



# Language Disorders: Autism and Other Pervasive Developmental Disorders

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[Q2] [Q3]

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[Q4]

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[1]. Included under the umbrella of the autism spectrum are the [Q5] disorders defined in the *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 *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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39 significant variability in the specific symptoms and degree of severity. Lan-  
40 guage abilities may range from being nonverbal to developing language that  
41 is highly idiosyncratic with echolalia, scripted speech, and unusual prosody  
42 (tone or inflection). At least half of all children who have autism have men-  
43 tal retardation; those who do not have nonverbal cognitive delays are con-  
44 sidered to be high functioning even though they may have significant  
45 impairments in adaptive functioning and language or communication.

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

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

### 78 79 **Social communication and language deficits in children who have autistic** 80 **spectrum disorder**

[Q6]

81  
82 Because of the increasingly broad nature of symptoms that now are  
83 included under the autism spectrum, language deficits in ASD vary

84 dramatically across the different diagnoses and also within a single diagnos-  
85 tic category. Most, but not all, children who have ASD have receptive and  
86 expressive language impairments. As is the case with many developmental  
87 disorders (eg, specific language impairment), the prevalence of ASD is  
88 higher in boys than in girls. For autism, the ratio is about 3 or 4:1, but  
89 this ratio approaches about 10:1 for Asperger's disorder [1]. Unique deficits  
90 in social or pragmatic aspects of communication distinguish the communi-  
91 cation impairments in ASD from other developmental disorders [9]. These  
92 impairments manifest differently in children of different developmental  
93 levels and across the autism spectrum. Thus, evaluations of children for  
94 ASD always must include assessments of language and social communica-  
95 tion abilities in light of the child's cognitive level [10]. Early and appropriate  
96 diagnosis of ASD in young children requires knowledge of typical develop-  
97 mental milestones of language development and social communication, espe-  
98 cially preverbal communication. Research on very young children who  
99 have ASD has focused on impairments of joint attention (the sharing of  
100 an experience, affect, or intention with another person through coordination  
101 of gaze and gesture or vocalization) as an important early sign of the social  
102 communication deficits in ASD [11]. Current guidelines and available  
103 screening tools for ASD in the first 2 years of life tap these early nonverbal  
104 communicative skills that involve joint attention deficits and apparent lack  
105 of receptive language skills [12]. The milder the early symptoms of ASD are,  
106 the more subtle the deficits in social communication may be, delaying  
107 diagnosis.

### 109 **Language in toddlers and preschoolers**

111 Typically, developing infants are competent communicators well before  
112 they speak their first words. The development of joint attention and the  
113 sharing of experiences during the first year of life are critical prerequisites  
114 for more complex forms of social communication [13]. By the age of 9 to  
115 12 months, infants develop gaze-monitoring and social-referencing skills,  
116 or the ability to observe others' focus of attention or affect by shifting  
117 gaze between people and objects [14]. This ability is seen, for example,  
118 when a baby "reads" her parent's face in a pediatrician's office to look  
119 for an indication of whether this strange adult is safe. In the same period,  
120 infants acquire understanding of gestures, single words, and phrases, ini-  
121 tially in the context of social games or routines. They start using simple ges-  
122 tures or vocalizations to communicate requests or comment by reaching,  
123 pushing away, calling out, or waving. Around the first birthday, babies  
124 can respond to joint attention by following another's point to look at the  
125 object indicated, point to indicate an object, and later point so show or share  
126 an experience with another person [15,16].

127 Development of language comprehension and expression follow nonver-  
128 bal precursors to spoken language. By the first birthday, most children say

129 their first words and can understand many more words and some phrases.  
130 Initially meaning is linked to context, and children learn names of objects  
131 only in a single setting [17]. Between 12 and 18 months of age, there is a gradual  
132 growth in receptive and expressive vocabulary, with increasing freedom  
133 from context, and sometimes overgeneralization of words that are known  
134 (eg, any gray-haired woman is “grandma”). Around 18 months, they can  
135 “read” others’ communicative intentions through eye gaze and gestures  
136 [18]. From 18 to 24 months, children have an explosive increase in vocabulary  
137 and an understanding of communicative norms such as the back-and-forth  
138 of a conversational exchange [19]. During the second year of life, in  
139 concert with increasing complexity of communication skills, toddlers have  
140 more complex play skills, advancing from constructive to functional to  
141 imaginary play, imitating actions they have seen and using them in the con-  
142 text of play. By the second birthday, most children have hundreds of words  
143 that are not tied to use in specific context and start putting together simple  
144 phrases or “sentences” of two or three words [20].

145 This predictable developmental progression goes awry in ASD. Most tod-  
146 dlers who have ASD have delays in acquiring language and significantly de-  
147 creased vocal output [21]. Acquisition of language is slower than in other  
148 children who have language delays, often is related directly to cognitive  
149 level, but may lag behind development in other areas [22]. Children who  
150 have ASD typically use words to label, request, or protest, as a way of reg-  
151 ulating their environment, rather than for purely social reasons, such as to  
152 comment or to initiate a social interaction [23]. Some children remain non-  
153 verbal, although as more children are identified early and receive intensive  
154 early intervention from the time of diagnosis, that number seems to be drop-  
155 ping [9]. It is unclear whether some of the improvement in outcome may re-  
156 sult from diagnosis of children who have milder symptoms from the outset.  
157 Some children who have ASD and who never acquire spoken language may  
158 also have apraxia, or oral-motor impairment, impacting their ability to com-  
159 municate verbally [9]. Usually, receptive language also is impaired in ASD  
160 and often seems to lag behind expressive language, although this unusual  
161 profile may reflect difficulties in testing young children’s comprehension be-  
162 cause of their lack of social responsiveness [24]. Children who have stronger  
163 language comprehension tend to have more advanced play skills and better  
164 comprehension of social interactions [25].

165 Babbling and other vocalizations that are present are often unusual in  
166 tone, including repetitive screeching, groaning, humming, “raspberries,”  
167 or echolalia. Echolalia can be immediate or delayed, and most children  
168 who have ASD and who speak use echolalia early in language acquisition,  
169 but its frequency decreases with time. Some typically developing children  
170 also have transient immediate echolalia as a means of learning and consol-  
171 idating vocabulary, and it sometimes is observed in children who are lan-  
172 guage delayed or blind [26]. Immediate echolalia can consist of the final  
173 word of another person’s sentence or a complete sentence, demonstrating

174 the characteristic pronoun reversal often seen in, although not unique to,  
175 autism (eg, a child's saying, "You want juice?" after being asked if he wants  
176 juice). Delayed echolalia can be taken from video, books, or past conversa-  
177 tions (eg, the child's finding an adult and saying, "Are you sleepy?" to indi-  
178 cate he wants to nap). It is not uncommon for a child who has ASD with  
179 little or no spontaneous language to repeat commercials or large chunks  
180 or dialogue from movies. In clinical practice, parents sometimes describe  
181 "imaginary" play of their child who has autism, which consists of using  
182 toys to act out dialogue repeated verbatim from a previously watched video.  
183 Echolalic phrases may be complex but usually are uttered as a chunk, as if  
184 a single word. Although echolalia in children who have ASD seems to be  
185 a vocal stereotypy or self-stimulatory behavior, it sometimes is functional,  
186 allowing children to make requests, self-soothe, participate in a social rou-  
187 tine or interaction, or gain time to process language [27].

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

199 Young children who have ASD demonstrate reduced use of the nonverbal  
200 communicative behaviors that precede spoken language in typically devel-  
201 oping children. Affected children use fewer gestures, show decreased use  
202 of gaze to indicate and interpret meaning, and do not initiate or respond to  
203 bids for joint attention. Unlike other children who have language delays,  
204 children who have ASD do not compensate for their lack of speech with ges-  
205 tures [21]. Rather than using symbolic gestures like pointing or waving, af-  
206 fected children may use gestures associated with physical contact such as  
207 leading, pushing, or moving another's hand to a desired object. Problem be-  
208 haviors such as aggression, self-injury, and tantrums can serve communica-  
209 tive functions in children on the spectrum.

210 Impairments in joint attention are now part of the *DSM* criteria for au-  
211 tism ("lack of showing, bringing, or pointing out objects of interest"). Indi-  
212 viduals who have ASD rarely communicate for purely social reasons or  
213 enjoyment. Young children who have ASD do not show the usual precurs-  
214 sors to joint attention, such as social referencing, sharing affect, following  
215 the gaze of another, or pointing [29]. Children who have ASD often are de-  
216 scribed by parents as being "in their own world" and do not attend to voices  
217 around them, although they usually respond to other nonvocal auditory  
218 stimuli. Typical children learn words by hearing them used in a social

219 context, which requires joint attention. When young children not respond to  
220 social stimuli, they miss vital learning opportunities for developing lan-  
221 guage. When they do not monitor others' gaze, they may make incorrect  
222 associations between objects and words [30]. Thus impairment in joint atten-  
223 tion may be a primary deficit that leads to delays in language.

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

234

235

### 235 **Language in school-aged children and beyond**

236

237 By the time typical children enter school, they are able to speak fluently,  
238 have acquired a rich vocabulary, and use full sentences. In the early school  
239 years, children master the more complex grammatical structures of their na-  
240 tive language; however, vocabulary continues to grow throughout the life-  
241 span. Pragmatic and discourse skills continue to develop as children  
242 become more effective communicators, becoming more sensitive to their lis-  
243 tener's perspective and telling more complex and well-structured narratives  
244 [33].

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

254 There is considerable variability in the rate at which language progresses  
255 among verbal children who have ASD. Children with higher levels of IQ,  
256 receptive language, imitation, and joint attention skills tend to make greater  
257 gains [11]. In general, verbal children who have ASD do not have problems  
258 with articulating speech sounds [37]. They also can score quite highly on  
259 tests of vocabulary knowledge, although they may not understand or use  
260 words referring to emotions, thoughts, and other mental states [38]. Some-  
261 times children or adults will use idiosyncratic words or phrases or made-up  
262 words (eg, "cuts and bluesers"), and their speech might be quite persevera-  
263 tive [39]. With respect to grammatical knowledge, there are different



264 subgroups, with some children achieving average or above-average scores  
265 on standardized tests (about 25% of verbal children who have ASD), but  
266 the majority remains delayed [37]. Like children who have specific language  
267 impairment, children who have ASD who have impaired language have partic-  
268 ular difficulty mastering grammatical morphology, especially for marking  
269 tense (eg, using “-ed” to construct the past tense, as in “John painted the  
270 house”) and related complex syntactic structures [40].

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

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

299 At the heart of the language problems found in everyone who has ASD  
300 are difficulties in the area of language pragmatics, the ability to use language  
301 effectively in a variety of social contexts [9]. Children who have ASD use  
302 language in limited ways, rarely to comment, request information, acknowl-  
303 edge their listener, or to describe events [46]. They may fail to follow polite-  
304 ness rules, make irrelevant remarks, and in conversations with other people  
305 have problems taking turns and may talk either too much or too little [7].  
306 When asked to narrate events from their lives or stories, children who  
307 have autism often include irrelevant or inappropriate content and have dif-  
308 ficulty taking into account their listener’s needs (eg, by failing to establish

309 clear reference or by presenting events in a confused or disorganized way)  
310 [47]. These pragmatic problems seriously impede the social adaptation of  
311 both children and adults who have ASD and can lead to disruptive behavior  
312 in the classroom, on the playground, or in employment situations.  
313

### 314 **Clinical implications and recommendations** 315

316 Although many parents have attributed their children's onset of symp-  
317 toms to immunizations at the age of 15 months (cf. [48]), retrospective anal-  
318 yses of first birthday party home videotapes have shown signs of impairment  
319 of social relatedness and communication before that time [49–51]. Usually  
320 there is no period of unequivocally normal development, although abnor-  
321 malities may not be noted unless regression occurs or language skills lag  
322 far behind peers, usually between 18 and 24 months [52]. Numerous studies  
323 have shown significant delays from first parental concern that “something is  
324 wrong,” to referral to a specialist for evaluation, to diagnosis [53]. With im-  
325 proved awareness in both lay and professional communities of the early  
326 signs of autism, the interval between recognition of symptoms and diagnosis  
327 is dropping. Despite this improvement, several recent studies have shown  
328 disparities in age of diagnosis based on socioeconomic factors including eth-  
329 nicity, rural residence, and income. [54] This disparity has important clinical  
330 implications, because there is ample evidence that early and intensive ther-  
331 apy in young children who have ASD positively impacts outcomes in lan-  
332 guage and cognition in many children [55]. Parent advocacy groups have  
333 been promoting a sense of urgency for early diagnosis of autism because  
334 of the promise of better outcomes with treatment. Routine pediatric visits  
335 are the most appropriate place for identification of early signs of ASD. It  
336 is critical for pediatric providers to be familiar with the earliest signs of im-  
337 paired social communication in ASD and with screening recommendations.

338 A number of different studies have identified some of the earliest observ-  
339 able signs of ASD [56]. Not surprisingly, the findings are most robust in chil-  
340 dren who are most severely affected, who can be diagnosed earliest. The  
341 retrospective studies of parental report and home video studies mentioned  
342 previously showed decreased social interactions, less social smiling, de-  
343 creased range of facial expressions, lack of response to name, decreased  
344 pointing and showing, fewer vocalizations, decreased orientation to faces,  
345 and decreased imitation [49–51]. A number of screening tools have been de-  
346 veloped to identify ASD before 24 months. These tools include many of  
347 these early signs and have found that they discriminate between ASD and  
348 other developmental disorders, such as language or cognitive impairments  
349 [57–60]. More recently, a number of centers have initiated longitudinal stud-  
350 ies of infants at high risk for ASD, younger siblings of children already di-  
351 agnosed, who have an approximately 10% risk or higher of being on the  
352 autism spectrum. To date, initial reports from those studies have shown dif-  
353 ferences at 12 months in children later diagnosed as having ASD in the



354 following behavior patterns: decreased receptive language; use of fewer ges-  
355 tures; atypical eye contact, visual tracking, and visual attention; impaired  
356 orientation to name; decreased imitation; decreased social smiling and social  
357 interest; and temperamental differences [61,62] Interestingly, siblings who  
358 were not later diagnosed as having ASD also used fewer gestures than  
359 controls [21].

360 Prospective research is revealing signs of ASD at the first birthday, and  
361 screening guidelines are beginning to reflect these advances. The challenge  
362 for pediatric providers lies in distinguishing children for whom language de-  
363 lay is a sign of the late talker from one whose language delay requires reme-  
364 diation. In children who require further assessment and treatment, language  
365 delay may be a symptom of developmental language disorder, global devel-  
366 opmental delay, or ASD. Screening guidelines and tools point to ways in  
367 which these distinctions can be made in the primary care setting. The Amer-  
368 ican Academy of Neurology and the Child Neurology Society published  
369 a practice parameter for screening and diagnosis of autism, including red  
370 flags requiring immediate referral for further evaluation that focus on ex-  
371 pressive and nonverbal communication, joint attention, and regression: no  
372 babbling, or pointing, or other gestures by 12 months; no single words by  
373 16 months; no two-word spontaneous (not echolalic) phrases at 24 months;  
374 and any loss of any language or social skills at any age [63]

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

388 Pediatricians are increasingly expected to recognize the subtle, early signs  
389 of ASD, before language delay is evident, and to respond quickly with ap-  
390 propriate referrals. Pediatricians should familiarize themselves with screen-  
391 ing tools designed specifically to identify autism and should know how to  
392 refer parents to community resources, including specialists with expertise  
393 in ASD and early intervention programs. In addition, it is vital to have a par-  
394 ticularly high index of suspicion in siblings of children who have ASD, es-  
395 pecially those who have language delay, because their risk for the disorder  
396 is many times that of the general population.

397 Because children who have ASD face new challenges with each transition  
398 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.

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