

Tools for Early Screening of Autism Spectrum Disorders in Primary Health Care – A Scoping Review

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Abstract

Background. Autism spectrum disorder (ASD) is a neurodevelopmental disorder that manifests itself in early childhood. Early diagnosis of these disorders allows for the initiation of early therapy, which is crucial for the child's further functioning in society.

Objectives. This review aims to gather and present the existing ASD screening tools used in primary care and adapted to different countries conditions linguistically and culturally.

Eligibility criteria. We searched for English-language publications on ASD screening tools for children aged 0-3 years suitable for use in primary care (i.e. free, requiring no additional training or qualifications).

Sources of evidence. Four databases were explored to find English studies on ASD screening tools intended for the rapid assessment of children aged 0–3.

Charting methods. The information sought (specific features of the questionnaires relevant to primary health care workers, psychometric and diagnostic values of a given cultural adaptation of screening tools, and the linguistic and cultural changes made) were extracted and collected to create profiles of these tools.

Results. We found 81 studies which met inclusion criteria and underwent full data extraction. Three additional data sources were included. These allowed to create 75 profiles of adaptations for 26 different screening tools and collect data on their psychometric values and characteristic features.

Conclusions. The results of our review show that there are several diagnostic tools concordant culturally and linguistically with a given population for early ASD screening in primary care setting. They could be an effective method of accelerating the diagnostic process and starting personalized therapy faster. However, most tools have significant limitations – some are only available for research purposes, while others do not have scientific evidence to prove their effectiveness.

Introduction

Autism spectrum disorder (ASD) is a category of neurodevelopmental disorders characterized by challenges concerning social skills, speech development and behavior [1]. The cause of ASD is not known - it was suggested that the etiology includes many factors, including genetic, infectious or metabolic ones [2]. These disorders occur in all racial, ethnic and socioeconomic groups [3]. The prevalence is yet to be clearly defined; however, the World Health Organization (WHO) estimates that ASD occurs in 1 in 160 children worldwide [4]. However, this estimate varies considerably depending on the research method and country. For example, in Israel, it is 4.8%; in Iceland – 3.13%; in the United States – 1.7%; in Qatar – 1.14%; in Iran – 0.06% [5–10]. Thus, the percentage of individuals with ASD in the population depends primarily on diagnosis methods. The growing number of registered cases of ASD in recent years probably results

from a greater number of diagnosed adults and children than changes in the frequency of the autism spectrum phenotype in the population [11].

Diagnosis of ASD is a long-term and multi-stage process aimed at recognizing existing disorders and assessing a child's functioning on many levels. It begins with observing the child by parents, guardians, or other people who have contact with the child. It is also necessary to exclude other diseases that may cause symptoms similar to ASD. For this reason, consultations with other specialists (e.g., audiologists, laryngologists, geneticists) are necessary. The final stage is the definitive diagnosis by a team of specialists (psychiatrists, psychologists, special educators, or speech therapists) [12].

The role of family doctors and pediatricians in early diagnosis of ASD

Family doctors or pediatricians working in a primary care clinic most often observe a child during infancy and early childhood, especially as part of well-child care visits, qualifications for vaccinations, or visits due to common infectious diseases. This fact enables careful observation of the child's development and behavior in the critical period for diagnosing ASD, which means that the general practitioner (GP) may be first to notice the behavioral signs of disorders [13–15].

A desirable situation from the patients' point of view is the GP taking the role of a "gatekeeper," i.e., a person who notices the first "red flags" in the child's behavior, analyzes the concerns raised by parents, and decides about the need for further specialist consultations [16]. During the aforementioned visits, parents ask questions about the symptoms they notice and express concerns about their child's development [13]. Unfortunately, there are still frequent situations when doctors marginalize, minimize, or ignore the concerns raised by parents [17]. This may be due to organizational reasons related to primary health care structure (e.g., limited consultation time, excessive workload) [16, 18]. Moreover, identifying some ASD-specific features (e.g., sensory disorders) requires – apart from experience in this matter – devoting more time to patients than is generally provided for a visit in primary care clinics [19]. Another problem that hinders early diagnosis in primary care is the insufficient knowledge of doctors about ASD. A study conducted in 2020 showed that only 23% of primary care physicians (PCPs) had sufficient knowledge about ASD, and the percentage of such doctors was higher in countries with higher income [20]. For example, in Pakistan, only 44% of GPs knew the concept of autism, and only 42% of them had any further knowledge about it [21]. The driving force to improve the knowledge and skills of PCPs in the field of ASD may be the growing public awareness of the issue. Unfortunately, the spread of the term "autism" in society produced mixed results. On the one hand, greater awareness of the problem allowed many families to get help and additional financial resources; on the other hand, it also led to an uncontrolled public debate and spread of unfavorable stereotypes and untruths about ASD and its etiology [22–24]. A better method of spreading knowledge about ASD is special training for doctors by experts [25].

Possibilities of early detection of ASD

Identification of autism spectrum disorders is challenging in the early stages of life when changes in development are rapid and symptoms – often subtle [26]. However, early diagnosis is a necessary first step to implement effective therapy appropriate to the child's needs at a critical time of development – the younger the child at the time of ASD diagnosis, the better therapy results [27–29].

In order to increase the effectiveness of PCPs in the early diagnosis of ASD, numerous screening questionnaires have been developed, which their proponents claim to be some of the most beneficial health policy innovations ever created for children with ASD [30]. On the other hand, ASD screening is criticized in terms of cost-effectiveness or time constraints and the low psychometric properties of tests, especially in very young children [31–33]. However, there is evidence suggesting that including screening tools in routine medical appointments may result in earlier and more accurate identification of children who need further help than relying solely on clinical impressions, which is particularly important when care providers are less experienced in diagnosing ASD [34, 35]. Since the effectiveness of detecting ASD using various questionnaires (understood as the percentage of true positive results) increases with age, very early diagnosis of the youngest children is one of the major therapeutic problems. For such patients in whom screening is associated with tests of low psychometric properties, developmental follow-up is essential later in life. A solution to these problems may be developing novel and better diagnostic methods that take into account both the age and gender of the child [36].

Since 2006, the American Academy of Pediatrics (AAP) has recommended routine diagnosis of ASD at 18 and 24 months of age during well-child care visits [37]. Children who receive a positive screening result should be sent for further ASD evaluation to an early intervention center and referred to an audiologist to rule out hearing impairment, as recommended by the AAP [38, 39]. Over 14 years, these activities significantly increased the prevalence of ASD and made primary care facilities the main places of early diagnosis of ASD [40]. Following these recommendations resulted in more than 50% of American children undergoing screening for autism spectrum disorder [41–43]. In addition, the increasing availability of screening significantly lowered the age of ASD diagnosis in the US, with diagnosis before the age of 4 made in 71% of children (2018) compared to 58% in 2014 [40, 44].

In turn, the recommendations of the US Preventive Services Task Force indicate the lack of sufficient evidence in favor or disadvantage of performing ASD screening in children, for whom no concerns of ASD have been raised by their parents or a clinician [45].

Aim of the study

The main aim of this scoping review was to demonstrate available, culture-specific and language-adapted tools for the early screening of autism spectrum disorders, that can be used by healthcare professionals working in primary care. We were interested in gaining better insight into their psychometric properties and cultural adaptations, which is particularly important due to the social diversity of cultures [46]. Our final goal is to identify the most relevant tools for screening for ASD in primary care.

Materials And Methods

In this research we used the five-step approach described by Arksey and O'Malley to conduct a scoping review: 1) identifying the research question, 2) identifying relevant studies, 3) selecting the studies, 4) charting the data, 5) collating, summarizing and reporting the results [47]. The whole process was dynamic and iterative, with each step discussed with a group of investigators.

Identifying the research question

Our scoping review focused on answering the question: What are the suitable ASD screening questionnaires available that can be used in primary health care, and what are their characteristics? By suitability we mean a free (available in the public domain or after contact with the authors), short screening questionnaire, completed by a parent or clinician, characterized by good psychometric values and requiring no additional training in order to use it.

Identifying relevant studies

The primary search strategy was developed collaboratively by all authors. We conducted an online search using four different scientific databases containing articles concerning medical and psychological sciences (PubMed, EBSCO, Scopus, and Web of Science) to find publications related to the early diagnosis of children with ASD. We used Mendeley to collect and organize the references. The search began in March 2021 and included all publications written in English and released from January 1980 to May 2021.

The initial search results included a large number of studies related to developmental screening processes and provided guidance and recommendations for the use of screening tools (e.g. AAP guidelines) [37]. The results also included research describing the development and validation of tools, the adaptation of screening tools, and comparisons between individual instruments. Using previously published scientific research on ASD screening tools and our literature search, we compiled a list of tools used for this purpose [48, 49].

As names of screening tools were not mentioned in the title or keywords of many peer-reviewed papers, we also performed individual searches to identify them. Therefore, at each stage of the search for screening tools (step 1), an individual search (step 2) was performed using the name of each instrument indicated in the general search results. In addition, we adapted the search string to the thesaurus of three other databases. Finally, using a snowball approach, we added articles of the reference lists if they met the inclusion criteria mentioned below but were not listed in the initial search. The exact terms we used in the searches can be found in S1 File.

Unfortunately, not all the information sought by us was available in peer-reviewed scientific publications. Therefore, we collected information about screening instruments from several sources. For example, we checked test reviews and articles describing psychometric properties in peer-reviewed journals, manuals, technical papers, doctoral dissertations, and information from test publishers or distributors.

Inclusion criteria

1. Studies on tools intended for diagnosing children from 0 to 3 years of age;
2. Research describing the use of the tool published in English (or at least an abstract providing the necessary information);
3. Research on the tool intended for screening or rapid assessment, not a formal diagnosis of ASD.

Exclusion criteria

1. Studies on tools intended for formal diagnosis (for this reason, instruments such as the Autism Diagnostic Observation Schedule (ADOS) or Bayley Scales of Infant Development (BSID) were excluded from the study). We also excluded more complex tools beyond the competencies of family doctors, requiring additional training or completion of training authorizing to use them (e.g., Ages and Stages Questionnaire (ASQ), Social Responsiveness Scale (SRS-2), Achenbach System of Empirically Based Assessment (ASEBA), Parents' Evaluation of Developmental Status (PEDS), or Autism Spectrum Rating Scales (ASRS));
2. Studies on tools intended for screening children older than three years. For this reason, the publication omits, for example, the Social Communication Questionnaire (SCQ), which, according to the authors of the tool, is intended for screening of children over four years of age;
3. Research on diagnostic tools used in screening for other developmental disorders.

Information on screening tools was not always readily available; therefore, the decision to include a particular instrument was made based on the best current knowledge. After individual searches, some tools were excluded as they were replaced with a newer, improved version.

Selecting the studies

We imported all titles of our search into Rayyan software and deleted duplicates [50]. Reviewers in pairs (MSO and MBF, AK and MSe) read the titles and abstracts of the studies found following the search strategy to determine their eligibility. Then, studies were categorized as "include" or "exclude." In the event of contradictory information or disagreement, all the authors responsible for the publication made a final decision after a discussion. Finally, full texts of the selected studies were retrieved for a final review and distributed among the researchers in the same pairs. As before, authors jointly decided to include or exclude given publication for this scoping review in case of doubt.

Charting the data

Data from all studies included in the review were extracted and collected in an Excel spreadsheet to create an appropriate profile for each tool and determine its suitability for use in a primary care setting. The spreadsheet presents information about the purpose of the instrument, children age range, required time to complete the questionnaire, information whether an assessment report (e.g., filled in by a parent or guardian) or a direct assessment was used (e.g., observation of a child's behavior), and its psychometric and diagnostic properties. We were also interested in knowing whether any cultural changes were made in a given questionnaire adaptation. The same pairs of reviewers involved in the study selection extracted data from selected studies using an Excel sheet and discussed the

discrepancies. To calibrate our data extraction, MSo prepared a calibration exercise on five studies, which improved data extraction.

Collating, summarizing and reporting the results

After extracting the data, we created tool profiles to standardize the available information about their characteristics, properties, and application in primary care. Each tool that met the inclusion criteria for the study received its profile with data on the name, abbreviation, time of completing the questionnaire, and the person responsible for completing it. In addition, each adaptation of the questionnaire received its line on the spreadsheet for the country for which the validation was prepared, the language into which the text was translated, psychometric and diagnostic data (i.e., reliability, sensitivity, specificity, positive and negative predictive value), and the population in which the study was conducted (with an indication of the specific features of this population). Additionally, we marked in the spreadsheet whether a given version of the questionnaire is the original version and whether the adaptations were subject to linguistic and cultural changes. Figures were rounded to the second decimal point.

Results

The initial search yielded 330,225 titles, of which 227,371 were duplicates. After the first screening of titles and abstracts, we assessed 154 full text studies and finally identified 81 studies, which met inclusion criteria and underwent full data extraction. Three additional data sources were included outside of database searches, e.g. test manuals available on-line (see Fig. 1). All collected data are presented in Table 1.

Study characteristics

The studies described research from 37 countries; most studies originated from the US (N = 18), Australia (N = 5), and South Korea (N = 4). In addition, one article reported a study conducted in nine Arabic countries (Egypt, Kuwait, Jordan, Oman, Qatar, Saudi Arabia, Syria, Tunisia, and Lebanon), and one from the US conducted on a group of Nepalese refugees from Bhutan [91, 101]. The number of scientific papers published during the period under review was relatively stable, with an increase over the last five years.

Study objectives

The studies included in the review had varied purposes; however, a significant majority focused on determining the psychometric values of the tools. Reliability (defined as Cronbach's alpha) was provided in 46 of all studies (one study reported only the factor analysis of the instrument), sensitivity was assessed in 53 studies, specificity in 51 studies, positive predictive value (PPV) in 47, and negative predictive value (NPV) in 36 studies. Two studies aimed to determine the cut-off points for the study population for a given tool [76, 77]. One study aimed to demonstrate the need for further research on the cultural and linguistic adaptation of screening questionnaires and simplifying the wording used in them

[101]. Finally, one study was designed to test the stability of the cross-cultural measurement, and one aimed to identify possible difficulties related to translating the ASD screening questionnaires to adapt them to other languages and cultures [78, 82].

Study populations

The number of participants included in the studies differed significantly, ranging from 13 to 52,026 [101, 102]. 34 studies included more than 1,000 children, while six had more than 10,000 participants.

Children from the general population were included in 46 studies. In eight papers, the research was based only on a group of children at risk. One study was conducted in a group of typically developing children [68]. In the case of three publications, the characteristics of the studied population were not specified. The remaining publications concerned both children with a low and high risk of ASD. It is worth noting the different understanding of the term “high-risk children” in individual papers, as risk groups, for example, included siblings of children diagnosed with ASD, children already diagnosed with ASD or other developmental disorders, or suspected of developmental delay, etc.

Tools characteristics

In the course of the study, we were able to identify 26 different autism spectrum disorder screening tools that met our study criteria.

We would like to point out that while researching the information about tools, we found mixed data on the availability of the Checklist for Autism Spectrum Disorder (CASD) for professionals who are not psychologists or have not completed the appropriate training. Nevertheless, we decided to include CASD in this publication as a tool available to PCPs.

Original versions of questionnaires

The original versions of questionnaires come from 13 countries. Most of them (as much as 35%, N = 9) were created in the US. Only two questionnaires were developed in low- and low-middle-income countries (Uganda and Sri Lanka) [51, 112, 135]. An even greater disproportion could be observed in the languages in which the original versions of the tools are available. Of the 35 original language versions (some questionnaires such as CASD, JA-OBS, and PAAS were prepared in two languages, and INCLLEN-ASD even in nine), almost half (N = 17) were in English.

Number of language versions and cultural adaptations of ASD screening tools

Data from selected publications allowed us to create 75 profiles of different versions of the adaptations or original versions of ASD screening questionnaires. Most tools were prepared in one country in one language version. At least one questionnaire was tested in a total of 45 different countries. The largest number of various questionnaires was available in the US (11), Australia and South Korea (4 each), China, the Netherlands, and Turkey (3 each).

Some questionnaires in one study were translated into multiple languages simultaneously; however, at least one tool was available in 35 different languages. In some countries, the questionnaires were adapted to the local dialect (e.g., the Spanish versions of M-CHAT were adapted to Spanish, Mexican, Chilean, and Argentinian respondents) [86, 88, 100, 104]. Most of the questionnaires were available in English (N = 21), Spanish (N = 7), Chinese (N = 6), Dutch (N = 4) and Korean (N = 4).

At this point, it is worth mentioning that there are many translations of the questionnaires, such as M-CHAT or Q-CHAT, available on the websites of organizations involved in developing them. For example, the most popular M-CHAT is available in 73 versions, but most lack research published in international journals [136, 137]. The situation is similar with the Japanese and Spanish BITSEA versions [138].

Most language versions of the individual questionnaires were translated directly into the language of the surveyed population, sometimes with minor changes. However, for example, in the Argentinian version of the M-CHAT questionnaire, the dialect was changed to match better Spanish used in Argentina. Likewise, in the Taiwanese version of STAT, two items were changed to suit the Taiwanese population better [86, 130].

In addition, cultural changes were made in nine adaptations. For example, phonemes were adapted to the language, and the type of assessed play or the type of toy shown to children was changed to capture their interest.

Psychometric values

When searching for information on different versions of questionnaires, we focused primarily on reliability, sensitivity, specificity, PPV, and NPV. We made the decision not to include validity data in our review due to the considerable variation in the methodology used across studies (different types of validity measured by various means) or other psychometric values (such as positive or negative likelihood ratio) due to the small number of studies containing these data and the desire to simplify the table as much as possible to facilitate its use by practitioners.

Out of all 75 profiles, we were only able to complete 20 of them containing all the five values sought.

Reliability. Internal reliability of the test is a measure defining the consistency of items included in a given scale, i.e., it determines to what extent the items included in a given factor or scale are similar to each other or whether they test the same phenomenon. The most common measure of reliability is Cronbach's alpha (α) [139]. In the profiles we created, this measure ranges from 0.53 to 1.00. Using the rule of thumb and other different qualitative descriptors methods, 6 of the studies had excellent reliability ($\alpha > 0.93$), 2 – strong (0.91–0.93), 12 – reliable (0.84–0.90), 14 – relatively high (0.70–0.83), and 13 had reliability below 0.70 [140].

Sensitivity. Test sensitivity is the ratio of the true positives to the sum of the true positives and the false negatives. A sensitivity of 100% would mean that all individuals with existing disorders would be diagnosed. Values of reported sensitivity in 53 profiles varied from 0.18 to 1.00. Most of the tests (N = 42)

scored above 0.70. There is a significant discrepancy between the sensitivity values between linguistic adaptations of the same type of questionnaire (e.g., M-CHAT used in the US and Sri Lanka), resulting potentially from an inadequate cultural adaptation of the tool [106, 110].

Specificity. Test specificity is the ratio of the true negatives to the sum of the true negatives and false positives. A specificity of 100% would mean that all healthy individuals in the test performed would be marked as healthy. Specificity was calculated for 51 of the above-mentioned versions of questionnaires and ranged from 0.51 to 1.00. In 37, specificity exceeded 0.80.

Positive predicting value (PPV). PPV is equal to the proportion of true positives out of all positives and determines the probability that a positive test result is accurate. PPV of the questionnaires in the studies included in the review ranged from 0.01 to 1.00, showing a significant variety. Noteworthy is the considerable increase in PPV after the follow-up interview was used in the American version of M-CHAT, showing an increase from 0.11 to 0.65 [110].

Negative predicting value (NPV). NPV is the proportion of true negatives out of all negatives; it determines the probability that a negative test result is accurate. All versions of questionnaires, except one (DBC-ES with NPV = 0.48), for which NPV was calculated, had NPV greater than 0.73 [73].

Person completing the questionnaire

ASD screening questionnaires can generally be divided into questionnaires filled in by people who have constant contact with the child (parents or guardians) or independent observers – specialists (e.g., doctors, nurses, psychologists, etc.). Most (15 out of 26) tools were intended to be filled by parents, and specialists only dealt with possible doubts arising while filling in the questionnaire and calculated the result of the test. These also tools underwent cultural adaptation much more often than those in which a specialist assessed the child. Some instruments were by definition predisposed to a given professional group, e.g., the assessment of a child's development using the JA-OBS test is performed by nurses [84].

Time of completing the questionnaire

Most of the questionnaires listed above should not take more than 10–20 minutes for parents or specialists to complete, and some only take 5 minutes. For example, according to the authors, the shortened version of Q-CHAT (Q-CHAT-10) takes less time than 5 minutes [121]. On the other hand, BeDevel can take over 40 minutes to complete, and INCLen-ASD takes 45–60 minutes [56, 83].

Discussion

Our research revealed many tools for early ASD screening that can be employed in primary care (26 different instruments in 75 adaptations). An ideal tool for ASD screening seems to be a free and short instrument with items suitable for assessing development, with good psychometric properties, corresponding to the entire studied population, using plain language (low-reading level), easy to assess by people with no experience in psychometrics (easily score-able), providing simple and clear guidance

on what to do after screening [141]. Unfortunately, it is unclear which existing tools are best suited for this, so further development of both instruments and research into their use is necessary. Furthermore, there is a possibility that it will be appropriate to create an entirely new tool, which will be much more effective than the existing ones. The problem is further exacerbated by the small number of meta-analyses on the effectiveness of given screening tools. Another issue is that evaluating the usefulness of some of the questionnaires mentioned above is based on studies conducted a long time ago.

Due to the large sample size and high psychometric and diagnostic values, APSI, BITSEA, CESDD, CSBS-DP, M-CHAT, SACS, and STAT deserve recognition among the tools mentioned above. Furthermore, among all questionnaires included in the study, CSBS-DP, M-CHAT, and STAT are recommended by the Centers for Disease Control and Prevention (CDC) for ASD screening in the United States [142]. An additional positive in favor of M-CHAT is the multitude of language versions that were at least partially validated; the second such questionnaire is Q-CHAT.

From the perspective of primary care workers, it is also important to reduce the occupational encumbrance of implementing another examination tool which is the responsibility of the PCPs. Hence, it seems that it would be favorable for PCPs to implement screening questionnaires filled out by a parent. On the other hand, questionnaires in which a neutral observer assesses the child are more effective in detecting early symptoms of autism spectrum disorders.

Still, the main problem for PCPs will be choosing the right tool to carry out ASD screening. Positive experiences from the United States, where a mass ASD screening system was implemented successfully, indicate the suitability of using ASD screening tests in primary health care [40, 44]. Unfortunately, experiences from the US cannot be transferred directly to other countries. Furthermore, it is crucial for early diagnosis of ASD to have tools that respond to cultural and linguistic differences (as well as the local perception of “disability”). Hence, the use of mismatched tools may be inappropriate [46]. For example, in Jamaica, the percentage of parents reporting that their child shows developmental delays compared to peers is significantly higher than in Bangladesh or Pakistan [143]. The global application of ASD screening, especially in low- and middle-income countries (LMICs), is associated with many problems because most existing tools were developed in North America or Europe, but they are used – often without any significant modifications – in countries whose cultures differ significantly from those in which they were created. In particular, our study shows that there is a lack of tools to identify children with ASD in Africa and other LMICs [144, 145].

Another difficulty is the availability of some of the tools. A large part of the instruments that met the study criteria is available only for scientific use. And even if access to them is free, it requires contact with researchers and the authors’ consent for further use.

The lack of available screening tests for individual populations, incomplete validation, or limited availability is not the only difficulty in popularizing early diagnosis of ASD. Screening tests have limited sensitivity - some of the children who received negative screening will receive in subsequent years of their life diagnosis of ASD [146]. Hence, it is not only necessary to pay attention to the dissemination of

screening but also to remember the necessity of further continuous monitoring (follow-up) of children's development [146]. The situation is further exacerbated by the fact that there are no readily available (e.g., in the public domain) rapid tests for older children (aged 30–60 months) as is the case with other psychiatric disorders, e.g., Vanderbilt ADHD Assessment Scales for attention-deficit hyperactivity disorder or Screen for Child Anxiety Related Disorders (SCARED) for anxiety disorders. This makes it necessary to decide whether the child should be referred for further tests based solely on the experience of the primary care worker, which may delay the diagnostic process.

This is not the only reason for delayed ASD diagnosis in children. The example of the United States demonstrates that causes for delay may be due to imperfections of the public health system and low predictive values of the tests (especially in children scoring close to the cut-off limits). Examples of such restrictions include the following:

1. not all children receive healthcare as infants,
2. not all children who are receiving healthcare are screened - only 8-28% of pediatricians in the United States use ASD screening tools in their daily practice [147, 148],
3. not all screened children undergo additional consultations in case of a positive result [42, 43] – only 31% of children with a positive screening test were referred for further diagnosis, 20% to an early intervention center and 36% to an audiologist [149]; these values are slightly higher in another study [42].

The above data show that even with the widespread of the idea of screening, it does not allow for a complete diagnosis of all children with ASD, which, in this case, results mainly from neglect of the diagnostic process on the part of health care workers.

From an ethical point of view, it should be noted that lowering the age at which the diagnostic process begins in the population will result in an increased number of “false positive” cases, which entails a lot of stress experienced by the families of children that could be difficult to counteract in primary health care. Another problem is that it implies the rising cost of additional evaluation processes in children developing correctly.

The most controversial issue regarding universal ASD screening in children is the cost-effectiveness of ASD screening, primarily due to the moderate accuracy of current tools and the low prevalence of the disorder [45]. Attempts to estimate the cost-effectiveness of ASD screening indicate that universal screening may not be financially sound mainly due to delays in further diagnostic and therapeutic steps [33]. Eliminating the waiting time for further consultations with simulation models showed that the initial high cost incurred for screening might be offset by future savings resulting from improved functioning of ASD patients in society. However, the same analyses conducted in high-risk children showed the cost-effectiveness of screening. Nevertheless, the significant benefits of early intervention justify attempts to further refine this strategy for the early detection of autism spectrum disorders [150].

Limitations of the study

There are several limitations to this review. This study includes only scientific publications whose full text was in English or had an abstract containing most of the necessary data to create a profile for the tool. Because the goal of researchers studying early diagnosis of ASD is the implementation of the instruments in a given country, some existing research may have been excluded due to the publication of results of the validation process in languages other than English in local peer-reviewed journals.

The review was carried out mainly by using search string for publications in four scientific databases, potentially limiting the results.

It should also be noted that researchers carried out the measurements of psychometric and diagnostic properties of various tests in different ways, making it impossible to compare their parameters without taking into account the methodological details contained in the source texts.

Conclusions

The results of our review show that there are several diagnostic tools for early ASD screening in a primary care setting for which the full validation process was carried out and showed high psychometric and diagnostic values in this research. These tools could effectively accelerate the diagnostic process and lead to a faster start of personalized therapy. As some examples show, they could also become the basis for preparing almost equally effective adaptations of screening tests for different populations, especially after introducing cultural and linguistic modifications.

Unfortunately, for a large part of the tools, no changes other than accurate translation were made to fit the questionnaire to the characteristics of the particular population. Furthermore, only partial validation studies were carried out in many cases, which means that using them in everyday practice may be ineffective. Finally, the more culturally different two populations are, the more a tool designed for one will be less effective for the other.

Therefore, it appears necessary to continue research on adaptations of existing ASD screening methods and attempt to improve them and constantly increase the knowledge of health care professionals about ASD, improve the follow-up process, and further evaluate the cost-effectiveness of the ASD screening process.

Practical implications

This review highlights the available options for early diagnosis of ASD in primary care from a global perspective, indicating the importance of psychometric and diagnostic values in choosing the most suitable tool for everyday practice.

Declarations

Ethics approval and consent to participate

Not applicable.

Consent for publication

Not applicable.

Availability of data and materials

All data generated or analysed during this study are included in this published article and its supplementary information files (S1 File).

Competing interests

The authors of this publication are members of a research project, preparing the Polish version of the CSBS-DP questionnaire. None of the authors report receiving compensation from any author of the original version of the questionnaire.

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Authors' contributions

MSo and MMBF were responsible for conceptualization of the study and original draft preparation. MSo was responsible for data curation. All authors were responsible for formal data analysis, investigation and methodology. MMBF and MSe supervised research. All authors read and approved the final manuscript.

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Tables

Due to technical limitations, table 1 is only available as a download in the Supplemental Files section.

Figures

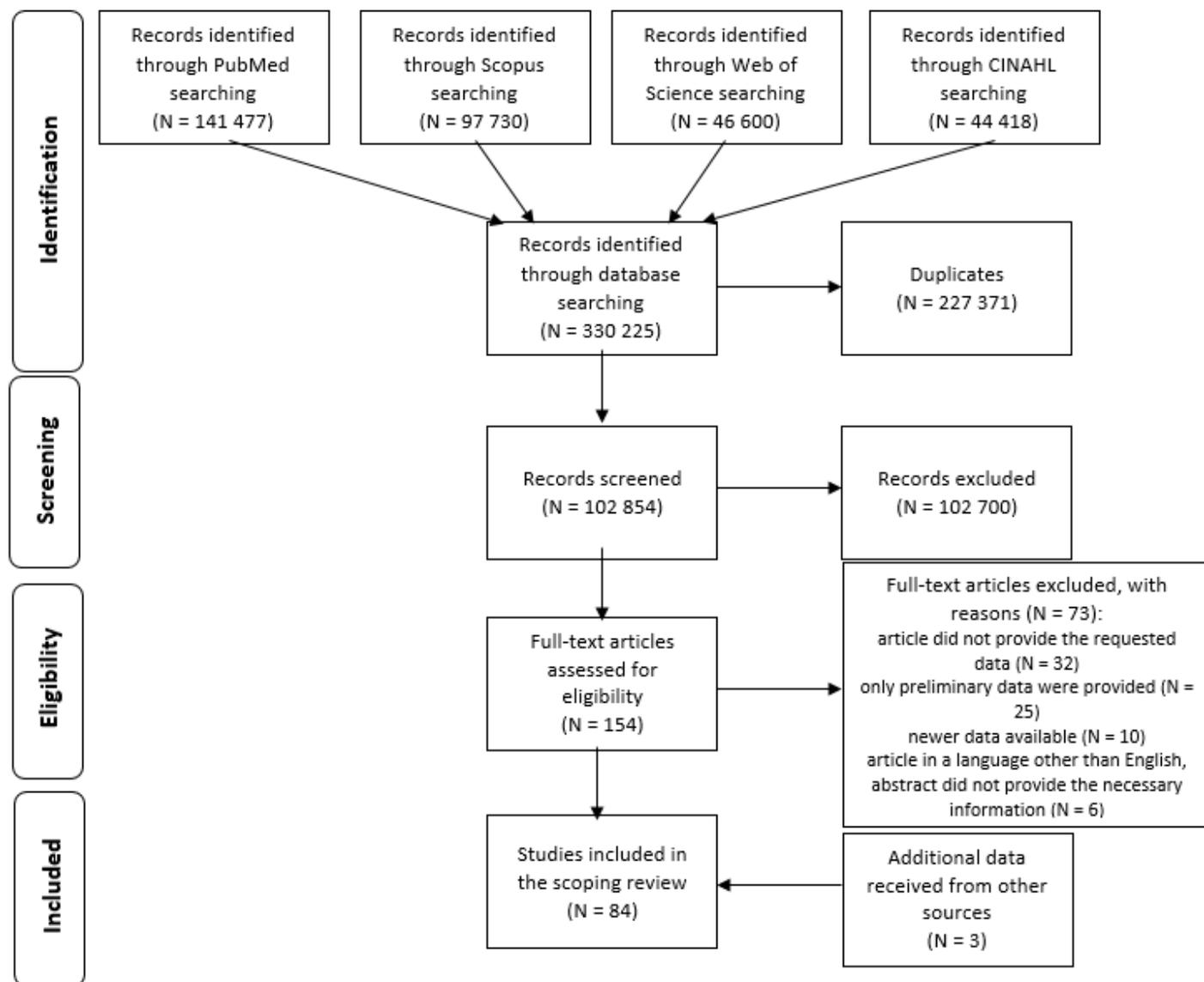


Figure 1

Prisma flow diagram.

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