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SYMPTOM PRESENTATIONS AND CLASSIFICATION OF AUTISM SPECTRUM DISORDER IN EARLY CHILDHOOD: APPLICATION TO THE *DIAGNOSTIC CLASSIFICATION OF MENTAL HEALTH AND DEVELOPMENTAL DISORDERS OF INFANCY AND EARLY CHILDHOOD (DC:0–5)*

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Abstract

Over the past 5 years, a great deal of information about the early course of autism spectrum disorder (ASD) has emerged from longitudinal prospective studies of infants at high risk for developing ASD based on a previously diagnosed older sibling. The current article describes early ASD symptom presentations and outlines the rationale for defining a new disorder, Early Atypical Autism Spectrum Disorder (EA-ASD) to accompany ASD in the new revision of the ZERO TO THREE *Diagnostic Classification of Mental Health and Developmental Disorders of Infancy and Early Childhood (DC:0–5)* (in press) alternative diagnostic classification manual. EA-ASD is designed to identify children who are 9 to 36 months of age presenting with a minimum of (a) two social-communication symptoms and (b) one repetitive and restricted behavior symptom as well as (c) evidence of impairment, with the intention of providing these children with appropriately tailored services and improving the likelihood of optimizing their development.

Keywords

autism spectrum disorder; early atypical autism spectrum disorder

In 2010, Yirmiya and Charman argued that there were insufficient data to advocate for the diagnosis of an *autistic disorder prodrome*. In the past 6 years, a great deal of new information has emerged on the early course of autism spectrum disorder (ASD), primarily from studies of infants at high risk by virtue of having an older sibling with ASD (e.g., Bryson, Zwaigenbaum, Brian, Roberts, Szatmari, Rombough, & McDermott, 2007; Chawarska et al., 2014; Macari et al., 2012). Prospective studies of infants within a familial high-risk design follow younger siblings of children with ASD from early infancy or the prenatal period, prior to the onset of symptoms, and compare them to a low-risk contrast group consisting of infants with a typically developing older sibling without a family history of ASD. The surprisingly high recurrence risk among these high-risk siblings, which was

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found to be 18.7% in a large, multisite prospective study (Ozonoff et al., 2011), has allowed investigators to study the early developmental course of large numbers of children who go on to develop ASD. Such studies have greatly contributed to further understanding of the initial development and early markers of ASD, thus making the potential for earlier identification and appropriate interventions increasingly possible.

The goals of this article are to (a) describe early ASD symptom presentations and (b) outline the rationale for creating a new disorder, Early Atypical Autism Spectrum Disorder (EA-ASD) in the ZERO TO THREE *Diagnostic Classification of Mental Health and Developmental Disorders of Infancy and Early Childhood* (*DC:0–5*) (in press) manual. ASD is now included in the *DC:0–5* in a manner that is developmentally specified for very young children and fully aligned with the *Diagnostic and Statistical Manual of Mental Disorders* (*DSM-5*; American Psychiatric Association, 2013) criteria. The EA-ASD diagnosis is designed to capture children who are within the window of risk for developing ASD, and are evidencing multiple ASD symptoms with impairment, but do not currently meet and have never met the full criteria for ASD. Informed by international studies of infant siblings of children with ASD that have revealed a broad window of risk for the emergence of full ASD criteria, marked variability in age at onset for stable ASD diagnoses that occur between 12 and 36 months of age (Ozonoff et al., 2015; Zwaigenbaum et al., 2015), and patterns of slow regression through the window of risk for many children who go on to meet full ASD criteria (Landa, Gross, Stuart, & Bauman, 2012; Ozonoff et al., 2014), we believe that there is now sufficient evidence to introduce a disorder for infants and toddlers who present impairing, subthreshold ASD symptomatology.

In contrast to the three social-communication and two restricted and repetitive behavior symptoms required to meet diagnostic criteria for ASD in the *DSM-5* and the *DC:0–5* (in press), a minimum of two social-communication symptoms and one restricted and repetitive behavior symptom are required for making a diagnosis of EA-ASD. Moreover, EA-ASD is only appropriate for use with infants and toddlers who are in the age window of risk for ASD (9–36 months; Baird et al., 2000; Bryson, Rogers, & Fombonne, 2003; Charman et al., 2005; Lord et al., 2006); do not meet criteria and have not been previously diagnosed with ASD; and are evidencing clear impairment, but do not evidence sufficient symptoms to meet full *DSM-5*/*DC:0–5* ASD diagnostic criteria. This diagnosis is not intended to apply to children whose behavior is better explained by a language or an intellectual delay/disability/disorder or other psychopathology.

The corpus of work emerging from longitudinal infant sibling studies has provided clear evidence that the onset of ASD is often gradual and marked by a slowing developmental progression of social-communication competencies. Indeed, several excellent recent comprehensive reviews of this literature (Jones, Gliga, Bedford, Charman, & Johnson, 2014; Mitchell, Cardy, & Zwaigenbaum, 2011) have highlighted that ASD onset can be gradual and that there is a relatively broad window of risk between 12 and 36 months of age. In this article, we offer our clinical perspective, selectively reviewing the prospective infant sibling literature and retrospective home-video studies to provide limited empirical support for EA-ASD. To facilitate early identification, we also offer examples of how ASD symptoms can present in the infant–toddler and preschool periods.

AUTISM SPECTRUM DISORDER IN EARLY CHILDHOOD

ASD is characterized by impairments in social interaction and communication, and the presence of restricted and repetitive behaviors (American Psychiatric Association, 2013; World Health Organization, 1992; ZERO TO THREE, in press). Once considered rare, ASD is now among the most common neurodevelopmental disorders, with current estimates in the United States of 1 in 68 children affected (Centers for Disease Control and Prevention, 2014). Accurate and early identification of ASD is critical, particularly given the growing prevalence (Fombonne, 2009), considerable family and societal costs (Ganz, 2007), and recognized importance of early intervention (Seida et al., 2009; Woods & Wetherby, 2003). Early identification of ASD coupled with subsequent high-intensity, evidence-based early intervention are effective in improving language outcomes and ameliorating many of the serious symptoms characteristic of and associated with the disorder (for a review, see Bradshaw, Steiner, Gengoux, & Koegel, 2015). Measurement is the first step in determining appropriate treatment; thus, increased efforts have been directed toward improving tools and methods for earlier detection and diagnosis of ASD in the general population (Robins & Dumont-Mathieu, 2006). Coupled with the push for increased screening and early identification (Charman, 2014) is the need to have a formal diagnosis that supports careful monitoring and appropriate intervention for children who are manifesting emerging signs of ASD with functional impairment. The burgeoning body of knowledge resulting from prospective studies of high-risk infant siblings has elucidated a clear picture of the wide individual variation in developmental trajectories of infants who go on to develop ASD. Findings from these studies as well as those from retrospective studies provide the rationale for the *DC:0–5* EA-ASD.

Findings From Prospective and Retrospective Studies

Longitudinal prospective studies of infants with older sibling(s) diagnosed with ASD (high-risk infant siblings), who are at significantly increased risk for developing ASD (18.7 vs. 1.5% in the general population; Ozonoff et al., 2011), have identified early emerging symptoms of the disorder. These studies have documented that in general, ASD symptoms emerge during a window of risk that opens in the second half of the first year of life and dramatically narrows at approximately 36 months of age (Baird et al., 2000; Bryson et al., 2003; Charman et al., 2005; Lord et al., 2006; Ozonoff et al., 2015; Zwaigenbaum et al., 2015). Critically, the presentation of ASD characteristics is heterogeneous (Kim, Macari, Koller, & Chawarska, 2015), and there is great variability in age at onset (Ozonoff et al., 2015; Zwaigenbaum et al., 2015). This body of literature has documented robust evidence for the emergence of ASD symptoms as early as 12 months of age as well as the ability to predict ASD diagnostic outcome at 36 months from the presence of a high number of symptoms at 12 and 18 months of age (Bryson, Zwaigenbaum, Brian et al., 2007; Chawarska et al., 2014; Macari et al., 2012). A diagnosis, such as EA-ASD would enable children presenting with subthreshold ASD symptoms (that are not better explained by the presence of a language disorder or global developmental delay) to be more closely monitored during a critical stage of development.

When children meet full criteria for ASD prior to 3 years of age in both high-risk and general population samples, the diagnoses are stable over time, although studies to date have published stability rates that range widely and vary significantly across *Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition, Text Revision* (*DSM-IV-TR*; American Psychiatric Association, 2000) categories: 63 to 100% stability for Autistic Disorder (e.g., Ben Itzchak & Zachor, 2009; Chawarska, Klin, Paul, Macari, & Volkmar, 2009; Kleinman et al., 2008; Takeda, Koyama, Kanai, & Kurita, 2005; van Daalen et al., 2009), 17 to 100% for Pervasive Developmental Disorder-Not Otherwise Specified (e.g., Chawarska, Klin, Paul, & Volkmar, 2007; Kleinman et al., 2008; Mahli & Singhi, 2011), and 82 to 100% for dichotomized *DSM-IV-TR* ASD outcomes (Anderson, Liang, & Lord, 2006; Lord et al., 2006; Turner, Stone, Pozdol, & Coonrod, 2006). However, a recent systematic review of the diagnostic stability of ASD (Woolfenden, Sarkozy, Ridley, & Williams, 2012) has found that the stability increases when diagnoses are made later in childhood, as diagnoses made after 5 years of age were more stable over time than were diagnoses made earlier.

Findings from prospective studies of high-risk infant siblings coupled with studies of the stability of early diagnoses provide a foundation for understanding the manifestations of very early symptoms of ASD as well as the marked individual differences in the course of symptoms in the first years of life and beyond. Moreover, this foundation is scientifically rigorous, as prospective studies have eliminated many of the limitations encountered in earlier retrospective research, such as retrospective recall bias and selective home videotaping (Ozonoff et al., 2010; Palomo, Belinchón, & Ozonoff, 2006). At the same time, research that has been done with children at high genetic risk for ASD may not generalize to a general population sample. Moreover, most prospective studies for children at risk for ASD have reported mean findings rather than the sensitivity and specificity of symptom thresholds, and a majority has focused on prediction from 12 months of age. In the next sections, we offer a clinical perspective on evidence for the early emergence of symptoms across the two broad domains of ASD symptomatology: Social-Communication and Repetitive and Restricted Behaviors (RRBs). Table 1 describes specific examples of symptom presentations across the infancy/toddlerhood and preschool periods. All examples in the infancy/toddlerhood period also may be observed in the preschool period, particularly among children with delays/disabilities in language and cognitive development.

Social-communication symptoms in early childhood—Many prospective studies have found that at 6 months of age, high-risk infants who go on to later receive an ASD diagnosis show no differences in most social and communicative behaviors when compared to infants who do not receive a future diagnosis (Frohna, 2007; Landa & Garrett-Mayer, 2006; Ozonoff et al., 2010; Ozonoff et al., 2014; Yirmiya et al., 2006, Zwaigenbaum et al., 2009). However, by 12 months of age, many (but not all) infants later diagnosed with ASD can be differentiated from those who remain unaffected by a myriad of abnormal social-communication skills, including impaired social nonverbal behaviors, impaired social relationships, and impaired socioemotional reciprocity (Bryson, Zwaigenbaum, Brian et al., 2007; Frohna, 2007; Landa & Garrett-Mayer, 2006; Yimriya et al., 2006; Zwaigenbaum et

al., 2005). The following sections review the specific impaired behaviors and skills within each of these areas of social communication.

Significant limitations in social nonverbal behaviors—Prospective studies of high-risk infants over the last decade have provided evidence that by 12 months of age, some infants who go on to receive a diagnosis of ASD show impaired social nonverbal behaviors, or communicative strategies besides the use of language (Rowberry et al., 2015; Zwaigenbaum et al., 2005). Social nonverbal behaviors that are commonly assessed include eye contact, gestures, body language, and facial expressions as well as the coordination and integration of these behaviors flexibly across contexts.

Eye contact is used for a variety of social purposes in typically developing children, including for requesting and sharing interest, enjoyment, and other internal states. Zwaigenbaum et al. (2005) found atypical eye contact at 12 months old, measured on the Autism Observational Scales of Infancy (AOSI; Bryson, Zwaigenbaum, McDermott, Rombough, & Brian, 2007), a semistructured, interactive, play-based assessment, to be a significant predictor of ASD diagnosis at 24 months in a sample of high-risk infant siblings. In addition, Nichols, Ibañez, Foss-Feig, and Stone (2014) found that high-risk infant siblings who went on to receive a diagnosis of ASD showed a significantly lower rate of eye contact during administration of the Screening Tool for Autism in Toddlers and Young Children (Stone, Coonrod, & Ousley, 2000) at 15 months, as compared to both high-risk siblings who did not receive a future ASD diagnosis as well as low-risk typically developing children.

Gestures allow a child to symbolically represent requests, the objects in his or her environment, and aspects of these objects, especially prior to extensive language development. Mitchell et al. (2006) reported that 12-month-old, high-risk infant siblings both produced and understood fewer gestures than did low-risk infants, as measured by the MacArthur Communicative Development Inventory-Infant Form Words and Gestures (CDI-WG; Fenson, Dale, Reznick, & Bates, 1993). The parent-reported CDI-WG takes into account both “early gestures” (e.g., giving, showing) and “late gestures” (e.g., nodding head “yes”). Similarly, Yirmiya et al. (2006) found that at 14 months, high-risk infant siblings who went on to receive a diagnosis of ASD initiated fewer requesting gestures than did low-risk infants. Other prospective studies have shown that high-risk infants who go on to receive an ASD diagnosis have a smaller vocabulary of gestures and show delays in the acquisition of communicative and symbolic gestures (Landa, Holman, & Garrett-Mayer, 2007; Talbott, Nelson, & Tager-Flusberg, 2015). A decreased use of distal pointing continues to be seen in high-risk infants who go on to meet diagnostic criteria for ASD. For example, Rozga et al. (2011) observed that 12-month-old, high-risk infants later diagnosed with ASD were less likely to point than both high-risk infants who did not go on to receive an ASD diagnosis and low-risk controls.

Imitation is an important skill in early childhood development, as it provides a powerful means for young children to learn from and interact with others in their environment. In samples of high-risk siblings, both Zwaigenbaum et al. (2005) and Macari et al. (2012) found reduced imitation ability at 12 months of age to be a significant predictor of an ASD diagnosis at 24 months. Although not included as a symptom of ASD in the *DSM-5*|*DC:0–*

5, many studies have documented deficits in imitation among older children with ASD (Rowberry et al., 2015; Young et al., 2011), and it appears to be an important early marker of the emergence of ASD.

Significant limitations in social relationships—The *DSM-5* lists "deficits in developing, maintaining, and understanding relationships" (p. 54) as one of the mandatory diagnostic criteria for ASD. Prospective studies over the last decade have shown evidence that young children who go on to receive a diagnosis of ASD show significant and persistent deficits in many of the behaviors that lay the groundwork for developing sophisticated relationships in later childhood. For example, Cornew, Dobkins, Akshoomoff, McCleery, and Carver (2012) found that high-risk infants who met diagnostic criteria for ASD at 36 months of age sought social information from their environment at a significantly slower pace at 18 months of age, as compared to both low- and high-risk infants who did not later meet ASD diagnostic criteria. The authors concluded that social referencing, the strategy by which infants acquire social information about objects and events in their environment, has a predictive value for later ASD diagnosis.

A major focus of study in prospective research continues to be joint attention, or the ability to coordinate attention between people and objects (Bruner, 1975; Mundy & Jarrold, 2010). Response to joint attention (RJA) involves the skill of using another person's nonverbal communication (i.e., eye gaze, head turn, pointing, gestures, facial expression) to understand what he or she is paying attention to. Sullivan et al. (2007) found that the RJA abilities of 14-month-old, high-risk infants predicted their ASD outcome status at 24 months. Initiation of joint attention (IJA) is best described as directing another person's attention with social intent (i.e., for sharing an experience). Landa et al. (2007) reported that 14-month-old, high-risk infants who later met diagnostic criteria for ASD were less likely to engage in IJA than were other outcome groups.

In addition, both Nichols et al. (2014) and Zwaigenbaum et al. (2005) reported the predictive utility of social smiling, which involves the integration of a facial expression of positive affect and the orientation of eye gaze toward another person. Zwaigenbaum et al. (2005) found reduced frequency of social smiling at 12 months of age, measured on the AOSI, to be a significant predictor of an ASD diagnosis at 24 months in a sample of high-risk infant siblings. Moreover, Nichols et al. found that high-risk siblings who went on to receive a diagnosis of ASD showed a significantly lower rate of social smiling at 15 months of age, as compared to both a group of high-risk infants who did not receive a future ASD diagnosis as well as a group of typically developing infants. Furthermore, Ozonoff et al. (2010) found that at 18 months of age, frequency of social smiling distinguished infants who went on to receive a diagnosis of ASD from those who did not. Frequency of vocalizations/verbalizations directed toward others, another relational behavior, has been the focus of many studies over the last decade. Interestingly, Ozonoff et al. (2010) reported that while the frequency of directed vocalizations increased in the typically developing group between 6 and 12 months of age, it significantly decreased in the infants who went on to receive an ASD diagnosis.

Significant limitations in socioemotional reciprocity—Response to name is a skill that emerges by 4 to 6 months of age in typically developing infants. Prospective studies have shown that a deficit in this area does not emerge until 12 to 18 months of age among high-risk infants subsequently diagnosed with ASD (Brian et al., 2008; Nadig et al., 2007; Zwaigenbaum et al., 2005); in many children diagnosed with ASD, response to name remains a deficit through the preschool period. Other socioemotional reciprocity skills that have been found to be atypical in high-risk infants later diagnosed with ASD include decreased social engagement between 6 and 12 months (Ozonoff et al., 2010), reduced attentiveness to their mothers at 12 months (Wan et al., 2013), lower engagement with a researcher at 12 months (Hutman et al., 2010; Macari et al., 2012), decreased shared positive affect and orienting to a target in response to gaze/point prompts at 14 months (Brian et al., 2008; Landa et al., 2007; Sullivan et al., 2007), and reduced social referencing at 17 to 20 months (Cornew et al., 2012). In addition, Bryson, Zwaigenbaum, Brian et al. (2007) reported reduced social engagement, shared enjoyment, and nonverbal communication by 12 months of age.

Thus, a wide range of early emerging social-communication differences have been documented between 12 and 36 months of age when comparing infant siblings at risk for ASD and low-risk siblings, including differences in (a) using and understanding social nonverbal behaviors; (b) developing, maintaining, and understanding social relationships; and (c) engaging in socioemotional reciprocity. The aforementioned studies have highlighted group differences, but also note that there is dramatic individual variation in the patterns and trajectories of social-communication difficulties and deficits that precede the onset of ASD.

RRB symptoms in early childhood—The second domain of ASD symptoms is RRBs, which includes stereotyped or repetitive motor and vocal mannerisms, inflexible adherence to routines or rituals, preoccupations and restricted interests, and sensory behaviors. Although children with typical development may exhibit RRBs at young ages (e.g., flapping when excited), these behaviors decrease in frequency over time (DiGennaro Reed, Hirst, & Hyman, 2012). The early emergence of RRBs and its predictive validity of ASD are not as well established as social-communicative domains of functioning in prospective studies. However, there have been several promising findings that provide a broader picture of RRBs in the early emergence of ASD (e.g., Barber, Wetherby, & Chambers, 2012; Kim & Lord, 2010; Ozonoff, Heung, Byrd, Hansen, & Hertz-Picciotto, 2008; Stronach & Wetherby, 2014; Watt, Wetherby, Barber, & Morgan, 2008; Wetherby et al., 2004). The following sections review the specific behaviors present in early childhood that fall into each of the RRB categories described earlier.

Stereotyped or repetitive motor and vocal mannerisms—Well-documented motor stereotypies in young children include small hand mannerisms (e.g., finger flicking/twisting, hand posturing), and larger and/or complex repetitive body movements (e.g., arm flapping/waving, toe-walking, jumping, body posturing, spinning) (Kim & Lord, 2010; Ozonoff et al., 2008; Stronach & Wetherby, 2014). In one of the earliest prospective studies addressing RRBs in young children with ASD, Loh et al. (2007) examined RRBs generated during the administration of the AOSI at 12 and 18 months and found arm waving to be the only

behavior that distinguished a diagnosis of ASD at 36 months. Wetherby and Morgan (2007) developed the Repetitive and Stereotyped Movement Scales to specify coding of four repetitive body movements (i.e., flapping, stiffening, rubbing, and patting) and nine objectoriented repetitive behaviors that are often observed in play with toys (i.e., spinning, rocking, rolling, collecting, swiping, rubbing, moving, lining, and clutching). Wetherby and colleagues (Barber et al., 2012; Watt et al., 2008; Wetherby et al., 2004) compared ASD and age-matched language delayed groups (12–23 months old) and found that repetitive movements (either with body or with object) were more commonly observed in the ASD group (Wetherby et al., 2004). When followed over time, the ASD group demonstrated both higher frequency and longer duration of repetitive behaviors with objects and with the body (Barber et al., 2012; Watt et al., 2008). More work in this area is needed, as several studies of children at high risk for ASD have reported elevated rates of repetitive movements in all siblings at risk, not just those who go on to develop ASD, with exploratory analyses highlighting differential prediction to ASD for different patterns of RRBs (e.g., Damiano, Nahmias, Hogan-Brown, & Stone, 2013).

In addition to repetitive object- and body-focused behaviors, stereotyped or repetitive behaviors include vocal mannerisms such as echolalia (i.e., repeating another person's speech with the same intonation pattern) and repeating the same syllable(s), sound, word, or phrase over and over (DiGennaro Reed et al., 2012; van Santen, Sproat, & Hill, 2013). While these behaviors are observed in very young children with ASD, they have received less attention in the high-risk literature.

Inflexible adherence to routines or rituals—In typical development, ritualistic habits and compulsions have been documented to increase between the ages of 12 and 24 months, but decrease after 48 months (Evans et al., 1997). Early in development, children with ASD show an "insistence on sameness," or a desire for repetition (Leekam et al., 2007), which can cause significant distress when rituals/routines are disrupted. This symptom can manifest in the following ways: extreme distress at small changes, challenges with transitions, and the need to follow the same routine every day (American Psychiatric Association, 2013).

Preoccupations and restricted interests—Young children with ASD commonly have narrow interests that occupy their focus and attention. Restricted interests often seen in young children with ASD include trains, cars, dinosaurs, mechanical objects (Porter, 2012), and balls. Zwaigenbaum et al. (2005) found that high-risk infant siblings show delayed disengagement of visual attention, sometimes referred to as "sticky attention," which has been found to predict later social-communicative impairment. In addition, it has been hypothesized that this "sticky attention" may be an early precursor to preoccupations or restricted interests (Stronach & Wetherby, 2014). However, recent studies using eye tracking have not found problems in disengagement (Fisher et al., 2015). Early intervention services for children with ASD often address the ability to be redirected and the skill of flexibility to maximize age-appropriate learning opportunities (Boyd, McDonough, & Bodfish, 2012).

Sensory behaviors—Atypical sensory-oriented behaviors also have been found to be an early identifier of ASD. Analyzing typical versus atypical behaviors during a free-play

session with 12-month-old, high- and low-risk infants, Ozonoff et al. (2008) found that children diagnosed with ASD at 24 or 36 months displayed at least one atypical behavior (most frequently, unusual visual examination/inspection of objects, including peering at objects from unusual angles and/or focusing on parts of objects rather than the whole object) that was more than 2 standard deviations above the mean of a group of children with a typical outcome. Zwaigenbaum et al. (2005) found that parents reported that infants with a 24-month diagnosis of ASD showed more frequent and intense distress reactions to a variety of stimuli at 12 months than did other high- or low-risk infants (i.e., sensory overresponsivity). Similarly, parents have reported increased sensitivity to sensory input in 6-month-old infants who go on to receive an ASD diagnosis at 36 months, as compared to infants with typical development (Clifford et al., 2013). Unusual sensory responses commonly seen in young children with ASD include covering ears when there is a moderately loud noise; repeated rubbing, licking, and/or mouthing of unusual objects such as wooden blocks or sandpaper; not appearing to be bothered by very loud noises or painful experiences such as hitting one's head on the bottom of a table; and engaging in repeated self-injury (DiGenarro Reed et al., 2012).

EA-ASD

Many 12-month-old children who do not fully meet *DSM-5*/*DC:0–5* criteria for ASD have begun to evidence high risk for ASD through the manifestation of many of the requisite symptoms which cause functional impairments. Infants and toddlers who are exhibiting signs of ASD without meeting full criteria need to be monitored closely over time, as ASD symptoms typically emerge gradually from 9 to 42 months (e.g., Kim et al., 2015; Landa et al., 2007; Ozonoff et al., 2015; Sacrey, Bennett, & Zwaigenbaum, 2015; Shumway et al., 2011). Moreover, infants with emerging, impairing symptoms are likely to benefit from interventions designed to engage them in age-appropriate social interactions and learning opportunities. The diagnosis of EA-ASD is designed for infants and toddlers who appear to be on a developmental trajectory toward meeting full *DSM-5*/*DC:0–5* criteria for ASD, but do not meet full criteria for ASD. EA-ASD is appropriate for children who appear to be losing social-communication skills and/or are plateauing or failing to gain chronological age and/or mental age expected social-communication skills *and* are beginning to show RRBs. Thus, symptoms in both of the domains of the *DSM-5*/*DC:0–5* ASD diagnostic criteria must be present. Moreover, EA-ASD cannot be assigned if children meet or have met criteria for ASD.

Although we set a high symptom threshold and require impairment, we recognize that some children diagnosed with EA-ASD may be manifesting symptoms of ASD consistent with the broader phenotype of ASD and will not go on to develop full ASD criteria or receive a diagnosis of ASD. This is particularly likely to be the case among relatives of individuals with ASD (Ozonoff et al., 2014). We foresee that some readers may be concerned about this diagnosis resulting in an increase in unnecessary service provision. Despite the possibility that some children diagnosed with EA-ASD will not develop ASD, the requirement of functional impairment should minimize identification of children who are not in need of intervention and monitoring. Findings from the infant sibling consortium support the fact that a high percentage of infant siblings at risk for ASD who do not ultimately develop the

full disorder have significant functional impairments in communication and social interaction that warrant intervention (Charman, 2014; Messinger et al., 2013). Unfortunately, it is not clear what percentage of these children would meet criteria for EA-ASD. We hope that specifying criteria for EA-ASD will lead to greater research in this area.

As described earlier, diagnostic criteria for EA-ASD includes the manifestation of at least two social-communication symptoms and one RRB symptom as well as the presence of functional impairment. This differs from the three social-communication and two RRB symptoms required to meet diagnostic criteria for ASD, and ensures continuous monitoring and treatment of children within the window of risk who are presenting with concerning behaviors that are causing everyday impairments.

CONCLUSION

While research continues to expand our understanding of the early presentation, course, and stability of the *DSM-5*/*DC:0–5* ASD diagnosis and individual variations in the developmental trajectories of those displaying symptoms in infancy and toddlerhood, we believe that there is sufficient evidence to identify, monitor, and treat both young children who meet full criteria for ASD and those (a) who are within the window of risk for developing ASD; (b) present with many symptoms of ASD, but do not meet full criteria for ASD; and (c) evidence functional impairment. Thus, the *DC:0–5* alternative classification system includes *both* a developmentally specified ASD diagnosis that is aligned with the *DSM-5* as well as the new diagnosis of EA-ASD. EA-ASD is intended to identify children 9 to 36 months of age (who do not and have never met full criteria for ASD) and are presenting with a minimum of two social-communication deficits and one RRB as well as evidence of impairment, with the intention of providing these children with appropriately tailored services and improving the likelihood of optimizing their development. Although some children who receive a diagnosis of EA-ASD may not go on to develop ASD and may represent presentations consistent with the broader phenotype (Ozonoff et al., 2014), the requirement of impairment is included to focus on children in need of intervention services. By aligning the *DC:0–5* diagnosis of ASD with the *DSM-5* diagnosis of ASD and including EA-ASD for subthreshold cases with impairment, we hope to improve communication across interdisciplinary providers and to ensure a mechanism for reimbursement for continued monitoring and appropriate services.

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TABLE 1

Developmentally Salient Behavioral Manifestations of Specific Social-Communication (SC) and Restricted/Repetitive Behavior (RBB) Symptoms

Domain Criteria for SC and RRB Symptoms		
Symptom	**Infant/Toddler Presentation**	**Preschool Presentation***
SC1: Limited or Atypical Socioemotional Responsivity, Sustained Social Attention, and/or Social Reciprocity as Evidenced by at Least One of the Following:		
Atypical social approach	• Shows limited social initiation (e.g., only initiates social interaction to get help or make requests) • Shows unusual social approach (e.g., walks backward and leans on caregiver without eye contact) • Uses adult's hand as a tool to complete a goal or uses adult's body for comfort without checking in/social referencing	• Intrusively touches another person to get their attention • Rarely approaches adult caregivers or peers • Uses routinized or "scripted" social approach (e.g., child uses the same greeting, mannerisms, or questions when approaching familiar or unfamiliar caregivers or peers)
Reduced or limited ability to engage in reciprocal social games or activities that require taking turns	• Shows reduced or lack of interest in turn-taking games (e.g., peek-a-boo, rolling a ball back and forth) • Only engages in reciprocal games that are embedded in familiar routines (e.g., child will only play peek-a-boo with a specific blanket) • Requires repeated prompting to take his or her turn	• Demonstrates reduced or lack of interest in turn-taking games (e.g., simple board games, hide and seek) • Makes limited requests for a turn in a social game or with a desired toy • Shows difficulty with back-and-forth conversation (e.g., may interrupt or walk away while someone is asking a question or elaborating on an answer)
Reduced or limited ability to initiate joint attention to share interests or emotions or seek information about objects of interest in the environment	• Exhibits reduced or lack of showing or bringing objects of interest to others • Shows reduced or lack of initiation of joint attention (e.g., child may reach for, but not direct an adult's attention to, a desired, out-of-reach object by using strategies such as pointing and eye contact) • Shows reduced distal pointing with coordinated eye contact to indicate interests	• Shows reduced asking of questions to learn about items of interest • Often persists in solving problems independently even when clearly frustrated or stuck, will not seek help from others • Rarely shares enthusiasm about accomplishments
Infrequent or restricted responses to social interaction	• Shows limited response when adult tries to get his or her attention (e.g., avoids others' efforts to make eye contact) • Rarely smiles in response to being smiled at • Rarely responds to name being called • Sometimes appears not to hear	• Shows limited response to praise (except in the context of familiar routines) • Shows limited or lack of shared enjoyment in social interactions • Shows limited awareness of others' affective response (e.g., may talk for an extended period of time about a special interest regardless of conversational partner's level of interest)
Rare and/or restricted, or lack of, initiation of social interaction	• Prefers to play alone • Rarely tries to engage others in his or her play (e.g., by showing or	• Rarely tries to engage others in his or her play (e.g., by sharing toys, asking to join in an activity) • Uses unusual routinized or limited strategies for initiating social

Domain Criteria for SC and RRB Symptoms		
Symptom	**Infant/Toddler Presentation**	**Preschool Presentation***
	bringing objects of interest to others) • Sometimes enjoys activities with others, but the focus is on the toy/activity rather than the person in the social interaction (e.g., enjoying blowing bubbles or rolling cars back and forth)	interaction (e.g., approaching others without eye contact and using very few of their available words to communicate)
SC2: Deficits in Nonverbal Social-Communication Behaviors as Evidenced by at Least One of the Following:		
Difficulties understanding or using nonverbal communication	• Shows difficulty understanding gestures and facial expressions (e.g., head nodding/shaking) • Uses limited nonverbal communication strategies like gestures (e.g., waving) and facial expressions • May not follow gaze or pointing	• May not notice when others are distressed or excited unless given verbal information • Shows limited ability to understand others' intentionally communicative body language and facial expressions (e.g., a mother's facial expression communicating that a child has done something wrong) • Rarely understands sarcasm (older preschoolers only)
Lack of or restricted integration of nonverbal and verbal behaviors	• Uses one communication strategy at a time (e.g., will point to something out of reach without coordinating eye contact or vocalization) • Uses a limited range of gestures	• Uses gestures inflexibly and rarely integrates eye contact and/or verbalizations • Shows limited use of conventional gestures (e.g., shrugging, head nodding/shaking, counting on fingers, showing age on fingers)
Atypical use of eye contact or turning away from others in social contexts	• Uses limited eye contact during play • Uses limited eye contact while making requests • Avoids eye contact in social interactions	• Uses limited eye contact in conversation or when seeking information • Shows delayed eye contact when asking questions • May show improved eye contact when highly motivated (e.g., may make eye contact when asking for favorite food/toy/activity)
Restricted range of facial expressions and limited nonverbal communication	• Uses reduced facial expressions to express emotions • Only directs extreme/exaggerated facial expressions to others (e.g., only when very happy or very sad)	• Does not use facial expressions to communicate needs, wants, or desires • May appear muted or "flat"
SC3: Peer-Interaction Difficulties as Evidenced by at Least One of the Following:		
Problems adapting behavior to accommodate varying social demands across social contexts	• Shows difficulty modifying his or her behavior across social contexts (e.g., has trouble sharing toys) • Behaves in overly familiar ways with strangers	• Shows difficulty changing behavior to fit various social contexts (e.g., may laugh/smile inappropriately) • Asks inappropriate questions • Shows limited understanding that rules vary by context (e.g., home, school, public settings)

Domain Criteria for SC and RRB Symptoms		
Symptom	**Infant/Toddler Presentation**	**Preschool Presentation***
Difficulties engaging in pretend or imaginative play	• Shows reduced or lack of pretend play routines (e.g., does not hug baby doll) • Uses pretend play materials for other purposes (e.g., plates and utensils are stacked or rolled)	• Shows limited imaginative play skills (e.g., is unsure what to do when peers suggest playing "house") • Prefers to play with cause and effect or construction toys
Limited or lack of interest in peers and in playing with other children	• Prefers to play alone instead of with other same-age children • Exhibits lack of interest in babies or same-age children	• Prefers to play alone • Shows limited interest in making friends • Is unaware of or indifferent to same-age peers • Does not comfort peers when hurt
Two of the Following Four Repetitive and Restricted Behaviors Must Be Present:		
Stereotyped or repetitive babbling or speech (including echolalia), motor movements, or use of objects and toys	• Repeats sounds over and over (e.g., "dadada") with the same intonation pattern • Immediately echoes another's speech with the same intonation pattern • Flaps arms, flicks fingers, or engages in other complex body postures • Plays repetitively with toys or objects	• Repeats the same words, phrases, or sentences over and over (e.g., media commercial) with the same intonation pattern • Repeats a question that is asked verbatim
Rigidly maintains routines with excessive resistance to change; demands sameness and shows distress in response to change; or ritualized use of stereotyped, odd, or idiosyncratic verbal phrases, or nonverbal behaviors	• Shows difficulty with transitions between activities • Gets upset and cannot be easily redirected when focused on a toy or activity • Uses odd words to communicate (e.g., says "moy" for "more" even though no articulation problem is present)	• Insists on adherence to specific multistep routines and shows distress in response to change • Uses overly formal or pedantic language (e.g., refers to self by own name) • Uses unusual phrases to communicate (e.g., consistent use of a phrase from a television show for a specific purpose)
Highly circumscribed, specific interests that manifest in extreme fixation on an item or topic of interest	• Shows unusually strong interest in a specific object (e.g., toy train, car) and shows significant distress when it is removed	• Shows unusually strong interest in specific topics (e.g., historical dates, dinosaurs, train schedules, weather, nature) and shows significant distress when topic is no longer discussed
Atypical responsivity to sensory inputs (either over- or underresponsive) or unusual engagement with sensory aspects of the environment	• Seeks sensory inputs (e.g., holds objects very close to ears, visually examines objects out of the corner of eye, sniffs or licks objects) • Shows strong adverse reactions to sensory input (e.g., covers ears during loud sounds)	• Refuses to wear certain materials or clothing styles (e.g., refuses to wear jeans or belt) • Is bothered by being in certain types of motion (e.g., swinging, sliding)

*Behavior examples listed for infancy/toddlerhood often manifest in the preschool period.