

ON OR OFF THE “SPECTRUM”?

The Complexity of Screening and Diagnosing Autism Spectrum Disorder (ASD)

Scott Shaffer, MD, and Joaquin Fuentes, MD

Recent estimates by the Centers for Disease Control (CDC) have suggested that 1 in 68 children are diagnosed with an autism spectrum disorder (ASD).¹ As the number of children diagnosed has risen steadily over the past decades, there has been an understandable surge of interest in the condition amongst health care professionals, teachers, the general public, and certainly amongst parents and advocates. Moreover, the DSM-5 issued a new set of diagnostic criteria that combined several DSM-IV-TR diagnoses, including autistic disorder, pervasive developmental disorder not otherwise specified (NOS), and Asperger’s Disorder into the new, singular diagnosis of autism spectrum disorder (ASD). The DSM-5 defines ASD as a neurodevelopmental condition characterized by impairments in social communication and interaction, as well as “restricted and repetitive patterns of behavior.”² Child and adolescent psychiatrists, along with their colleagues in pediatrics, neurology, and psychology, are now being asked more and more questions about how to screen for and identify children who may be “on the spectrum.”

Clinicians and researchers have known for a long time that earlier diagnosis generally leads to earlier identification of needs, more appropriate treatment for the child, better educational planning, and decreased family stress.³ Therefore, it is crucial that healthcare providers who see children (and adults) familiarize themselves with the typical signs and symptoms suggestive of ASD, as well as with screening and diagnostic tools used to help confirm this diagnosis. Clinicians often wonder how they should integrate their own clinical judgment with the results of a rating scale or diagnostic measure. This article will conclude with two diagnostic dilemmas which address this issue.

Developmental Signs of ASD

Research indicates that symptoms of ASD may be detectable in infancy, and by two years of age, a diagnosis by a professional is considered reliable.¹ Children with ASD can present in different ways depending on their age.

The prospective follow up of babies-at-risk (that is, children that were born in the family after the diagnosis of ASD in an older sibling) constitutes an extremely useful research venture, as about 20% develop ASD them-

selves.⁴ This high-risk sample allows us to see the first nonspecific symptoms observable at around six months of age, notably not social aspects but motor delays, such as head lag (weak head and neck control).⁵ Then, in the following months, one can observe unusual and unexpected developmental aspects, like over-fixation on vision on objects, decreased interest in people, and slowed development of language. Characteristics of progressive social isolation, as well as non-functional interest in objects and poverty of verbal and non-verbal communication become manifest between 18 and 24 months (a one-hour video presentation of high-risk babies by expert Lonnie Zwaigenbaum is available online at <http://sfari.org/sfari-community/community-blog/webinar-series/2013/webinar-lonnie-zwaigenbaum-discusses-high-risk-infant-studies>).

Later in development, preschool-age children (age two to four years) typically demonstrate important milestones in their social development. For example, by two years of age, most children respond to their first name, copy the actions of other children and adults, become excited when with other children, and point to objects and pictures when they are named. Two year old children with ASD often exhibit a significant lack of these skills and are clearly less interested in others.

By the time typically developing children are three years old, they show affection for other children without prompting or concern for a child who is crying, and they can participate in conversations using two to three sentences. Three year old children with ASD often do not demonstrate these early signs of empathy, and they frequently have absent or delayed speech.

By four years of age typically developing children enjoy doing new things and their play with others becomes more creative and cooperative. Four year olds with ASD are resistant to change and start to demonstrate restricted interests and stereotyped movements.

Healthy school age children (age five to twelve years) further develop their social skills and are able to adapt to changing expectations and environments. Self-stimulatory behaviors that were present earlier typically are no longer present by this stage. School age children with ASD frequently have difficulty coping with change,

and they often continue to demonstrate self-stimulatory behaviors that can become more prominent.

Adolescents and adults face increased social demands, and they typically have developed the skills to navigate their social worlds. In contrast, adolescents and adults with ASD often struggle to fit in socially with their peers.⁶

Screening for ASD


Pediatricians are often the first healthcare providers to identify concerns suggestive of ASD, although frequently the initial worries come from the parents. The American Academy of Pediatrics (AAP) recommends that all children be screened for general developmental progress by nine months of age.³ There are many general developmental screening tools used by clinicians. For example, the Denver-II Developmental Screening Test (<http://denverii.com/denverii>) is frequently used to screen expressive and receptive language, gross motor, fine motor, and personal-social skills. It is published by Denver Developmental Materials and is available for \$40.⁷

AAP also recommends that all children be screened specifically for ASD at 9 months, 18 months and 24 or 30 months.¹ The AAP recommends that additional screening be considered if a child is at high risk for ASD. Risk factors include having a sibling with ASD, preterm birth, and low birth weight.³ Additional indications for further evaluation include no babbling by 12 months, no gesturing (pointing, waving bye-bye) by 12 months, no single words by 16 months, no 2-word spontaneous phrases by 24 months, or loss of language or social skills at any age.^{2,3} While there are multiple screening measures available, the Modified Checklist for Autism in Toddlers (M-CHAT) and the Childhood Autism Spectrum Disorders Test (CAST) are commonly used as both are free, have been adapted cross culturally, can be applied for different ages, and have a wealth of research supporting them.² The M-CHAT will be highlighted here. The CAST is applicable to older children, 4 to 11 years old, and was initially established to screen for Asperger's disorder.

ASD Screening with M-CHAT R/F

The Modified Checklist for Autism in Toddlers (M-CHAT R/F, freely available at <https://m-chat.org>) is a screening tool created by Diana Robins which identifies children at risk for ASD.⁸ It was revised in 2009 in order to reduce the number of cases that screen as false positives.⁸ The M

-CHAT R/F is based on parental report of children's current skills and behaviors. It can be filled out by a parent, requires no special training to use, and is available for free in the public domain. It is validated for use in children between 16-30 months of age. It was developed to ensure high sensitivity, meaning it will pick up as many cases of ASD as possible. This ensures that more patients who truly have ASD will obtain a positive result.⁹ There is a high false positive rate associated with it, meaning that a significant number of these patients will not eventually meet criteria for ASD, but they may have other developmental problems meriting intervention. This high false positive rate has been decreased with a new two stage process.



**“Research indicates
that symptoms of ASD
may be detectable in
infancy”**

The parent questionnaire takes less than two minutes to score. Children are scored either as low-risk, medium-risk, or high-risk based on the parents' answers to 20 questions. Children scored as low-risk do not need further evaluation unless they are less than two years of age and will require subsequent standard developmental screens or if there is clinical concern. Children scored as medium-risk should receive the “second stage” questions of the M-CHAT R/F. If the child continues to screen positive, the child should be referred for a diagnostic evaluation by a

specialist with expertise in ASD as well as be referred to early intervention programs. If a child is scored as high risk, he or she should be referred for a diagnostic evaluation and the second stage questions are unnecessary.⁸

If a clinician is concerned about a child with a negative M-CHAT, a referral to a specialist should still be made, but the use of the M-CHAT may help clarify the nature of the referral (e.g. “although the child screened negative, I have noticed certain behaviors that merit further evaluation”). A positive M-CHAT R/F and follow-up interview does not necessarily mean a diagnosis of ASD, as many patients will not ultimately meet criteria for the diagnosis. However, the patients who screen positive but subsequently do not have ASD should be evaluated for other developmental delays.

Diagnostic Evaluations

Specialists will typically employ a diagnostic measure to assist with the diagnosis of ASD. Two such measures, the Childhood Autism Rating Scale (CARS-2) and the Autism Diagnostic Observation Schedule (ADOS) will be highlighted here.

Childhood Autism Rating Scale (CARS-2)

The CARS-2, which is a diagnostic observation instrument first developed by Schopler et al., is a 15-item structured interview and observation instrument which can be used for children over two years of age.¹⁰ The CARS-2 is published by Western Psychological Services and is available for \$175 (<http://www.wpspublish.com/store/p/2696/childhood-autism-rating-scale-second-edition-cars-2>). The CARS-2 can be administered by a range of professionals with knowledge of normal child development.¹⁰ It takes about 30-45 minutes to administer. The CARS-2 is not a standardized measure and should not be used to diagnose a child with ASD independent of clinical judgment. It can provide useful data to help support a diagnosis of ASD, or facilitate necessary referrals.

With the CARS-2, fourteen domains assess behaviors associated with ASD, and the fifteenth domain rates general impressions of ASD. Each item is scored on a scale from 1 to 4. Total scores can range from a low of 15 to a high of 60. Scores below 25.5 indicate that the patient likely does not have an ASD, scores between 25.5-36.5 indicate mild to moderate symptoms of ASD, and scores between 37-60 are consistent with severe symptoms of ASD. Perry et al., demonstrated that in a sample of 274 preschool children, there was an agreement rate of 88% between classification made by the CARS and the DSM-IV.

While the CARS has high sensitivity, it appears to over diagnose young children with intellectual disability as having ASD. Consequently, children can be incorrectly diagnosed and unnecessary referrals for expensive intervention services can occur, as is the case for all screening instruments.¹¹ Filipek states that instruments used to diagnose ASD should have moderate sensitivity and good specificity.³ The CARS demonstrates strong internal consistency and inter-rater reliability.³ In 2010, a second edition for high functioning individuals, CARS2-HF, was developed including a version meant for children who have greater intellectual and verbal abilities.

Autism Diagnostic Observation Schedule (ADOS)

The ADOS is a semi-structured observational standardized assessment that includes investigator-led activities to assess communication, reciprocal social interaction, play, stereotypic behavior, restricted interests, and other abnormal behaviors. The ADOS is published by Western Psychological Services and is currently at a price of \$1,995 for each kit (<http://www.wpspublish.com/store/p/2647/autism-diagnostic-observation-schedule-ados>).

Training is required to administer the ADOS. The interview takes about 30-45 minutes.³ It consists of four modules designed to be administered to individuals according

to their level of expressive language.³ During the assessment, social interactions referred to as “presses” occur in which a range of social initiations and responses is likely to appear. The main goal of the ADOS is to provide interactions that elicit spontaneous behaviors in standardized contexts.

The ADOS was first introduced in the 1980s as a method of standardizing direct observations of social behavior, communication, and play in children suspected of having autism. It was originally proposed as a complementary instrument to the Autism Diagnostic Interview, (ADI) which later became the Autism Diagnostic Interview-Revised (ADI-R).¹² The ADI-R consists of 93 items and is meant to be administered by an experienced clinician who has received training to use the instrument. It takes between 2-3 hours, and it is published by Western Psychological Services and is available for \$85 (<http://www.wpspublish.com/store/p/2645/autism-diagnostic-interview-revised-adi-r>).¹⁰

The ADOS-2 was published in 2012 and can be given to patients from age one year old through adulthood. There are five modules. The toddler module is used for children ages 12-30 months who have nonverbal mental ages of at least 12 months and do not consistently use phrase speech. Module 1 is used for children ages 31 months and older who have little or no phrase speech. Module 2 is used for children who have phrase speech but are not verbally fluent. Module 3 is used for children and young adolescents who are verbally fluent, and Module 4 is used for adolescents and adults who are verbally fluent.

Part of the testing is play-based and part of it is more structured. The presses are coded using a standardized rubric and then the scores are tallied using a diagnostic algorithm. Each press is given a score between 0 and 3, with 3 representing a more significant impairment. The individual items are grouped by domain including communication, reciprocal social interaction, and restricted and repetitive behaviors. The diagnostic algorithm score is then calculated which is compared to predetermined cutoff values, corresponding to diagnostic classifications of autism, autism spectrum, or non-spectrum. The toddler module uses different classifications which include little to no concern, mild to moderate concern, and moderate to severe concern. In addition, a comparison score is included which is meant to compare autism severity between individuals and over time. The score is on a scale of 1-10, with 10 being the highest severity score.

The use of the ADOS and ADI-R together provide more diagnostic clarity than when they are used alone. Using these two measures, combined with clinical judgment,

can ensure the highest levels of sensitivity and specificity.¹³

Diagnostic Dilemma #1

Jill is a 7 year old girl who presents with her parents complaining that she “drives them crazy.” She is always in her own world and she is extremely distractible. She is very oppositional and only interested in what she wants to do at any given moment. She is emotionally labile. She is so hyperactive that other children do not want to be around her. She becomes upset when routines change. Yesterday, she had a tantrum in school when there was a fire drill because the teacher would not let her play on the swings. She is described as fearless. She has some sensitivity to texture as she does not like the feeling of the tags on her shirt.

During her evaluation, she engages in reciprocal conversation with you about her interests and school. You decide to administer a CARS as Jill’s parents are anxious for answers regarding her diagnosis and you are not trained to administer an ADOS, nor is it available in the clinic. After interviewing the parents, she receives a score of 31 on the CARS-2, which is consistent with “mild to moderate symptoms of autism spectrum disorder.” You were surprised as your clinical judgment did not suggest ASD.

This case is an example of when a clinician’s judgment that a child is not “on the spectrum” differs from a diagnostic instrument which produces a score consistent with an ASD diagnosis. This patient has many symptoms which may be consistent with diagnoses such as attention-deficit/hyperactivity disorder (ADHD) and oppositional defiant disorder (ODD). Some categories on the CARS, such as Emotional Response and Activity Level, have significant overlap with these diagnoses, which consequently inflated this patient’s score. The clinician is also dependent on the parent’s reporting, which can sometimes result in over- or under-reporting of symptoms. A referral to a clinician who specializes in ASD is recommended for a complex case such as this.

Diagnostic Dilemma #2

Johnny is a 7 year old boy who has been having a difficult time making friends. When he is interacting with his peers, he wants to talk about different models of airplanes and other children quickly lose interest. He has difficulty reading their body language when they clearly are not in-

terested in what he is saying. He has a difficult time understanding sarcasm and humor. Johnny is very sensitive to loud sounds. He has trouble adjusting to changes in routine. He had a terrible day in school yesterday when there was an unannounced fire drill. He had a tantrum that lasted hours. Johnny repeatedly watches documentaries about airplanes, and he can recite the dialogues by memory. You suspect that Johnny may have an ASD and you decide to refer him to your colleague who is trained to administer the ADOS. During the ADOS, Johnny is friendly and cooperative with testing. The ADOS score is below the cutoff for ASD. You are surprised as your clinical judgment suggested that Johnny had ASD.

This case is an example of when a clinician’s judgment that a child has an ASD is not supported by a diagnostic instrument. Johnny is quite intelligent and verbal. If he has already received interventions such as speech therapy, he has likely made progress through the years. A careful developmental history is essential as the clinician needs to establish whether there is a history of symptoms that the patient may have “grown out of” thanks to interventions he has received. As the ADOS is observational in nature, important information that can be obtained in the clinical history is not necessarily reflected in the score.

“The use of the ADOS and ADI-R together provide more diagnostic clarity than when they are used alone”

Take-Home Summary:

Screening for Autism spectrum disorder (ASD) is part of routine evaluation for all children. Screening and diagnostic tools exist to assist in diagnosis and results should be considered within the context of clinical observations and expected developmental milestones.

Conclusion

ASD can often be challenging and complex to diagnose as they can have vastly different presentations in patients of different ages and abilities. While there are several screening and diagnostic instruments that are valid and reliable, as in other conditions in child and adolescent psychiatry, they must be used in combination with solid clinical reasoning based on an effective interview and comprehensive history.

References:

- Centers for Disease Control and Prevention. Screening and Diagnosis for Healthcare Providers. <http://www.cdc.gov/ncbddd/autism/hcp-screening.html> and <http://www.cdc.gov/ncbddd/autism/facts.html>. Accessed July 24, 2014.
- Fuentes, J, Bakare M, Munir K, et al. Autism Spectrum Disorder. In Rey JM (ed), JACAPAP e-Textbook of Child and Adolescent Mental Health. Geneva: International Association for Child and Adolescent Psychiatry and Allied Professions 2014, Geneva.
- Filipek PA, Accardo PJ, Baranek GT, et al. The Screening and Diagnosis of Autistic Spectrum Disorders. J Autism Dev Disord. 1999; 29(6):439-484.
- Ozonoff S, Young GS, Carter A, et al. Recurrence risk for autism spectrum disorders: a Baby Siblings Research Consortium study. Pediatrics. 2011 Sep;128(3):e488-95. peds.2010-2825. Epub 2011 Aug 15.
- Flanagan JE, Landa R, Bhat A, Bauman M. Head lag in infants at risk for autism: a preliminary study. Am J Occup Ther. 2012 Sep-Oct;66(5):577-85.
- Centers for Disease Control and Prevention. Developmental Milestones. <http://www.cdc.gov/ncbddd/actearly/milestones/>. Accessed July 24, 2014.
- Committee on Children with Disabilities. Developmental Surveillance and Screening of Infants and Young Children. Pediatrics, Jul 2001, Vol. 108 Issue 1, 192-96.
- Robins DL, Casagrande K, Barton M, Chen CM, Dumont-Mathieu T, Fein D. Validation of the Modified Checklist for Autism in Toddlers, Revised with Follow Up (M-CHAT-R/F). Pediatrics. 2014 Jan;133(1):37-45.
- Robins D. M-Chat: Free. On Line. Instant. <http://www.m-chat.org/print.php>. Accessed October 15, 2014.
- McClintock J, Fraser J. Diagnostic instruments for autism spectrum disorder: A brief review. New Zealand Guidelines Group, April 2011.

About the Authors

Scott Shaffer, MD, is an Assistant Professor of Pediatrics and Psychiatry and Behavioral Sciences at the Albert Einstein College of Medicine of Yeshiva University, New York. His interests include medical education and treating patients with autism spectrum disorder. He is also a board member of the New York Council for the American Academy of Child and Adolescent Psychiatry, and serves on the national advisory council of the Gold Humanism Honor Society.

Joaquin Fuentes, MD, is a corresponding member of the American Academy of Child and Adolescent Psychiatry. He practices in Donostia / San Sebastián, Spain, and currently serves in the Steering Committee of Dr. Paramjit Joshi's AACAP Presidential Initiative. Counselor of the Executive Committee of the International Association for Child and Adolescent Psychiatry and Allied Professions (IACAPAP) until 2018. He served as a mentor to Dr. Shaffer during the Medical Students and Residents Mentorship Program at the AACAP 56th Annual Meeting in Hawaii, 2009. Dr. Fuentes has received research support, has served as unrestricted speaker for, or has served on the advisory boards of Eli Lilly and Co., Janssen, Shire, Roche and Medice. He has not received any financial support to produce this paper.

GET CONNECTED TO AACAP!

AACAP provides exceptional educational resources and opportunities worth exploring:

- Medical Students, Residents, and Early Career Psychiatrists:** AACAP offers mentorship and educational programs; information on rotations, residency, and careers; networking; AACAP conference programs for trainees; membership, and much more! Visit www.aacap.org, and click on the [Medical Students and Residents](#) section.
- Resources:** Under "Resources for Primary Care" at www.aacap.org, you can find Practice Parameters, patient handouts, Clinical Practice resources, membership, and more. See [Member Resources](#) to learn about becoming a mentor!
- JAACAP** content is available at www.jaacap.com. AACAP members enjoy free online access to JAACAP content and to the *Child and Adolescent Psychiatric Clinics North America* and *Psychiatric Clinics of North America*.

Learn more about becoming an AACAP member at www.aacap.org.