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A Systematic Review of Instruments for Early Detection of Autism Spectrum Disorders

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Abstract

Only a small percentage of children with Autism Spectrum Disorders are diagnosed before they are three years old, although earlier detection and intervention would reduce the disabilities associated with the disorder. In addition, as children get older, treatments are more costly and difficult and the results less satisfactory. Considering the importance of detecting autism early, the objective of this study is to identify the instruments that can be used to detect signs of autism before children are 2 years old, and which of these instruments have been validated in the Spanish population. By searching in several research databases, we compared the existing instruments and their main characteristics. We found that some instruments can be used to assess a possible autism spectrum disorder before children are 2 years old, with acceptable sensitivity, specificity and reliability indexes. However, only a few instruments have been validated for the Spanish population, some of which have not been specifically designed for early detection. For this reason, a tool needs to be developed to detect the warning signs of autism spectrum disorders before the age of 2 which can be applied as part of the protocol for pediatric check-ups.
Key words: autism spectrum disorders, early detection, systematic review.

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Novelty and Significance

What is already known about the topic?

- The incidence of Autism Spectrum Disorders has increased in the last two decades and only a small percentage of children are diagnosed before age three.
- Several studies have identified important features in development at 9 months of life that are not present in children who are later diagnosed with Autism Spectrum Disorders.

What this paper adds?

- We found nine instruments that can be used to diagnose Autism Spectrum Disorders before two years of age and have acceptable sensitivity, specificity and reliability indexes. There are few validated instruments for Spanish population, although the existing ones are the most relevant in this field.
- It is necessary to develop tools for detecting warning signs of autism spectrum disorders that can be applied within the protocol of pediatric check-ups.

Autistic spectrum disorders (ASD) are neurodevelopmental disorders characterized by persistent deficits in social interaction and communication, manifested by the presence of qualitative impairment in verbal and nonverbal communication with a social purpose, deficiencies in social-emotional reciprocity, and an inability to develop, maintain and understand relationships with peers. Moreover, the existence of restricted and repetitive

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patterns of activities, behaviors or interests that may manifest as motor and/or verbal stereotypes, excessive inflexibility in routines, restricted interests and activities or unusual sensory behaviors have also been detected (American Psychiatric Association, 2013).

The literature shows that the incidence of ASD has increased in the last two decades (Lyall *et alii*, 2017; Fortea, Escandell, & Castro, 2013). The Center for Disease Control and Prevention in the United States estimates that 1 in 110 children aged 8 years old and younger have an ASD diagnosis (Dixon, Granpeesheh, Tarbox, & Smith, 2011). Baio (2012) and Mahoney, Minter, Burch, and Stapel-Wax (2013), cite a higher prevalence of 1 in 88 in the United States. In Spain, a recent study obtained a prevalence of 0.61% ASD in the Canary Islands (Fortea *et alii*, 2013). Two indisputable facts influence this increase: the conceptual and definitional change, and the significant improvement in tools and evaluation processes. In fact, this implies that individuals with diverse profiles, difficulties and skills who are not covered by the classical definition are placed in the same category (Grupo de Estudios de los Trastornos del Espectro Autista, 2004).

In terms of screening, the children that are most affected are those with a delayed diagnosis, many of them only being diagnosed after visiting various professionals. Only a small percentage of children are diagnosed before age three, and in many cases it is their teacher who identifies difficulties when they start school. One study cited by Klin, Klaiman, and Jones (2015) of surveillance records from the USA Centers for Disease, Control and Prevention (Wiggins, Baio, & Rice, 2006), notes that even among children who are initially assessed for a possible ASD at an average age of four years old, professionals do not diagnose them before they are, on average, at least five years old. Some features of social, communicative and symbolic development in the first two years of the lives of these children are still not sufficiently taken into account by professionals specializing in care during early childhood (Ferre, Palanca, & Crespo, 2008).

This diagnostic delay contradicts the view expressed by parents who observe warning signs manifested in the development of their children at an earlier age (Zablotsky *et alii*, 2017): 30% of parents of children with ASD suspected developmental problems before their child's first birthday; 50% at 18 months; and 80% at 2 years (Chawarska et alii, 2007; Wetherby, Brosnan-Maddox, Peace, & Newton, 2008). Nevertheless, the median age of diagnosis in the United States continues to be 5.5 years old (Shattuck *et alii*, 2009). In the Canary Islands, Fortea *et alii* (2013) found that in 79% of cases it was the family itself who first became suspicious that something was wrong with the child's development. Sixty-nine percent of children were first diagnosed before their third birthday, and 32% before their second. The diagnostic delay was around 16 months. These authors conclude that most parents of children with ASD become aware of changes in their children's development at around 18 months, and insist that early detection improves prognosis. Similarly, Chakrabarti and Fombonne, (2005) using ADI-R with parents of autistic children, concluded that the average age at which parents recognized that there was a problem was 23.4 months, the average delay between first recognition of the problem and seeking professional help was 4 months, and diagnosis was received at 32 months.

A precise definition of the particular development of individuals with ASD, in terms of these characteristics in the first 24 months of life, enables us to anticipate the possible presence of difficulties associated with the spectrum before the child is 2 years old. This, in certain cases, could advance diagnosis, which is the starting point for planning and implementing early intervention programs that encourage the development of the functions that are limited. Early stimulation of children with signs of risk has

been shown to significantly reduce the disability associated with the disorder (Canal *et alii*, 2015). With the appropriate stimulation, the neurological plasticity at this age provides children with better personal tools for their social environment in the future. It is more difficult to treat the autistic disorder after three years old, when it has become internalized. The cost of treatment also increases, and the results are less satisfactory the older the child is (Canal *et alii*, 2015).

The study by Veness *et alii* (2012) is one of the few studies we have found that detected signs of ASD before 2 years old. The authors found no difference at 8 months, but began to find differences at 12-24 months in children probably suffering from ASD, mainly indicated by their intentional communication. However, it is important to note that the study has little statistical power due to the small sample it uses. Other studies have identified important features in development at 9 months old that are not present in children who are later diagnosed with ASD (Libertus, Sheperd, Ross, & Landa, 2014; Ozonoff *et alii*, 2010; Palomo, 2012; Rivière, 2000). Bölte *et alii* (2013) and Ozonoff *et alii* (2015) emphasize the need for longitudinal studies beginning early to detect autism, and argue that it is possible to diagnose the condition before 2 years old, when the problem is usually identified. This could be an added issue, since intervention time is lost, thus leading to a decline in the child's quality of life.

According to Matson, Rieske, and Tureck (2011), early detection and diagnosis is the keystone of early treatment. Scales that are more specific for very young children are therefore required. Some tests can identify warning signs at the evolutionary level before the first year of life, such as the Haizea-Llevant Scale (Fernández, Fuentes, & Rueda, 1991), which can be applied in routine visits in the core areas of health care until the age of 4. An application for detecting developmental disorders early called the Early Detection System of Developmental Disorders (EDSDD) has recently been developed. This application can be used by parents to detect any problems in the psychomotor development of children aged from 3 to 36 months (Alcantud, Alonso, & Rico, 2015). Although these tests allow the identification of developmental problems at earlier stages, they are not specifically designed to assess ASD.

Considering the importance of detecting autism early, the objective of this study is to identify the assessment instruments for detecting signs of autism before the child is 2 years old. We also look at which of these instruments are currently validated in the Spanish population. Specifically, we identify and compare the existing instruments and their main characteristics (number of items, psychometric properties, number of subjects used to develop the instrument, etc.). This comparison of the different instruments is of particular interest for doctors and researchers working in this field, helping them to decide which is the most relevant instrument according to their objectives and the characteristics of each test.

Method

Search strategy

We searched articles in indexed journals in Web of Science, Scopus, PubMed, PsycNET, ERIC and Google Scholar from their beginning until January 2018. The following keywords were used in the search engines: Autism* AND (child* OR infant* OR small child*) AND early detection* AND (instrument* OR questionnaire* OR list* OR assessment* OR scale* OR report* OR record* OR test* OR measurement* OR

interview*). All the articles were empirical studies or reviews in English or Spanish. We did not filter by author, date, journal type, country, subject area, funding source, journal title, or editor. We added a filter for the document type by selecting "article" or "journal article" depending on the option allowed by the database ("article" in Web of Science and Scopus, and "journal article" in PubMed, PsycNET and ERIC). We also added a filter for the language by selecting English and Spanish in the databases that had the option (Web of Science, Scopus and PubMed). Finally, the PubMed and PsycNET databases had the option to filter by age group, so we selected "infancy" (from birth to 23 months).

Selection criteria

The articles were selected using the following criteria: they had to have been published in indexed journals; they had to discuss instruments used to detect possible autism before age 2, with particular focus on their psychometric properties; the instruments mentioned in each article had to focus only on autism and assess all areas that are usually affected as a result of this disorder. We did not include articles that were adaptations of instruments in a particular country, articles that discuss a combination of several instruments, articles that assess disorders other than autism, or articles that only assess a specific disorder within ASD (e.g. Asperger).

We used the PRISMA guidelines (Urrutia & Bonfill, 2013) obtaining a total of 760 references. Then we eliminated those references that were repeated in several databases, which left 421 references. After this, we added 2 articles identified in other sources. Of the total of 423 articles, 385 were excluded because they were irrelevant articles for the objective of the current study, or they did not fulfill some of the inclusion criteria specified above. More specifically, these articles were focused on instruments that do not allow an early detection of possible autism before age 2, they assess disorders other than autism, or they only assess a specific disorder within ASD (e.g. Asperger). Moreover, other articles were focused on adaptations of preexisting questionnaires in new countries. Of the remaining 38 articles, we excluded 29 because they were focused on instruments that do not evaluate all areas that are usually affected in this disorder. Finally, we obtained a final number of 9 articles that fulfill all the inclusion criteria, which were included in this review. Each of these 9 articles discusses a different instrument that can be used to screen for possible autism before 2 years old (with the exception of ADI-R and ADOS for which a new algorithm was developed so that they could be used in toddlers) and describes its psychometric properties. Other articles focused on the same instruments, but we selected only those that included the psychometric properties. The aim of this review was to find instruments used to detect possible autism at an early age. For this reason, the articles were selected so that these instruments and their psychometric properties could be identified. In other words, the ultimate aim of the study was not simply to find articles. The process can be observed in Figure 1.

Results

The search with the keywords and inclusion/exclusion criteria specified above, revealed a total of nine screening instruments for detecting possible autism before 2 years old (see Table 1). All these studies included the psychometric properties of the instruments.

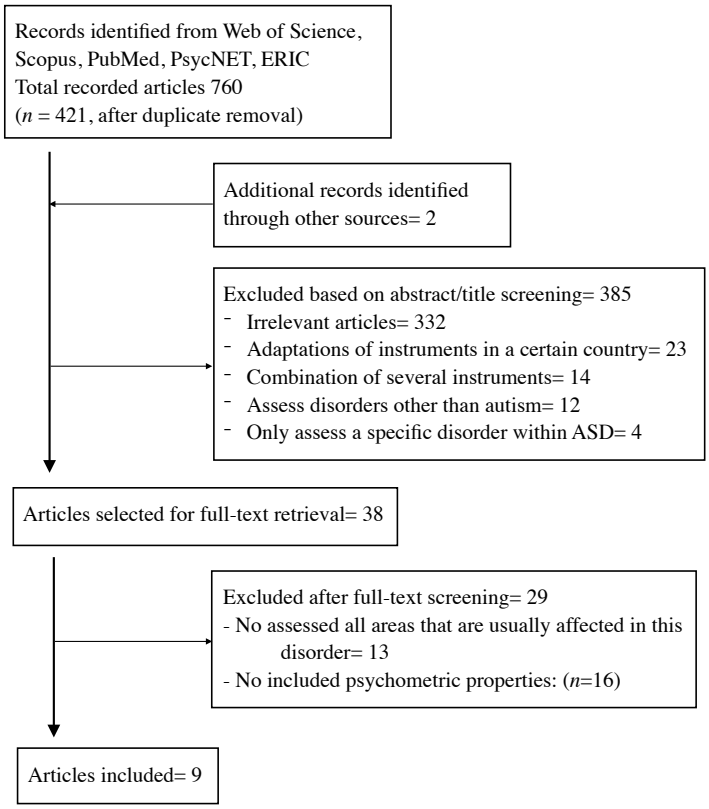

Figure 1. Flow diagram

The aim of all the articles found was to develop a screening tool to detect autism at early ages with acceptable psychometric properties. The only exceptions were ADI-R and ADOS, which were designed to develop new algorithms for toddlers.

To develop the screening instruments, all these studies used samples of children who were either developing normally or who had been diagnosed with autism. Each study had a different sample size, the smallest being the AOSI with 34 subjects and the highest being CESDD with 6808 subjects. The basic characteristics analyzed in each instrument were the children's age range, number of items, administration method, sensitivity, specificity, concurrent validity and reliability (see Table 2).

Some instruments used a small sample compared to their counterparts, such as the Toddler Autism Screening Questionnaire (TASQ), Autism Observation Scale for Infants (AOSI), and Autism Detection in Early Childhood (ADEC). It can also be seen that the strong point of some tests is their sensitivity, such as ADEC, BISCUIT and ESAT, while others stand out due to their degree of specificity, such as M-CHAT and CESDD. TASQ has high and balanced indexes, as it obtains 1 in sensitivity and .96 for specificity, similarly to the ADOS-Toddler Module, which obtains .91 for sensitivity and .94 for specificity. However, we do not have information on the sensitivity and specificity of

Table 1. Summary of the articles reviewed.

Articles	Instruments	Participants
Nah *et alii* (2014)	ADEC	70
Robins *et alii* (2001)	M-CHAT	1293
Kim & Lord (2012)	ADI-R algorithms for toddlers	829
Luyster *et alii* (2009)	ADOS-Toddler Module	182
Matson *et alii* (2009)	BISCUIT	276
Bryson *et alii* 2008)	AOSI	34
Tsai, Soong & Lotus (2012)	TASQ	77
Dereu *et alii* (2010)	CESDD	6808
Swinkels *et alii* (2006)	ESAT	478

Notes: ADEC= Autism Detection in Early Childhood; M-CHAT= The Modified Checklist for Autism in Toddlers= ADI-R: Autism Diagnostic Interview-Revised; ADOS= Autism Diagnostic Observation Schedule; BISCUIT= the Baby and Infant Screen for Children with Autism Traits; AOSI= Autism Observation Scale for Infants= TASQ: Toddler Autism Screening Questionnaire; CESDD= Checklist for Early Signs of Developmental Disorders; ESAT= Early Screening of Autistic Traits.

Table 2. Instruments for the early detection of signs of autism and their psychometric properties.

Name	Age range (months)	Items	Administration	Sensitivity	Specificity	Concurrent validity	Reliability
ADEC	12-36	16	With little training 10 minutes	1.0	.74-.90	ADOS (Lord *et alii*, 2000), ADI-R (Rutter, Le Couteur & Lord, 2003), DSM-IV-TR (APA, 1994).	α=.91
M-CHAT	18-30	23	Pediatricians and family practitioners with caution 5-10 minutes	.87	.99	Bayley (Bayley, 1993), VABS (Sparrow, Balla & Cicchetti, 1984), CSBS (Wetherby & Prizant, 1993), CARS (Schopler *et alii* 1980).	α=.85
ADI-R algorithms for toddlers	12-47	13-20	Professional 2-3 hours	.85	.70	ADI-R (Rutter, Le Couteur & Lord, 2003), ADOS-T (Luyster *et alii*, 2009), Module 1 or 2 of the ADOS (Lord *et alii*, 1999) PL-ADOS; (DiLavore, Lord, & Rutter, 1995), observation by two clinicians with DSM-IV criteria (APA, 1994).	α= .9 (social affect) α= .73 (restricted and repetitive behaviors) α= .87 (imitation, gestures and play)
ADOS-Toddler Module	12-30	41	Professional 30-45 minutes	.91	.94	ADI-R-algorithms for toddlers (Kim & Lord, 2012), ADOS (Lord *et alii*, 2000), direct observation by clinical psychologist.	ICC= .86 (algorithm total score)
BISCUIT	17-37	71	Professionals. 20-30 minutes	.93	.86	Clinical diagnosis.	α=.97 (diagnostic)
AOSI	6-18	18	Professionals 20 minutes	Not provided	Not provided	DSM-IV-TR, (APA, 1994), ADOS (Lord *et alii*, 2000), ADI-R (Rutter, Le Couteur & Lord, 2003), and judgment of experienced clinicians.	Test-retest: .68 and .61.
TASQ	18-24	15	Parental self-report.	.1	.96	Clinical diagnosis.	*k*=. 40-.92
CESDD	0-36	25	Child care workers with minimal training.	.80	.94	MSEL (Mullen, 1995), ADOS-G (Lord *et alii*, 2000).	Not provided.
ESAT	About 14 months	14	Health practitioners. 10-15 minutes	.90	.81	Clinical diagnosis	ICC= .81

Notes: ADEC= Autism Detection in Early Childhood; ADOS= Autism Diagnostic Observation Schedule; ADI-R= Autism Diagnostic Interview-Revised; M-CHAT= Modified Checklist for Autism in Toddlers; BAYLEY= Bayley Scales of Infant Development, second edition; VABS= Vineland Adaptive Behavior Scales; CSBS= Communication and Symbolic Behavior Scale; CARS= Childhood Autism Rating Scale; PL-ADOS= Pre-Linguistic ADOS; BISCUIT= Baby and Infant Screen for Children with Autism Traits; AOSI= Autism Observation Scale for Infants= TASQ: Toddler autism screening questionnaire; CESDD= Checklist for Early Signs of Developmental Disorders; MSEL: Mullen Scales of Early Learning; ADOS-G= Autism Diagnostic Observation Schedule-Generic; ESAT: Early Screening of Autistic Traits; α= Cronbach's alpha; *k*: Kappa; ICC= Intraclass correlation coefficient.

AOSI, which was not included in this article. In terms of reliability, BISCUIT, in its diagnostic part, is the instrument with the highest index, with a Cronbach's alpha of .97, followed by ADEC with an alpha of .91.

Many of the instruments use the clinical diagnosis of the child as concurrent validity, while others usually use the ADI-R, ADOS, and DSM-IV-TR criteria. Only CESDD and M-CHAT use other tools for concurrent validation.

The instruments reviewed here do not usually have many items, except BISCUIT, which has 71 items in the diagnostic part. The test that has the least items is ESAT, with 14 items. Looking at the different administration methods, we see that most of the instruments can be applied by a person with a minimum of training.

Special mention should be made of the traditional diagnostic tools for autism, ADI-R and ADOS, which have created a new algorithm for young children. Originally, ADI-R and ADOS were tools for testing autism in children over the age of 2 years old. However, a new algorithm has been developed for both instruments so that younger children can be assessed. More specifically, the new algorithm can be used with ADI-R to test children aged between 12 and 47 months, and with ADOS to test children between 12 and 30 months. But it should be taken into account that these instruments were not originally designed for these younger ages, so they do not contain specific content and items adjusted to these ages. We have included these two tools in this review due to their importance in specifically assessing autism, and because this algorithmic adaptation can be used to assess children younger than 2 years old.

Of the tests included in this review, only ADI-R (Rutter *et alii*, 2006), ADOS-Toddler Module (Esler *et alii*, 2015), and M-CHAT (Grupo Estudio MCHAT España, 2007) have a Spanish adaptation, for population from Spain. ADEC (Hedley, Young, Angélica, Gallegos, & Marcín Salazar, 2010) has a Spanish version adapted to the Mexican population.

Discussion

In this study, we reviewed the instruments that can be used to assess a possible autism spectrum disorder before 2 years of age. At present, several assessment instruments can be used for this purpose and have acceptable sensitivity, specificity and reliability indexes. Of the nine instruments reviewed, only two can be applied before 12 months old: the AOSI between 6 and 18 months, although it has the drawback of not having sensitivity or specificity indexes and of having been administered to a very small sample (n= 34), and the CESDD, which can be applied between 0 and 36 months old and has good specificity. We are surprised that we only managed to find these references even though a considerable number of publications show that many parents suspect problems in the development of their children before age 2, or even before the first year (Matson, Rieske, & Tureck, 2011), and other studies have identified important developmental features at 9 months old that do not appear in children with ASD (Libertus, Sheperd, Ross, & Landa, 2014; Ozonoff *et alii*, 2010; Palomo, 2012; Rivière, 2000; Veness *et alii*, 2012). We have also found the ADOS-Toddler Diagnostic Test (Lord *et alii*, 2000), which can be used between 12 and 30 months old, and the diagnostic interview ADI-R (Rutter *et alii*, 2003) which uses new algorithms to detect autism in children as young as 12 months old.

Almost all the instruments designed can be applied at around 18 months old, when the autistic regression tends to appear. Autistic regression refers to children who are developing normally but who are affected by an ASD and suddenly lose their developmental skills, especially language and social engagement. Of the instruments available, M-CHAT and TASQ have high sensitivity and specificity and have the

advantage that they do not have too many items so they are easy to administer. On the other hand, although BISCUIT has high diagnostic reliability, it has many items (71), which makes it less effective for use in the primary care of children, because it takes a long time to administer and requires specific training.

Nevertheless, most of these instruments require larger samples of autistic children, children with other development disorders and normative children, if they are to be shown to have high sensitivity and specificity. Taking into account that much of the research related to the aetiology of autism focuses on genetic and epigenetic factors, better instruments and diagnostic criteria are needed if ASD features are to be detected and longitudinal studies implemented before the age of 2 (Bölte *et alii*, 2013; Ozonoff *et alii*, 2015).

It should also be noted that few instruments have been validated for the Spanish population, although the ones that have been validated are the leading instruments in this field today: ADI-R (Rutter *et alii*, 2006), ADOS-Toddler Module (Esler *et alii*, 2015), and M-CHAT (Grupo Estudio MCHAT España, 2007). However, we believe that more tools should be validated in our country or new ones should be created in Spanish that can detect warning signs before 2 years of age.

In any case, it is positive that more and more instruments are being developed for the earlier detection of autism, and that these instruments contain fewer items. This means that they can be used in pediatric care contexts in which professionals have little time, so that children can be rapidly referred to specialized centers for further and more in-depth assessment. Consequently children with autism can be diagnosed earlier and the necessary protocols and interventions activate as soon as possible to improve their quality of life (Canal *et alii*, 2015). Even so, these early diagnoses should always be confirmed in the following years. In fact, the results of an assessment in children younger than 24 months old should specify that there is a certain degree of concern about the presence of warning signs of autism, avoiding a definitive diagnostic. It is necessary to develop a tool for detecting warning signs of autism spectrum disorders that can be applied within the protocol of pediatric check-ups. Therefore, in the regular medical checkups made during the first months of a baby's life this type of sensitive screening questionnaire could be administered at each periodical checkup.

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