in children aged 24 to 36 months. It is designed to discriminate between

autism and other developmental disorders. It is a second stage screening, being used by primary care providers to improve screening in some programs.

STAT consists of a 20-minute session with 12 activities that are observed in four domains:

- playing (two activities),
- asking-request (two activities),
- directed attention (four activities)
- motor imitation (four activities)

The overall score ranges from lowest 0 to 4 higher points.

ADVANTAGE: Understanding language is not necessary.

DISADVANTAGE: Training is required for administration and scoring.

4) (ITC) Infant-Toddler Checklist - Sensitivity and Specificity of 88.9%

Known as the "Babies and Toddlers Checklist" is a 24-item questionnaire that is a component of the other scale (CSBS-DP). ITC is a delay tracker communication for children ages 6 to 24 months. The ITC is freely available from Brookes Publishing Company.

5) POSI - The Parent's Observations of Social Interaction - sensitivity 83% and specificity 75%

This is a seven-item parental report screening tool for autistic spectrum disorder in children ages 18 to 35 months. It was developed as part of a comprehensive primary care screening.

The POSI includes five of the six critical M-CHAT items and two behavioral questions based on the criteria of the Diagnostic and Statistical Manual of Mental Disorders (DSM).

LATE SCREENING TOOLS FOR AUTISM

Unfortunately, even with the advancement of technology and research on the diagnosis and screening of autism, most pediatric patients do not benefit from the advantages offered by the concept of early intervention. The ideal situation in which the patient is screened, analyzed and investigated before 30 months of age is quite rare. Most are diagnosed and included in treatment programs only after 3 years, sometimes even 4 or 5 years old.

Asking of pediatricians in São Carlos the advice about this, seems that this failure has the origin first in prejudice and second in the lack of knowledge itself. Sometimes the parents note that "something is wrong" with their son / daughter, but do not take action, sometimes even for fear of facing reality, just as sometimes because of the unfortunate "let's let the child arrive in her time". The school teacher realizes that the child has communication and socialization problems, but it prefer to remain silent.

According to the Brazilian Society of Pediatrics (2019)¹⁻¹⁰ "this tardy diagnostic aspect has been directly associated with low family income, ethnicity, few stimulation, few observation about children's development by parents, health

professionals, educators and caregivers. The belief of family members and health professionals that "must wait for the child's time", even when the child has obvious delays, is one of the factors that directly interfere with early detection." (SOCIEDADE BRASILEIRA DE PEDIATRIA, 2019)¹⁻¹⁰.

Even so, there are screening instruments (scales) for preschool and school age. We list here, Social Communication Questionnaire, Childhood Autism Spectrum Test, Autism Spectrum Screening Questionnaire, Autism Spectrum Quotient, AQ-Child.

As the purpose of this work is not the late diagnosis, these scales will have only a brief description. (AUGUSTYN, DURYEA, TORCHIA, 2019, p. 9)¹⁻¹⁰:

1. Social Communication Questionnaire is designed for use in preschool and school age children. The Social Communication Questionnaire is a parent report screen consisting of 40 yes / no questions. There are two forms, one for children under six and one for children six and older.
2. Childhood Autism Spectrum Test - formerly known as the Asperger's Childhood Syndrome Test is a 37-item parent-completed questionnaire designed for use in children ages 4 to 11 years.
3. For children ages 7 to 16 with suspected Asperger's disorder (and other high-functioning autistic disorders) we have the high-functioning Autism Spectrum Screening Questionnaire that takes around 10 minutes to be apply and is actually a 27-item checklist on a three-point scale.
4. The Autism Spectrum Quotient is a self-administered quiz for adults with normal intelligence and consists of 50 questions that assess social skills, attention, communication and imagination.
5. AQ-Child is a parental reporting measure for 4-11 year olds.

DISCUSSION:

The Autistic Spectrum Disorder screening tools are more accurate than the general developmental screening tools for Autistic Spectrum Disorder identification.

However, both sensitivity (ability to identify young children with Autistic Spectrum Disorder) and specificity (ability to discriminate Autistic Spectrum Disorder from other developmental disorders such as language disorders and overall developmental delay) appear to be limited. There are many cases of children who have early positive patterns, according to which they are at risk for Autistic Spectrum Disorder. After competent reassessments, analyzes and tests, these children do not meet the diagnostic criteria for Autistic Spectrum Disorder.

In fact, if the goal is to optimize early identification of children at risk for autism, the sensitivity is more important than specificity, at least for first-level testing.

The importance of age is well-known and we warn from the beginning that screening tools used for young children may be ineffective, or at least few sensitive when we trying to apply them to preschoolers or school-age children.

We must not forget that older children with Autistic Spectrum Disorder and those who have received appropriate intervention can and do acquire some skills. This can impair screening testing, especially in the case of age-inappropriate application. (MAGLIONE, 2019, p. 130)¹⁻¹⁰

CONCLUSIONS:

Early diagnosis of autism spectrum disorder today is no longer a simple experimental choice. The recent experience and the results obtained through the application of various strategies show a great way to improve the final result. Today, we have strong evidence that early diagnosis leading to early therapy is the right way for a case of autistic spectrum disorder to regain hope of integration into society. There is scientific evidence that the concept of early intervention may in some cases even prevent

the complete clinical installation of autistic spectrum disorder.

The first step is always a competent screening, based especially on teamwork. Published research recommend that all children be screened, even if they are not suspected of having autism spectrum disorder. Several scales and various protocols are described in the literature, each presenting its own advantages and disadvantages, and it is obvious that none of these screening instruments is the ideal method. However, it needs to be clear from the beginning that a screening test is not an instrument to diagnose, but rather to make a first selection that will later be ascertained by a multiprofessional team.

It is very important to know that parental concerns are critical parameters and need to be carefully considered. Moreover, it is recommended to health professionals to encourage all those who interact with a child: teachers, nannies, parents, relatives, caregivers to observe and report any strange behavior of the child, even if that is "positive" - any atypical behavior of a child can change a perspective.

Finally, we remember that the age of maximum cerebral malleability is 12-24 months. Early intervention has to start at this age, as the chances of integration and recovery of some skills are maximum for the autistic. Any delay, neglect and omission of symptoms can cause irreversible damage.

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