Language Disorders: Autism and Other Pervasive Developmental Disorders

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Autism spectrum disorders (ASD) are increasingly common neurodevelopmental disorders that encompass a range of clinical presentations characterized by functional impairments in a triad of symptoms: (1) limited reciprocal social interactions, (2) disordered verbal and nonverbal communication, and (3) restricted, repetitive behaviors or circumscribed interests\cite{1}. Included under the umbrella of the autism spectrum are the disorders defined in the \textit{Diagnostic and Statistical Manual of Mental Disorders, fourth edition, text revised (DSM-IV TR)} as pervasive developmental disorders (PDD): autistic disorder, Asperger’s disorder, and pervasive developmental disorder, not otherwise specified (PDD-NOS). Rett’s disorder and childhood disintegrative disorder also appear in the \textit{DSM} classification under the rubric of PDD, but they usually are considered to be distinct from the autism spectrum. The terminology used for ASD can be confusing, because clinicians and families may use different terms to describe the same clinical entities, often using the terms “autism,” “autism spectrum disorder,” and “PDD” interchangeably. In this article, the term “ASD” is used to include autistic disorder, Asperger’s disorder, and PDD-NOS.

Young children who have autistic disorder generally exhibit marked impairments in all three domains of the triad before the age of 3 years. Often these children are referred for evaluation around the age of 2 years because of language delay, lack of interest in social contact with children or adults, and atypical, perseverative play (eg, focusing on spinning wheels, flashing lights, or non-toy objects such as pieces of thread), although there is

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significant variability in the specific symptoms and degree of severity. Lan-
guage abilities may range from being nonverbal to developing language that
is highly idiosyncratic with echolalia, scripted speech, and unusual prosody
(tone or inflection). At least half of all children who have autism have men-
tal retardation; those who do not have nonverbal cognitive delays are con-
sidered to be high functioning even though they may have significant
impairments in adaptive functioning and language or communication.

Much less common than the other forms of ASD, Asperger’s disorder is
sometimes incorrectly thought of as a mild form of autism. The degree of
functional impairment in social interactions of affected individuals is vari-
able but may be quite profound [2]. The diagnostic category itself has
been controversial, and experts disagree on whether it should be considered
a distinct disorder from autism [3]. As defined in *DSM-IV-TR* [4], early lan-
guage development in Asperger’s disorder is not delayed, (although the
*DSM*’s definition of normal language development of “single words used
by age 2 years, communicative phrases used by age 3 years” would, in gen-
eral, be considered moderately delayed), and intellectual abilities are at least
average but can be superior. Social interactions, including use of eye gaze
and body postures, development of friendships, and social reciprocity, are
atypical. Intense, restricted, or all-encompassing interests are common and
may be developmentally appropriate in content (eg, dinosaurs) or quite un-
usual (eg, the intricacies of a municipal recycling program). Most children
who have Asperger’s disorder are not identified until early school age,
when social difficulties with other children become impossible to ignore or
to explain away as mere quirkiness. Language skills, including articulation,
vocabulary, and grammatical abilities, may be preserved, although social or
pragmatic aspects of language, such as the ability to engage in the give-and-
take of social discourse, are impaired [5]. These children have been described
as “little professors” who may use advanced vocabulary, speak in mono-
logues or with a pedantic style, have difficulty with abstract or nonliteral
language, and have unusual prosody [6–8].

PDD-NOS is a term for individuals who show symptoms of ASD but do
not meet full *DSM* criteria for autistic disorder or Asperger’s disorder be-
cause of atypical or subthreshold symptoms or later age of onset. Changes
in the text revision of *DSM-IV* specified that this diagnosis is reserved for
individuals with, at minimum, impairment in reciprocal interactions with
language impairment and/or restricted interests or behaviors [4]. As a result,
it is a catchall diagnosis including children with a wide range of language
skills, cognitive abilities, and levels of functional impairment.

**Social communication and language deficits in children who have autistic
spectrum disorder**

Because of the increasingly broad nature of symptoms that now are
included under the autism spectrum, language deficits in ASD vary
dramatically across the different diagnoses and also within a single diagnostic category. Most, but not all, children who have ASD have receptive and expressive language impairments. As is the case with many developmental disorders (eg, specific language impairment), the prevalence of ASD is higher in boys than in girls. For autism, the ratio is about 3 or 4:1, but this ratio approaches about 10:1 for Asperger’s disorder [1]. Unique deficits in social or pragmatic aspects of communication distinguish the communication impairments in ASD from other developmental disorders [9]. These impairments manifest differently in children of different developmental levels and across the autism spectrum. Thus, evaluations of children for ASD always must include assessments of language and social communication abilities in light of the child’s cognitive level [10]. Early and appropriate diagnosis of ASD in young children requires knowledge of typical developmental milestones of language development and social communication, especially preverbal communication. Research on very young children who have ASD has focused on impairments of joint attention (the sharing of an experience, affect, or intention with another person through coordination of gaze and gesture or vocalization) as an important early sign of the social communication deficits in ASD [11]. Current guidelines and available screening tools for ASD in the first 2 years of life tap these early nonverbal communicative skills that involve joint attention deficits and apparent lack of receptive language skills [12]. The milder the early symptoms of ASD are, the more subtle the deficits in social communication may be, delaying diagnosis.

Language in toddlers and preschoolers

Typically, developing infants are competent communicators well before they speak their first words. The development of joint attention and the sharing of experiences during the first year of life are critical prerequisites for more complex forms of social communication [13]. By the age of 9 to 12 months, infants develop gaze-monitoring and social-referencing skills, or the ability to observe others’ focus of attention or affect by shifting gaze between people and objects [14]. This ability is seen, for example, when a baby “reads” her parent’s face in a pediatrician’s office to look for an indication of whether this strange adult is safe. In the same period, infants acquire understanding of gestures, single words, and phrases, initially in the context of social games or routines. They start using simple gestures or vocalizations to communicate requests or comment by reaching, pushing away, calling out, or waving. Around the first birthday, babies can respond to joint attention by following another’s point to look at the object indicated, point to indicate an object, and later point so show or share an experience with another person [15,16].

Development of language comprehension and expression follow nonverbal precursors to spoken language. By the first birthday, most children say
their first words and can understand many more words and some phrases. Initially meaning is linked to context, and children learn names of objects only in a single setting [17]. Between 12 and 18 months of age, there is a gradual growth in receptive and expressive vocabulary, with increasing freedom from context, and sometimes overgeneralization of words that are known (eg, any gray-haired woman is “grandma”). Around 18 months, they can “read” others’ communicative intentions through eye gaze and gestures [18]. From 18 to 24 months, children have an explosive increase in vocabulary and an understanding of communicative norms such as the back-and-forth of a conversational exchange [19]. During the second year of life, in concert with increasing complexity of communication skills, toddlers have more complex play skills, advancing from constructive to functional to imaginary play, imitating actions they have seen and using them in the context of play. By the second birthday, most children have hundreds of words that are not tied to use in specific context and start putting together simple phrases or “sentences” of two or three words [20].

This predictable developmental progression goes awry in ASD. Most toddlers who have ASD have delays in acquiring language and significantly decreased vocal output [21]. Acquisition of language is slower than in other children who have language delays, often is related directly to cognitive level, but may lag behind development in other areas [22]. Children who have ASD typically use words to label, request, or protest, as a way of regulating their environment, rather than for purely social reasons, such as to comment or to initiate a social interaction [23]. Some children remain non-verbal, although as more children are identified early and receive intensive early intervention from the time of diagnosis, that number seems to be dropping [9]. It is unclear whether some of the improvement in outcome may result from diagnosis of children who have milder symptoms from the outset. Some children who have ASD and who never acquire spoken language may also have apraxia, or oral-motor impairment, impacting their ability to communicate verbally [9]. Usually, receptive language also is impaired in ASD and often seems to lag behind expressive language, although this unusual profile may reflect difficulties in testing young children’s comprehension because of their lack of social responsiveness [24]. Children who have stronger language comprehension tend to have more advanced play skills and better comprehension of social interactions [25].

Babbling and other vocalizations that are present are often unusual in tone, including repetitive screeching, groaning, humming, “raspberries,” or echolalia. Echolalia can be immediate or delayed, and most children who have ASD and who speak use echolalia early in language acquisition, but its frequency decreases with time. Some typically developing children also have transient immediate echolalia as a means of learning and consolidating vocabulary, and it sometimes is observed in children who are language delayed or blind [26]. Immediate echolalia can consist of the final word of another person’s sentence or a complete sentence, demonstrating
the characteristic pronoun reversal often seen in, although not unique to, autism (eg, a child’s saying, “You want juice?” after being asked if he wants juice). Delayed echolalia can be taken from video, books, or past conversations (eg, the child’s finding an adult and saying, “Are you sleepy?” to indicate he wants to nap). It is not uncommon for a child who has ASD with little or no spontaneous language to repeat commercials or large chunks or dialogue from movies. In clinical practice, parents sometimes describe “imaginary” play of their child who has autism, which consists of using toys to act out dialogue repeated verbatim from a previously watched video. Echolalic phrases may be complex but usually are uttered as a chunk, as if a single word. Although echolalia in children who have ASD seems to be a vocal stereotypy or self-stimulatory behavior, it sometimes is functional, allowing children to make requests, self-soothe, participate in a social routine or interaction, or gain time to process language [27].

Up to a quarter of children who have ASD have regression of language between the age of 12 and 18 months. In most of these children there is not a dramatic loss of language in the midst of normal language development. Usually the child uses single words inconsistently, and they gradually disappear [28]. At the same time, parents often report social withdrawal and constriction of affect or change in temperament. Regression of language and social relatedness of this sort is unique to autism and should be responded to promptly in the pediatric setting. Regression of motor or other streams of development should raise concerns of other disorders, such as childhood disintegrative disorder, neurodegenerative or metabolic disorders, or seizure disorders such as Landau-Kleffner syndrome.

Young children who have ASD demonstrate reduced use of the nonverbal communicative behaviors that precede spoken language in typically developing children. Affected children use fewer gestures, show decreased use of gaze to indicate and interpret meaning, and do not initiate or respond to bids for joint attention. Unlike other children who have language delays, children who have ASD do not compensate for their lack of speech with gestures [21]. Rather than using symbolic gestures like pointing or waving, affected children may use gestures associated with physical contact such as leading, pushing, or moving another’s hand to a desired object. Problem behaviors such as aggression, self-injury, and tantrums can serve communicative functions in children on the spectrum.

Impairments in joint attention are now part of the DSM criteria for autism (“lack of showing, bringing, or pointing out objects of interest”). Individuals who have ASD rarely communicate for purely social reasons or enjoyment. Young children who have ASD do not show the usual precursors to joint attention, such as social referencing, sharing affect, following the gaze of another, or pointing [29]. Children who have ASD often are described by parents as being “in their own world” and do not attend to voices around them, although they usually respond to other nonvocal auditory stimuli. Typical children learn words by hearing them used in a social
context, which requires joint attention. When young children not respond to
social stimuli, they miss vital learning opportunities for developing lan-
guage. When they do not monitor others’ gaze, they may make incorrect
associations between objects and words [30]. Thus impairment in joint atten-
tion may be a primary deficit that leads to delays in language.

Play in autism is atypical, usually perseverative, and lacking in imagina-
tive themes. Several studies have found that children who have autism also
have impairments in imitation (eg, Ref. [31]). Typical children learn socially
by observing and imitating what they see around them, developing symbolic
play that later is elaborated into imaginary play. Young children on the au-
tistic spectrum demonstrate more solitary play including sensory explora-
tion of toys and other objects, constructive play (eg, lining up or
stacking), and trial-and-error learning of how things work. Higher levels
of play in ASD correlate with expressive language level, suggesting that
these children may use language skills to mediate play skills [32].

Language in school-aged children and beyond

By the time typical children enter school, they are able to speak fluently,
have acquired a rich vocabulary, and use full sentences. In the early school
years, children master the more complex grammatical structures of their na-
tive language; however, vocabulary continues to grow throughout the life-
span. Pragmatic and discourse skills continue to develop as children
become more effective communicators, becoming more sensitive to their lis-
tener’s perspective and telling more complex and well-structured narratives
[33].

Many children who have ASD still have very limited language by the time
they enter kindergarten, and their impairments in nonverbal communication
also persist [34]. These deficits in social communication are a significant bar-
rier to learning and may lead to increased problem behaviors. At this stage it
is quite common to introduce minimally verbal children who have ASD to
alternative communication systems such as manual signs or the Picture Ex-
change Communication System [35]. It is crucial to focus on developing lan-
guage skills in young children who have ASD because the presence of speech
before age 5 years is the strongest predictor for better outcomes [36].

There is considerable variability in the rate at which language progresses
among verbal children who have ASD. Children with higher levels of IQ,
receptive language, imitation, and joint attention skills tend to make greater
gains [11]. In general, verbal children who have ASD do not have problems
with articulating speech sounds [37]. They also can score quite highly on
tests of vocabulary knowledge, although they may not understand or use
words referring to emotions, thoughts, and other mental states [38]. Some-
times children or adults will use idiosyncratic words or phrases or made-up
words (eg, “cuts and bluesers”), and their speech might be quite persevera-
tive [39]. With respect to grammatical knowledge, there are different
subgroups, with some children achieving average or above-average scores on standardized tests (about 25% of verbal children who have ASD), but the majority remains delayed [37]. Like children who have specific language impairment, children who have ASD who have impaired language have particular difficulty mastering grammatical morphology, especially for marking tense (eg, using “-ed” to construct the past tense, as in “John painted the house”) and related complex syntactic structures [40].

Compared with other groups of children who have language impairment or mental retardation, the receptive language skills of children who have ASD seem to be relatively lower than their expressive skills [41]. Part of their difficulties in understanding language stem from limitations in the ability to integrate linguistic input with real-world knowledge, which may include their impaired understanding of the social world [25]. Another source of difficulty is the use of different types of cues to decipher the intended meaning of another person’s message. For example, children who have ASD have core impairments integrating nonverbal cues to help interpret verbal messages, especially in everyday social interactions [42]. Thus, they may not use facial expressions, body language, or intonation to determine whether a speaker’s intended message is affectionate, hostile, or teasing. Elliptical utterances, indirect requests (eg, “Can you take the garbage out?”) and non-literal language (eg, lies or ironic jokes) all depend on the ability to interpret intended meaning and thus may contribute to the overall comprehension difficulties experienced by children and adults who have ASD [43].

The speech of children and adults who have ASD usually sounds odd or unusual, and this oddity is one of the immediately recognizable clinical signs of the disorder. Defining the specific abnormalities so that clinicians could make reliable judgments has been quite challenging, however, perhaps because there are many different ways in which their language sounds peculiar. These abnormalities in intonation may be even more prevalent among people who have Asperger’s disorder and include flat, monotonic, or sing-song speech, nasal or high-pitched vocal tone, lack of affective quality, poor control of volume, and atypical stress patterns in words and sentences [44]. Problems with intonation are found in both expressive and receptive language: children who have ASD have difficulty distinguishing different stress patterns or interpreting emotional prosody [45].

At the heart of the language problems found in everyone who has ASD are difficulties in the area of language pragmatics, the ability to use language effectively in a variety of social contexts [9]. Children who have ASD use language in limited ways, rarely to comment, request information, acknowledge their listener, or to describe events [46]. They may fail to follow politeness rules, make irrelevant remarks, and in conversations with other people have problems taking turns and may talk either too much or too little [7]. When asked to narrate events from their lives or stories, children who have autism often include irrelevant or inappropriate content and have difficulty taking into account their listener’s needs (eg, by failing to establish
clear reference or by presenting events in a confused or disorganized way) [47]. These pragmatic problems seriously impede the social adaptation of both children and adults who have ASD and can lead to disruptive behavior in the classroom, on the playground, or in employment situations.

Clinical implications and recommendations

Although many parents have attributed their children’s onset of symptoms to immunizations at the age of 15 months (cf. [48]), retrospective analyses of first birthday party home videotapes have shown signs of impairment of social relatedness and communication before that time [49–51]. Usually there is no period of unequivocally normal development, although abnormalities may not be noted unless regression occurs or language skills lag far behind peers, usually between 18 and 24 months [52]. Numerous studies have shown significant delays from first parental concern that “something is wrong,” to referral to a specialist for evaluation, to diagnosis [53]. With improved awareness in both lay and professional communities of the early signs of autism, the interval between recognition of symptoms and diagnosis is dropping. Despite this improvement, several recent studies have shown disparities in age of diagnosis based on socioeconomic factors including ethnicity, rural residence, and income. [54] This disparity has important clinical implications, because there is ample evidence that early and intensive therapy in young children who have ASD positively impacts outcomes in language and cognition in many children [55]. Parent advocacy groups have been promoting a sense of urgency for early diagnosis of autism because of the promise of better outcomes with treatment. Routine pediatric visits are the most appropriate place for identification of early signs of ASD. It is critical for pediatric providers to be familiar with the earliest signs of impaired social communication in ASD and with screening recommendations.

A number of different studies have identified some of the earliest observable signs of ASD [56]. Not surprisingly, the findings are most robust in children who are most severely affected, who can be diagnosed earliest. The retrospective studies of parental report and home video studies mentioned previously showed decreased social interactions, less social smiling, decreased range of facial expressions, lack of response to name, decreased pointing and showing, fewer vocalizations, decreased orientation to faces, and decreased imitation [49–51]. A number of screening tools have been developed to identify ASD before 24 months. These tools include many of these early signs and have found that they discriminate between ASD and other developmental disorders, such as language or cognitive impairments [57–60]. More recently, a number of centers have initiated longitudinal studies of infants at high risk for ASD, younger siblings of children already diagnosed, who have an approximately 10% risk or higher of being on the autism spectrum. To date, initial reports from those studies have shown differences at 12 months in children later diagnosed as having ASD in the
following behavior patterns: decreased receptive language; use of fewer gestures; atypical eye contact, visual tracking, and visual attention; impaired orientation to name; decreased imitation; decreased social smiling and social interest; and temperamental differences [61,62]. Interestingly, siblings who were not later diagnosed as having ASD also used fewer gestures than controls [21].

Prospective research is revealing signs of ASD at the first birthday, and screening guidelines are beginning to reflect these advances. The challenge for pediatric providers lies in distinguishing children for whom language delay is a sign of the late talker from one whose language delay requires remediation. In children who require further assessment and treatment, language delay may be a symptom of developmental language disorder, global developmental delay, or ASD. Screening guidelines and tools point to ways in which these distinctions can be made in the primary care setting. The American Academy of Neurology and the Child Neurology Society published a practice parameter for screening and diagnosis of autism, including red flags requiring immediate referral for further evaluation that focus on expressive and nonverbal communication, joint attention, and regression: no babbling, or pointing, or other gestures by 12 months; no single words by 16 months; no two-word spontaneous (not echolalic) phrases at 24 months; and any loss of any language or social skills at any age [63].

The most recent guidelines for screening for developmental disorders in the pediatric setting were published by the American Academy of Pediatrics [64]. Although the algorithm is for general developmental screening, ASD figures prominently. The recommendations include routine developmental surveillance as part of every well-child visit and administration of formal screening tests during three visits in the first 3 years of life. The first formal screening is at the 9-month visit, when the provider is urged to evaluate nonverbal communication for early symptoms of autism, such as decreased eye contact, response to name, and pointing. At the 18-month visit, an autism-specific screening tool is recommended, because more general screening tools have not been found to have adequate sensitivity and specificity to identify ASD. Proposed federal legislation would mandate universal screening for autism, potentially transforming this algorithm into a standard of care.

Pediatricians are increasingly expected to recognize the subtle, early signs of ASD, before language delay is evident, and to respond quickly with appropriate referrals. Pediatricians should familiarize themselves with screening tools designed specifically to identify autism and should know how to refer parents to community resources, including specialists with expertise in ASD and early intervention programs. In addition, it is vital to have a particularly high index of suspicion in siblings of children who have ASD, especially those who have language delay, because their risk for the disorder is many times that of the general population.

Because children who have ASD face new challenges with each transition they make, first into preschool programs and then into elementary school, it
is important to continue to monitor their progress to ensure that their behavioral and language needs are being addressed in their educational programs [65]. Most children who have ASD need to have their language skills assessed on a regular basis to evaluate their receptive and expressive abilities and also, importantly, their pragmatic skills. It is important to recognize that ASDs are lifelong disorders, and although significant gains in language can be expected, especially during early childhood, difficulties in effective communication that are closely tied to their core social deficits continue to require close monitoring and referrals for comprehensive evaluation and treatment.

References


