

## General

# Autism Spectrum Disorder: recommended psychodiagnostic tools for early diagnosis

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### BACKGROUND

According to the DSM-5, Autism Spectrum Disorder (ASD) is a neurodevelopmental disorder characterized by persistent deficits in social communication and social interaction across multiple contexts and restricted, repetitive patterns of behaviour, interests, or activities. Autism is known as a “spectrum” disorder because there is wide variation in people’s experience of symptoms which cause clinically significant impairment in important areas of functioning.<sup>1</sup>

The prevalence is still unclear; however, the World Health Organization estimates that ASD occurs in 1 in 100 children worldwide.<sup>2</sup> Actually, this data could represent an underestimation, because most children are not diagnosed before the age of 3, although symptoms can appear at 12 to 18 months already.<sup>3</sup>

Moreover, considering the heterogeneity of this disorder and the lack of objective tools, the diagnosis of ASD is a long-term, multi-step process that aims to assess the child’s functioning on different levels. It begins with observation by parents or others who have contact with the child.<sup>4</sup> Considering this scenario, early diagnosis is necessary to implement an effective therapy tailored to the child’s needs at a critical time in development. Appropriate early treatment can reduce children’s symptoms and improve their overall development and quality of life, allowing them to gain social skills and the ability to act better in social situations, in order to achieve more autonomy in later life.<sup>5</sup> Moreover, in a preventive perspective, it is also essential to take into account the quality of life of siblings<sup>6</sup> and parents.<sup>7</sup>

It is important, hence, to understand what the early symptoms are. According to Tsang et al.<sup>3</sup> early warning signs for ASD are:

- at 12 months: no cooing or reciprocal babbling and no use of social gestures;
- at 18 months: no shared enjoyment and no meaningful single words;
- at 24 months: no spontaneous two-word phrase and no interest in other children;
- at any age: poor eye contact, no response when name is called and developmental regression, loss of existing language, words and social skills.

In this regard, family physicians or paediatricians working in a primary care practice most often observe a child during infancy and early youth. This fact allows close observation of the child’s development and behaviour during the critical period for the diagnosis of ASD, which means that the family physician may be the first to notice behavioural signs of disorders.<sup>4</sup>

This is why it is increasingly important having specific psychometric tools to screen early symptoms of ASD so that timely intervention can take place.

### RECOMMENDED PSYCHODIAGNOSTIC TOOLS

Currently, there are several screeners for children from the age of 12 months, 18–24 months and up to 30–36 months (Tab. 1). According to scientific evidence, the tools listed below represent those with better reliability and validity, despite their limitations and the challenging nature of ASD early diagnosis.<sup>8</sup>

Some tools are considered Level 1 screeners because they are intended to screen all children regardless of their risk for developmental disabilities, including ASD.<sup>8</sup> As already stated, *response to name* represent a “red flag” for ASD. Children aged 4–6 months listen significantly longer to their own names than to other names, suggesting that

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**Table 1. Recommended psychodiagnostic tools**

Full name of the tool	Number of items and administration strategy	Administration age (months)	Assessed dimensions	Sensitivity (Se) and Specificity (Sp)
<b>LEVEL 1 SCREENERS</b>				
<b>First-Year Inventory (FYI)</b>	63 item – caregiver checklist	12	Social communication and sensory-regulatory functions domains	Se: 44 %, Sp: 99 %. <sup>8</sup>
<b>Infant-Toddler Checklist (ITC)</b>	24 item – caregiver checklist	8-24	Social and communicative behaviours	Se: 88,9 %, Sp: 88,9 %. <sup>9</sup>
<b>Modified Checklist for Autism in Toddlers – revised with follow up (M-CHAT-R/F).</b>	20 item (+ in-depth item) – caregiver checklist	16-30	Wide range of behaviours	Se: 97 %, Sp: 99 %. <sup>10</sup>
<b>LEVEL 2 SCREENERS</b>				
<b>Autism Diagnostic Observation Schedule, Second Edition (ADOS-2)</b>	Standardized observation of behaviours using objects and coding	From 12 to adulthood	Communication, social interaction, play, imagination, and repetitive and/or restricted behaviour	Different Se and Sp for each module. <sup>11</sup>
<b>Screening Test for Autism in Two-Year-Olds (STAT)</b>	12 item – semi structured play-based interactive tool	12-36	Play, request, direction of attention and motor imitation domains	Se: 92 %, Sp: 85 %. <sup>8</sup>
<b>Parent Observation of Early Milestones Scale (POEMS)</b>	61 item – caregiver checklist	From 18	Wide range of behaviours	Se: 74 %, Sp: 87 %. <sup>8</sup>
<b>Autism Detection in Early Childhood (ADEC)</b>	16 item – clinician-administered play interaction tool	12-36	Wide range of behaviours	Se: 100 %, Sp: 90 %. <sup>8</sup>

the sound patterns of children's names are internalised early in life.<sup>12</sup> The lack of response may be a general marker for ASD between 12 to 18 months but a specific indicator of ASD by 24-months.<sup>13</sup> In fact, evaluation of name response should be included in each ASD evaluation. This task involves calling the infant's name in a clear voice at a normal volume up to 3 times while the infant is engaged with toys and seated in the parent's lap, not facing the examiner. Then the examiner codifies if the response has been produced at the first, second or third time, or there is not any response.<sup>14</sup> However, the response to name can be assessed in different ways.<sup>15</sup>

Another useful tool for screening risk of ASD is *First-Year Inventory (FYI)*.<sup>16</sup> This is a parent-rated questionnaire with 63 items with different response types for screening 12-month-olds. The items are based upon the empirical literature on early symptoms of ASD. The authors considered items covering the wide categories of "social communication" and "sensory-regulatory functions" and intentionally included some items indicating "generalised developmental delay" because of their strongest link to autism. The authors have identified eight constructs in the following two domains: (1) social-communication domain: social orientation and receptive communication, social-affective engagement, imitation and expressive communication; and (2) sensory-regulatory domain: sensory processing, regulatory patterns, reactivity and repetitive behaviour. The FYI provides a total score with a cut off of 17 representing the

presence of development risk. The FYI is currently only available for research purposes; however, the authors are in the process of testing its clinical utility.

An additional screening tool for risk of ASD is *Infant-Toddler Checklist (ITC)*. The ITC is a 24-item caregiver-report checklist for children aged 8–24 months, and it is part of the Communication and Symbolic Behaviour Scales Developmental Profile.<sup>9</sup> ITC can be compiled by a very familiar caregiver in about 5 minutes. Because the ITC was originally developed as a language screening tool, it focuses almost exclusively on social and communicative behaviours. In fact, there is a lack of items assessing repetitive behaviours, unusual sensory reactions, or temperament problems. An important limitation of this tool is the inability to distinguish ASD from other neurodevelopmental disorders, but it shows high level of specificity, high negative predictive value, low false-positive rate, and identifies children with developmental delay and ASD.<sup>17</sup>

The most well-used instrument in the United States and in Europe is the *Modified Checklist for Autism in Toddlers – revised with follow up (M-CHAT-R/F)*.<sup>10</sup>

The M-CHAT-R/F is a 20-item and 2-stage parent-report screening tool. When a child screens positive on the first part, a structured Follow-Up is administered, during which more information about at-risk item are collected to solicit examples and clarify child behaviours to confirm or disconfirm the caregiver's initial response. It is valid for screening toddlers between 16 and 30 months of age.

The primary goal of the M-CHAT-R is to maximise sensitivity, meaning to detect as many cases of ASD as possible. To reduce the risk of false positives follow-up questions (M-CHAT-R/F) have been developed. Total score at M-CHAT-R could be assessed as follows: (1) Low-risk (0-2): if child is younger than 24 months, screen again after second birthday; (2) Medium-risk (3-7): follow up version should be administered to get additional information about at-risk responses. If M-CHAT-R/F score remains at 2 or higher, the child has screened positive; (3) High-risk (8-20): it is acceptable to bypass M-CHAT-R/F and refer immediately for diagnostic evaluation and eligibility evaluation for early intervention.

Considering the early age for which screening tools are structured, it should not be forgotten that these are not diagnostic tools. In fact, next to these level 1 screening instruments, also level 2 screeners are useful to distinguish ASD children already identified as being at high risk of developmental disabilities and delay.

One of these is the *Autism Diagnostic Observation Schedule, Second Edition (ADOS-2)*, a semi-structured and standardised instrument designed to assess skills, such as communication, social interaction, play, imagination, and repetitive and/or restricted behaviour.<sup>11</sup> The administration of the ADOS can be done by professionals from different disciplines, such as psychological, medical or related, and lasts between 40 and 60 minutes depending on the chosen module or the responses of the child examined. During the administration, the examiner delivers the tasks set within each module, and codes the behavioural and linguistic responses for social and communication skills related to the diagnostic criteria of ASD.

There are five modules within the ADOS and they are divided according to age and verbal competence. It starts from The Toddler module, for children from 12 to 30 months without capability in the use of phrasal language, to Module 4, for adolescents and adults with excellent verbal fluency skills. Accordingly, code 0 indicates the absence of abnormal behaviour, code 1 highlights slightly unusual behaviour, code 2 points out abnormal behaviour and code 3 reports the presence of largely abnormal behaviour.<sup>11</sup>

Another interactive, semi-structured instrument to diagnose early ASD is the *Screening Test for Autism in Two-Year-Olds (STAT)*, for children between 12 and 36 months of age.<sup>18</sup> It focuses on social communication, joint attention, pretend play, gestural imitation and functional use of objects, through its 12 items divided into the domains of Play, Request, Direction of Attention and Motor Imitation. The STAT can be administered by various specialists such as psychologists, paediatricians, speech therapists, social workers and kindergarten teachers, and takes approximately 20 minutes to complete. The total score, which will determine the final diagnosis, will be given by the 'pass', 'fail' or 'refuse' rating given to each item. The advantage of STAT consists in assessing the risk of autism diagnosis at an early stage.

Another Level 2 screening tool is *Parent Observation of Early Milestones Scale (POEMS)*.<sup>19</sup> The POEMS is a 61 items caregiver-report checklist, with a best assessment performance in 18 months children.<sup>8</sup> The items address a wide range of behaviours: early social and communication skills or deficits, restricted interests, ritualistic, repetitive, non-functional behaviours, intolerance to transitions and waiting, difficulties with new foods, loud noises, sleeping, and toileting, problems with attention and visual tracking, and problems with motor agility and movement. Each item was rated on a scale of 1 to 4, where 1 represents typical, non-problematic behaviour and 4 represents extreme difficulty. By summing the results to each item, a total score is obtained.

To conclude, a further tool for early diagnosis is *Autism Detection in Early Childhood (ADEC)*.<sup>20</sup> The ADEC is a clinician-administered play interaction tool with 16 items for children aged 12–36 months. The specific behaviours that are observed during the administration of the ADEC are response to name, imitation, ritualistic play, joint attention and social referencing, eye contact, functional play, pretend play, reciprocity of smile, reaction to common sounds, gaze monitoring, following verbal commands, delayed language, anticipation of social advances, nestling, use of gestures, and task switching. Response scores for each item is between 0 (appropriate) and 2 (inappropriate), with a possible maximum score of 32.

In conclusion, according to health psychology, prevention and monitoring have a key role in the management of pathologies,<sup>21</sup> and particularly in neurodevelopmental ones, such as ASD or ADHD.<sup>22</sup> This topic is particularly critical given the major changes and difficulties brought by COVID-19 pandemic at different levels, like early diagnosis, prevention, stress and learning.<sup>23-27</sup> Despite their limitations, the discussed tools allow clinicians to diagnose autism for ensuring the best desirable intervention.

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Manuscript writing: Francesco Maria Boccaccio, Giuseppe Alessio Platania, Claudia Savia Guerrera, Simone Varrasi and Chiara Rosa Privitera; data collection: Francesco Maria Boccaccio, Claudia Savia Guerrera; writing-review and editing: Sabrina Castellano, Concetta Pirrone, Pasquale Caponnetto; Simone Varrasi; supervision: Sabrina Castellano, Concetta Pirrone.

#### CONFLICT OF INTEREST

The authors declare no conflict of interest.

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