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The Role of Tests and Tools in Accurate Diagnosis and Severity in Children with Autism Spectrum Disorders

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A B S T R A C T

Objective: The current research was conducted with the aim of studying and investigating the role of tests and tools in the accurate diagnosis of autism spectrum disorders.

Methods: A total of 83 children (56 boys and 27 girls) diagnosed with autism or one of the comorbid diagnoses were selected and underwent the GARS-3 scale and the Psycho-Educational Profile Revised (PEP-3) diagnostic test. Additionally, parents were asked to complete a checklist provided by the researcher. The age range of the individuals with autism was between 2 and 7 years, with an average age of 6.5 years.

Findings: The results of the statistical analyses of the data from this study indicated a significant role of diagnosis tools.

Conclusion: So, the proper and timely use of diagnostic tests plays a significant role in the accurate diagnosis of autism spectrum disorders and also in determining the severity of these disorders.

Keywords: *Test, Tool, Diagnosis, Severity, Autism*

1. Introduction

Until the mid-20th century, there was no name for the disorder now known as autism spectrum disorder or autism. In 1943, Dr. Leo Kanner at Johns Hopkins Hospital studied a group of eleven children who exhibited characteristics such as solitude, introversion, inability to produce certain expected bodily gestures, developmental delays, echolalia, pronoun reversal, monotonous voice and speech, excellent eidetic memory, stereotypical behaviors, insistence on environmental sameness and resistance to change, poor eye contact, and an abnormal relationship with others, and lifeless objects. Although Kanner labeled these children with autism, they were classified under mental

retardation or schizophrenia until the 1980s. It was later clarified that although this disorder could co-occur with childhood schizophrenia, they are distinct phenomena (Fombonne, 2003). These children have sensory processing disorders, thus exhibiting abnormal responses to sensory stimuli (such as avoidance behaviors and hypersensitivity) (Jasmin et al., 2009; Kientz & Dunn, 1997), emotional (Rapin, 1991), motor skills—particularly fine hand movements—daily activities, and play (Restall & Magill-Evans, 1994; Watling et al., 2001), and difficulties in learning natural language and speech (Corbett & Prelock, 2006) and in mimicking others (Schopler et al., 1984; Schopler et al., 2010). Overall, these children exhibit a wide range of psychological and medical disorders (Klin, 2006). Autism is a developmental neurological disorder in children, generally recognized as a neurodevelopmental brain disorder, but little is known about its exact origin (Järvinen-Pasley & Heaton, 2007). There is no general agreement about structural defects in the brain of these individuals, and no biomarker has been identified for a definitive clinical diagnosis. Thus, the diagnosis of autism is generally based on the observation of the aforementioned clinical symptoms (Tanel, 2009).

Since this disorder was recognized in the Diagnostic and Statistical Manual of Mental Disorders (DSM) as a distinct disorder (Sung et al., 2018), there has been considerable research related to autism spectrum disorders, attributed to the increasing prevalence—from a prevalence of 1 in 1000 in 2010 (Matson et al., 2012) to 1 in 68 in 2016 (Duda et al., 2016), with a prevalence of 3.8 per 10000 in Asia (Jin et al., 2018) and 2.95 per 10000 in Iran (Samadi & McConkey, 2015). Reasons for this increase include improved diagnostic levels, increased awareness, and a detailed definition of the disorder (Neggers, 2014). Regardless of the prevalence of autism spectrum, early diagnosis is important for the child and family to benefit from therapeutic-educational interventions and support systems (Van Daalen et al., 2009). As age increases, not only do costs increase, but the treatment process also becomes more difficult (Rey et al., 2019). Therefore, special attention is given to the development of screening tools for ages under 30 months (Al Maskari et al., 2018). The diagnosis of autism spectrum disorders is important as it leads to timely interventions, resulting in more effective social interactions and stereotypical behaviors in autistic children (Randall et al., 2018). Therefore, the development and compilation of identification tools should be a priority for screening and diagnostic purposes (Reza Gorji et al., 2021; R. Gorji et al., 2021).

Although the development of diagnostic tools for adults has always been considered, most tools emphasize diagnosis in early life (Wigham et al., 2019). Therefore, for diagnosing this disorder, in addition to clinical observations by specialists, the use of valid psychological tools is important (Durkin et al., 2015), which according to the American Psychiatric Association is carried out in two stages: initial screening and multi-axial diagnostic evaluation in the second stage (Pang et al., 2018). This process for an accurate and timely diagnosis at one year of age requires a developmental monitoring program, and otherwise, for ages under 18 months, parent questionnaires and other diagnostic tools can be used, overall highlighting the importance of tools for identifying autism spectrum disorders (Reza Gorji et al., 2021; R. Gorji et al., 2021).

Clinical specialists must first be able to diagnose mental disorders to treat them. The diagnostic process requires that clinical specialists have a systematic approach to classifying the disorders they see in their patients. The role of the diagnostic guide is to provide coherent diagnoses in individuals based on the presence or absence of a specific set of symptoms. Without an accurate diagnostic guide, it is impossible for a clinician to decide on the best treatment path for a specific patient. Researchers also use the diagnostic guide to provide a coherent vocabulary for researchers to use when reporting their findings. The diagnostic process involves using all relevant information to arrive at a name that describes the patient's disorder. This information includes the results of any tests given to the patient, information gathered from interviews, and knowledge of the patient's personal history (Reza Gorji et al., 2021; Randall et al., 2018).

Based on what has been discussed, there is an unavoidable need to design, develop, and standardize tests related to autism spectrum disorder for all ages and severity levels for the Iranian community. Therefore, to achieve this, it is first necessary to evaluate the existing tools in two areas and correct interviewing principles based on DSM5 regarding children with autism spectrum disorder, and also to address this issue using correct tools (a combination of several diagnostic tools) (Durkin et al., 2015; Samadi & McConkey, 2015). Thus, the norm-setting of tests designed in other countries also becomes important in our country.

The results indicate that overall, three groups of tools are distinguished from each other in this context: a group used for diagnosis in childhood, specifically tools related to

diagnosis from birth to 13 years of age; a group known for diagnosing autism spectrum disorders in adulthood; and finally, a group that can identify all age groups from birth to adulthood. Regardless of the age group these tools are designed for, they have been developed based on the DSM criteria from its third edition in 1980 to the fifth edition in 2013, and naturally, the latest tools are being developed based on the fifth edition, which has sensitivity and specificity coefficients over 0.9 (Neggers, 2014), thus the new tools have greater diagnostic power (Reza Gorji et al., 2021).

Cultural differences in identifying symptoms of autism are one of the reasons why the prevalence of autism spectrum disorders is reported to be lower in non-Western societies. Studies show significant differences in societies in rating non-verbal communication, socialization, verbal communication, insistence on environmental consistency, and restricted behaviors. Therefore, parents and professionals in a specific society may have a completely different approach to symptoms and syndromes that are problematic and abnormal in other societies and consider them normal. Migration of ethnic groups from one society to another also indicates that the prevalence of autism is higher than what had been reported among these groups in their original society. For example, the immigrant African and Middle Eastern communities in Sweden have a much higher prevalence of autism than the native Swedish population. It should be noted that the prevalence of autism in Sweden is among the highest reported globally, and the mentioned immigrant communities, considered as developing, not only lack official reports on the prevalence of autism but estimates predicted the occurrence of this disorder to be very rare (Barnevik-OLSSON et al., 2010; Gilliver et al., 2014).

As previously mentioned, timely identification and provision of services at the right time, known as timely interventions, have the greatest positive impact. Providing services and necessary guidance to parents and individuals with autism in the early stages of diagnosis is crucial (Wallace & Rogers, 2010). The first step in this process and providing appropriate services is screening children, which usually occurs between 18 to 24 months of age and in the pre-school stage. Various tools have been created for this purpose. Reports are usually provided by parents or caregivers and these tests are generally conducted with indirect information (Allison et al., 2012), but recently a practical test for autism screening has been developed that uses interactive activities lasting between 5 to 10 minutes to identify children at risk of autism (Choueiri & Wagner, 2015). Individuals who achieve the cutoff score are referred to specialists capable of conducting diagnostic tests. Most screening tools beneficial for identifying children with autism have been developed and refined in Western cultures, but their applicability in low and middle-income countries is questionable, and the validity of these tests decreases significantly if used in other societies (Wallis & Pinto-Martin, 2008).

Therefore, the selection of items included in the screening test, which are very important, and the effectiveness of the screening test in terms of sensitivity to these important items, require further study and should be carefully examined and considered among immigrant communities to the West as well. Based on this, a 10-item scale named Hiwa was proposed for use in Iran (Samadi & McConkey, 2014; Samadi & McConkey, 2015) derived from 800 individuals with autism using the second edition of the Gilliam Autism Rating Scale (GARS-2) (Reza Gorji et al., 2021) in a field survey in Iran for screening the preschool to 11-year-old age group (Samadi & McConkey, 2014). This test is based on the fourth edition of the Diagnostic and Statistical Manual of Mental Disorders and the tenth edition of the International Classification of Diseases. The Hiwa test consists of 10 selected items; 5 items relate to deficits in social interactions, 3 items reflect behavioral problems, and 2 items represent communication problems. Items 1, 5, and 9 are also present in the M-CHAT.

Therefore, the current research was conducted with the aim of studying and investigating the role of tests and tools in the accurate diagnosis of autism spectrum disorders.

2. Methods

2.1. Study design and Participant

This research is a descriptive-survey study with an applied objective. The primary goal of the researcher in conducting applied research is to derive principles and rules that can be applied in real-world and practical situations, assisting in the improvement of products and the efficiency of operational methods.

The initial target population for this study consisted of all children and adolescents aged 5 to 14 years, who were first-time attendees at private counseling centers, welfare centers, and special education schools across four geographical areas (North, South, East, and West) of Tehran, receiving diagnoses of autism spectrum and other comorbid neurodevelopmental disorders. However, due to significant challenges in sample collection, non-cooperation from

centers and even the national special education organization, the researcher was compelled to utilize WhatsApp groups. Using a snowball sampling method, participants were identified through mothers who were members of these groups. The study continued with a census of 51 samples diagnosed with autism spectrum disorders and 32 samples with other comorbid disorders. Inclusion criteria were having only one diagnostic label, physical health, and no use of any type of medicinal or non-medicinal treatment. Due to operational limitations, only physical health and age could be strictly controlled.

2.2. *Measures*

2.2.1. *Social Communication Questionnaire (SCQ)*

Created by Rutter, Bailey, & Lord (2003), this tool consists of 40 items designed to screen for autism in individuals over 4 years of age. SCQ assesses communication skills and social functioning based on parent or caregiver reports. It is scored on a dichotomous scale (yes/no), with higher scores indicating more severe symptoms of social and communication deficits. Its validity and reliability are well-established in the literature (Reza Gorji et al., 2021; R. Gorji et al., 2021).

2.2.2. *Childhood Autism Rating Scale (CARS)*

Developed by Schopler, Reichler, DeVellis, & Daly (1980), CARS includes 15 items that measure behavior on a scale from 1 to 4. It is used to assess the severity of autism based on behavioral observation across multiple subscales including relational and communicative aspects. The scale's scoring reflects the severity of autism from mild to severe, and it has demonstrated high reliability and validity (Reza Gorji et al., 2021; R. Gorji et al., 2021).

2.2.3. *Autism Diagnostic Interview-Revised (ADI-R)*

Authored by Lord, Rutter, & Le Couteur (1994), this is a structured interview used with caregivers, encompassing over 93 items. It covers the participant's full developmental history and assesses behaviors related to autism spectrum disorders. It is noted for its diagnostic validity and reliability in differentiating autism from other developmental disorders (Reza Gorji et al., 2021; R. Gorji et al., 2021).

2.2.4. *Autism Diagnostic Observation Schedule (ADOS)*

Created by Lord et al. (2000), ADOS is an observation scale that includes standardized tasks that allow the observer to judge the social and communicative behavior of the child. The tasks are divided among different modules which are suitable for children and adults across varying developmental and language levels. The ADOS is highly regarded for its clinical validity and reliability (Reza Gorji et al., 2021; R. Gorji et al., 2021).

2.3. *Data Analysis*

Data were analyzed using the Chi-square test to determine the relationship between the use of various diagnostic tools and the accuracy of autism spectrum disorders diagnoses. This statistical method was chosen to test for independence between observed frequencies in the categorized data obtained from the diagnostic assessments and the predefined diagnostic categories.

3. Findings and Results

As observed in Table 1, 50% of diagnoses were made using the autism diagnostic tool, 17% by other tools, and in 33.7% of cases, there is no information available about the diagnostic tool used.

Table 1

Frequency of Diagnostic Tools Used

Tool	Frequency	Percentage	Cumulative Percentage
Unknown Tool	28	33.7%	33.7%
Autism Diagnostic Tool	41	49.4%	83.1%
Other Tools	14	16.9%	100%

The chi-square value is significant (Table 2), indicating that the type of diagnostic tool used significantly affects the diagnosis ($p < 0.05$).

Table 2

Chi-Square Test Results and the Impact of Tools on Diagnosis

Tool	Missed Diagnosis	Correct Diagnosis	Incorrect Diagnosis	Total	Difference in Severity	Chi-Square Value (df=6)	p
No Tool	13	1	13	28	1	66	0.0
Autism Tool	11	26	0	41	4		
Other Tools	5	0	1	14	8		

As observed in Table 3, 47% initially had a severity score of 3 (severe) and 35% a score of 1 (mild). In secondary severity, 40% had a score of 1 (mild) and 31% a score of 3 (severe). The results indicate that severity is predictive in secondary diagnoses ($p < 0.05$), whereas it is not effective in initial diagnoses ($p > 0.05$).

Table 3

Frequency of Autism Spectrum Disorder Severity

Severity Level	Severity Score	Frequency	Percentage	Cumulative Percentage
Initial	1	29	34.9%	34.9%
	2	15	18.1%	53.0%
	3	39	47.0%	100%
Secondary	1	33	39.8%	39.8%
	2	24	28.9%	68.7%
	3	26	31.3%	100%

Table 4 show the significance of the diagnostic tools and the severity levels in influencing the accuracy of autism diagnoses within the studied population.

Table 4

Chi-Square Test Results and the Impact of Disorder Severity on Diagnosis

Severity Level	Missed Diagnosis	Correct Diagnosis	Incorrect Diagnosis	Difference in Severity	Chi-Square Value (df=6)	p
Initial					0.536	0.060
1	12	11	4	2		
2	5	6	2	2		
3	12	10	8	9		
Secondary					0.038	13.32
1	13	11	6	3		
2	7	6	2	9		
3	9	10	6	1		

4. Discussion and Conclusion

In recent years, there has been a noticeable trend towards the early diagnosis of autism spectrum disorders. This trend is driven by the efficacy of early interventions during the initial years of childhood for these disorders. For these interventions to be implemented, early diagnosis is imperative. To achieve this, we rely on tools that assist in screening and diagnosing. In the current study, various tools were examined for their advantages and disadvantages. The effective factors in diagnosis such as the use of tools, age, gender, evaluator expertise, and the severity of the disorder were analyzed. The findings suggest that the correct use of tools can play a significant role in the accurate diagnosis of autism spectrum disorders and their severity. Meanwhile, the age and gender of the children did not impact the identification and diagnosis of the disorder. Also, the expertise of the evaluator could not play a significant role, and anyone who has undergone the necessary training to use tools related to autism spectrum disorders can perform the diagnosis and screening.

From the information obtained from families, it was noted that most had initially sought a neurologist or pediatrician

after encountering behavioral problems in their child. These families were unaware of the existence of specialists in child psychology and doctors for these children. Since these specialists primarily rely on clinical signs and personal experience to identify these children and less on autism-specific tools, numerous misdiagnoses occur. It is possible that an individual may lack adequate experience and give an incorrect diagnosis. Conversely, someone working with autism diagnostic tools, even if not highly specialized, has definitely undergone training in using these tools and covers more than half the journey based on correct tools; the rest depends on the individual's accuracy and personal experience. This is particularly evident in differential diagnoses where a child with autism might be mistaken for having intellectual disability or hyperactivity, leading down a wrong therapeutic path that could cause irreparable harm to the child and incur significant costs for families. These families have spent considerable time in confusion and anxiety where incorrect diagnoses were made, consequently losing the golden period for treating their child. Therefore, it is recommended to exercise greater care and precision in identifying children with autism to expedite their identification and ensure timely intervention. Another point for consideration is the use of experts and specialists in exceptional children in this field. It may be that other professionals are trained on autism or hyperactivity and can make accurate diagnoses, but their perspective on the issue differs completely from someone who has studied and mastered all disorders related to children. This is clearly seen in evaluators with degrees in general psychology, education, or counseling who are committed to this issue. A specialist in children's issues considers all disorders at once, clearly seeing which the child is more aligned with or the possibility of two disorders co-occurring. Another important aspect to be considered is the coordination of relevant institutions like special education, welfare organizations, and medical and research centers in diagnosing autism. The special education organization specifically conducts screening for children with and without autism spectrum disorders at the start of elementary school, initially screening children with a Social Communication Questionnaire. If a child scores 9 out of 21, they must be further evaluated with the revised version of the Autism Diagnostic Interview for a final diagnosis, and if they meet the necessary criteria in this test, they are deemed a candidate for entry into a school with special needs for autism spectrum disorders. However, the welfare organization that covers rehabilitation centers has not provided specific guidelines for using any particular tests or tools, and ultimately medical and research centers make diagnoses based on the evaluator's interests (Reza Gorji et al., 2021; R. Gorji et al., 2021). This disparity in tool usage could be influential in increasing the number of people diagnosed with autism spectrum disorders (Neggers, 2014), highlighting the importance of using specific and uniform tools for autism.

5. Suggestions and Limitations

The study faced several limitations that may affect the generalizability and interpretation of its findings. Primarily, the reliance on snowball sampling through WhatsApp groups due to non-cooperation from centers may have introduced selection bias, limiting the diversity and representativeness of the sample. Additionally, the use of self-report measures for collecting data from parents might have led to response biases, affecting the accuracy and reliability of the data. The study also lacked a control group, which would have strengthened the causal inferences of the effectiveness of diagnostic tools. Moreover, the evaluators' varying levels of expertise and possible inconsistencies in using the diagnostic tools could have influenced the consistency and reliability of the diagnoses. These factors collectively suggest that the results should be interpreted with caution, and further research using more rigorous sampling and methodological approaches is needed.

Given the discussion, attention to the following points can be crucial in identifying and accurately diagnosing autism spectrum disorders, thus it is recommended that: 1 - Diagnostic tools based on DSM-5 are utilized and considered. 2 - Responsible organizations and related parties standardize the evaluation process to reduce confusion. Therefore, it can be suggested to use uniform tests for identifying autism spectrum disorders. 3 - It is also recommended to use complementary tests and not to rely solely on the results of one test. This is an important consideration that test creators strongly emphasize, believing that no test is perfect or 100% accurate. 4 - Another significant recommendation is to make more use of specialists in this field, as is naturally practiced in medicine. For example, while any doctor can treat a sick child, most families prefer to consult a pediatric specialist.

Authors' Contributions

All authors have contributed significantly to the research process and the development of the manuscript.

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Declaration

In order to correct and improve the academic writing of our paper, we have used the language model ChatGPT.

Transparency Statement

Data are available for research purposes upon reasonable request to the corresponding author.

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Declaration of Interest

The authors report no conflict of interest.

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Ethical Considerations

The study protocol adhered to the principles outlined in the Helsinki Declaration, which provides guidelines for ethical research involving human participants.

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