

Evaluation and Diagnosis of Autism Spectrum Disorders in Preschools

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ABSTRACT

Background: The purpose of this literature review is to document the standardized autism diagnostic tools that exist both globally and in the Greek population. More specifically, the current study focuses on autism assessment tools that can be administered from early childhood and are based both on clinical observation of the child and on data collection from questionnaires that are self-completed by parents or by the clinician.

Methods: To carry out this literature review, books, contemporary texts as well as the following search engines were used: Google Scholar, Science Direct and PubMed. After screening the content of the literature references so that they correspond to the topic of this study, 38 literature references were used.

Results: According to the present study, it appears that standardized diagnostic tools based on clinical observation and interaction with the examinee are few, while assessment scales that involve obtaining information through questionnaires prevail. More specifically, the standardized diagnostic tools for autism in Greece are insufficient as the only one mentioned in this article that has been translated into Greek is the M-CHAT and the M-CHAT-R/F.

Keywords: Autism; Assessment tools; Toddlers; Diagnosis

INTRODUCTION

Autism Spectrum Disorder is an extremely complex neurodevelopmental disorder, characterized by deviations from the typical developmental course [1]. Diagnostic criteria for the disorder are determined by the Diagnostic and Statistical Manual of Mental Disorders (DSM-5). Specifically, there are deficits in social communication and social interaction in a variety of contexts, as well as restricted and repetitive behavioral patterns, interests and activities. For the diagnosis, the symptoms must be present from early childhood and affect every day functioning and the deficits cannot be better explained by intellectual disability or global developmental delay [2].

Moreover, individuals in the autism spectrum, also present deficits in the “theory of mind”, which means difficulty in understanding the point of view of other people, something that affects their behavior in social and interpersonal areas [1].

There are signs that need to be evaluated, from early childhood. Specifically, children that may have autism spectrum disorder do not maintain eye contact, do not respond to smiling, do not react when hearing their name or a familiar voice, do not visually follow objects, do not make noises to attract other people's attention, do not respond to hugging, do not imitate movements or facial expressions, do not reach out their hands to be held and do not interact during playtime [1].

The exact etiology of autism is not fully known. It is considered that autism is primarily linked to the prenatal period of a child's development and specifically that it is related to the development of the brain in terms of structure and neurochemistry. It is therefore considered that this disorder is mainly biologically defined. It has been found, that it is more common in mothers who have been previously exposed to perinatal complications or infections, such as rubella, toxoplasmosis, chickenpox and parotitis [1]. According to Hnida

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and Deer, as referred by Kroncke et al. It is argued that autism can be attributed to a combination of environmental and genetic factors, because there are certain risk factors during pregnancy and early childhood that influence the manifestation of the disorder. Specifically, the age of the parents, the mother's exposure to environmental toxins, common womb diseases or maternal obesity may trigger the onset of this neurodevelopmental disorder [3].

LITERATURE REVIEW

The prevalence of autism spectrum disorders is estimated at a ratio of 2-5 births of children with autism spectrum disorders per 10.000 births, in the child population [1]. The numerical superiority in the prevalence of the disorder is boys, who are affected four times more than girls [1,4]

MATERIALS AND METHODS

Assessment of autism spectrum disorders

Early screening and diagnosis of Autism Spectrum Disorders is significant for determining the severity of the disorder and for detecting any possible comorbidity. Therefore, the results of diagnostic tools are useful, in order to create a personalized treatment program that will meet the needs of each individual [5-6]

Autism can be detected at 18 months of a child's life, however, the diagnosis can be made from 2 years of age and later by an experienced clinician. Since there are no medical tests, such as blood tests, through which autism can be diagnosed, the clinician must study the history as well as the child's behavior in order to make the diagnosis. The National Center on Birth Defects and Developmental Disabilities (NCBDD), states that children should be evaluated for the possibility of autism existence at 9 months, 18 months and at 24 or 30 months of age. The American Academy of Pediatrics (AAP) recommends that the autism assessment should be made at 18 months and then at 24 months of a child's age [7]. The results of the diagnostic assessment do not necessarily provide a final diagnosis and may indicate the need for a more extensive and formal developmental assessment, where the clinician could be able to identify if a child has indeed an autism spectrum disorder [8]. There are several diagnostic tools for autism which are carried out through clinical practice or through questionnaires answered by parents. The most widely used autism assessment scales are the Autism Diagnostic Interview, Revised (ADI-R) [9], the Autism Diagnostic Observation Schedule-Second Edition (ADOS-2) [10], the Childhood Autism Rating Scale- Second Edition (CARS-2) [11], the Diagnostic Interview for Social and Communication Disorders (DISCO) [12-13], the Modified Checklist for Autism in Toddlers, Revised (M-CHAT-R) [14] and the Autism Behavior Checklist (ABC) [15].

Diagnostic and screening tools for childhood autism

Autism Diagnostic Observation Schedule (ADOS) και Autism Diagnostic Observation Schedule-Second Edition (ADOS-2)

The ADOS-2 [16] was created to improve the accuracy and effectiveness of the ADOS diagnostic tool [17]. It is a new upgraded, semi-structured and standardized tool that assesses communication, social interaction, play and imagination in it, as well as restricted or repetitive behaviors. It was published by the Western Psychological Services. It is used in clinical practice as well as in research settings and is often referred to as a "gold standard" assessment and observation tool for pervasive developmental disorders [18].

It can be administered and interpreted by appropriately certified professionals from the fields of medicine, psychology or related professions. It lasts approximately 40 to 60 minutes, depending on the modules that are chosen to be administered and by the specific behavior that each examinee exhibits [16].

ADOS-2 was designed to be administered individually to each examinee by a clinician. It consists of 5 modules, one of which is administered, depending on the cognitive level and chronological age of the examinee. Each module has been designed to elicit communication, social interaction and behaviors that are potentially related to autism. ADOS-2 differs from other assessment tools for autism, because it requires the clinician to have constant interaction with the examinee during the whole administration process. Scoring must be done immediately after the administration and examinee's behavior should also be documented. The presence of a parent or caregiver is required during the assessment process for the Toddler Module, Module 1, and Module 2. The Toddler module is administered to children from 12 to 30 months of age, who communicate with single words and short phrases. Module 1 was designed for children aged 31 months and above, who communicate with single words and phrases and it consists of 10 activities. Module 2 is administered to individuals of all ages who communicate with phrases but do not have fluent speech and it consists of 14 activities. Module 3 assesses children with fluent speech aged 6 to 16 years and Module 4 is suitable for children and adolescents over 16 years of age [19].

The ADOS-2 kit contains most of the required objects and materials (some materials must be provided and renewed by the examiner), as well as 10 protocols for each of the 5 modules. The handbook is extensive and includes a review of the tool, guidelines for the module selection that is to be administered, directions for the administration and for the coding procedures of the results, as well as instructions for each module and example of cases, that may help the clinician in interpreting the results [10].

Autism diagnostic interview, revised (ADI-R)

The ADI-R diagnostic interview [9], is an inclusive interview for the parents, which aims to detect potential autism symptoms in their children. It is administered by a trained clinician with the use of a semi-structured form of interview. The full version of the ADI-R which is mainly used for research purposes, requires

approximately three hours to be administered, while the short version of the ADI-R, which only contains the diagnostic algorithm elements, may be used for clinical assessment and lasts approximately 90 minutes [9].

The ADI-R diagnostic interview can be administered to all age groups but primarily to children aged 4-5 years. The reason why it is administered in these age groups is because children at that age are old enough to exhibit a range of behaviors that may be related to autism and young enough not to experience significant changes that may occur with age [9].

This diagnostic tool, includes questions related to the present behavior of the child and its developmental history. Specifically, it contains questions about the family and the education of the examinee, previous diagnosis or treatment, in order for the clinician to understand the behavior, the developmental course, the language level, the social and communicative functioning level, the social development level and play skills, the interests and the behaviors of the examinee [20].

The elements that distinguish children with autism from those with other developmental delays are summarized in three algorithm scores, which calculate social difficulties, communication deficits and repetitive behaviors. The algorithm score distinguishes children with autism from those with other developmental disorders, such as severe perceptual speech disorder and other developmental disorders [9]. Despite the fact that ADI-R is a very useful tool for the assessment of autism, there are certain limitations. Specifically, it is not sensitive for children with mental age below 20 months as well as for individuals with IQ<20 [9]. Furthermore, it has been designed to confirm the first autism diagnosis and it is not suitable for re-evaluation [21].

Childhood autism rating scale-second edition (CARS-2)

The CARS autism assessment scale [22-23], is based on the observation and information obtained from parents and teachers and it can be administered to individuals aged from 18 months to 35 years [24].

The CARS-2 second edition of childhood autism assessment scale [25], remained the same with the previous edition for the administration in children aged below 6 years with communication difficulties or with an IQ below average, while CARS-2-HF, which was recently developed for the assessment of individuals with verbal fluency from 6 years of age and older with an IQ>80 [26].

This scale consists of 14 domains which assess behaviors related to autism, as well as one domain which assesses the general clinical presentation of the Autism Spectrum Disorder, to diagnose autism and differential diagnosis from other disorders [27]. The 15 assessment domains examine the relationship between the examinee and people, the imitation ability, the emotional response, the body use, the object use, the adaptive ability, the visual response, the auditory response, the perceptual ability, the fear or the anxiety, the verbal communication, the

non-verbal communication, the movement and the intellectual ability [28].

Each domain is scored on a scale ranging from 1 to 4. The highest scores are associated with a higher level of dysfunction and the total score of the scale can range from 15 to 60 points. Scores below 30 points indicate that the individual does not belong to the Autism Spectrum Disorder population, scores between 30 and 36.5 points indicate mild to moderate severity autism and scores from 37 to 60 points indicate the presence of severe autism. The psychometric properties of the CARS scale are very well documented [27].

Diagnostic interview for social and communication disorders (DISCO)

The DISCO diagnostic interview of social and communication disorders [12-13] is a comprehensive semi-structured interview which is used by clinicians to elicit information from the caregivers, about the development and behavior of the examinee. It was designed to detect a wide range of developmental skills and behaviors based on the autism spectrum. It includes questions about stereotypical and repetitive behaviors, repetitive motor behaviors, verbal stereotypes, unusual sensory interests and unusual reactions to sensory stimuli, persistence in routine and restricted interests [29]. This diagnostic tool has been designed to be administered in all age groups from early childhood to adulthood and is sensitive in detecting any level of ability. The initial goal is to understand the pattern of social behavior and communication over time, as well as the strengths and weaknesses of an individual's behavior. In this way, it is possible to classify all aspects of the autism spectrum from the mildest to the most severe [30].

Modified checklist for autism in toddlers, revised (M-CHAT-R)

The Checklist for Autism in Toddlers [31], which was one of the first standardized autism diagnostic tools, was designed to investigate the possibility that children who have not developed joint attention and symbolic play by the age of 18 months may have autism. This diagnostic tool was developed to be administered in children aged from 18 months and older. The scale administration lasts from 5 to 10 minutes and is easy to score. It consists of nine questions in part A, which are asked by the medical visitor or by the general practitioner and are answered by the child's parent. After that, the specialist who is administering the assessment scale, completes the five subtests of part B through direct observation of the child. There are five "key items" which are related to joint attention and symbolic play. The "key items" in part B are included to validate parents' answers in part A. The rest of the items provide additional information to discriminate a special autistic profile from other global developmental delays and they also provide the opportunity to all parents to answer positively in some questions [31].

Children who fail in all five "key items" are at a higher risk of developing pervasive developmental disorders [31].

The Modified version of the Checklist in toddlers (M-CHAT) [14] was then developed, due to the limited diagnostic value of the previous version, as it was not able to detect the autism signs 6 months after the first diagnosis [32]. The M-CHAT also included questions answered by parents. Then, the Quantitative CHAT (Q-CHAT) [33] was developed, in which closed-ended questions (yes/no) were replaced by open-ended questions. Although M-CHAT is one of the most widely used standardized autism diagnostic tools, it remains diagnostically unreliable over time, maybe because it is not sensitive in diagnosing older children [34]. For that reason, the Modified version (M-CHAT-R) [35] was developed, which consists of two stages of questions answered by parents, in order to identify the possibility of autism in toddlers aged 16 to 30 months. During the re-assessment process it is very likely that a child that did not present signs of autism, does not belong in the spectrum, but rather presents another developmental disorder [35]. The M-CHAT has been translated into Greek [36-38].

Autism behavior checklist (ABC)

Another diagnostic tool for assessing childhood autism is the Autism Behavior Checklist (ABC) [15]. This assessment scale is administered in individuals aged between 18 months and 35 years [24]. It describes autism spectrum behaviors and its aim is to detect corresponding behaviors in each examinee. It aims to evaluate the existence of potential autistic behaviors. The assessment form consists of 57 behaviors, which are divided into 5 subcategories. Specifically, 9 behaviors are related to sense, 12 to relating, 12 to stereotypes and the use of objects, 13 to communication and 11 to self-help and social interaction. The overall score of the scale is obtained by adding the results from each assessment area. Based on the score, examinees are classified in the following categories: normal individuals, individuals with severe emotional disorders, deaf/blind individuals, individuals with severe intellectual disability and individuals with autism [28].

DISCUSSION

Autism is a neurodevelopmental disorder, the etiology of which is not fully clear [39]. As autism symptoms affect a child's everyday functioning, early assessment is essential for designing an individualized therapy program that will aim to improve the deficits that each patient presents [5-6]. Given that this complex neurodevelopmental disorder cannot be diagnosed by any physical medical examination, the developmental history as well as the observation of the child is necessary to identify the present disorder [7]. According to the present literature review, few autism diagnostic tools that are based on the clinical assessment of the child have been detected and the most widely used of these is the Autism Diagnostic Observation Schedule-Second Edition (ADOS-2) [10] which can be administered to all age groups from children aged 12 months to adulthood [19]. Furthermore, the Childhood Autism Rating Scale-Second Edition (CARS-2) [25], which includes clinical observation, can be administered to children aged 18 months to 35 years [24]. For the most part, questionnaires have been designed to identify some neurodevelopmental disorder and the most widely used

are the Autism Diagnostic Interview, Revised (ADI-R) [9] and the Diagnostic Interview for Social and Communication Disorders (DISCO) [12,13], which can be administered in all age groups from early childhood to adulthood [20,30]. Moreover, a widely used diagnostic tool that consists of questions is the Modified Checklist for Autism in Toddlers, Revised (M-CHAT-R) [35], which was designed to investigate the possibility of autism existence in children aged from 16 to 30 months [35].

RESULTS

Taking everything into consideration, autism assessment both through clinical observation and through questionnaires that are self-completed by parents or by the clinician, is essential to establish the diagnosis of autism and subsequently design an individualized therapy program that will help each patient with the clinical symptoms that they present. According to the present literature review, it appears that diagnostic tools that include clinical observation are limited, while assessment scales that are based on questionnaires are more widely used. Moreover, in the present study it is noted that there are no standardized diagnostic tools for autism spectrum disorders in Greece, as the only ones available in Greek are the M-CHAT and the M-CHAT-R/F which are translated but not standardized. Therefore, there is a need to develop additional diagnostic tools adapted to the language level and age group of the individuals, based both on clinical observation and interaction with the child and on questionnaires that will be self-completed by the parents or by the clinician. Finally, it is concluded that the standardization of diagnostic tools globally and in the Greek population is necessary to thoroughly and scientifically diagnose this neurodevelopmental disorder, in order to design the appropriate therapeutic intervention that will begin from early childhood.

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