

Autistic spectrum disorders in preschool children

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ABSTRACT

OBJECTIVE To review existing data on early signs of autistic spectrum disorders (ASD) and on how these disorders can be distinguished from other atypical patterns of development, and to describe a developmental surveillance approach that family physicians can use to ensure that children with these diagnoses are detected as early as possible.

QUALITY OF EVIDENCE MEDLINE was searched from January 1966 to July 2000 using the MeSH terms autistic disorder/diagnosis AND diagnosis, differential AND (infant OR child, preschool). Articles were selected based on relevance to developmental surveillance in primary care and on experimental design, with emphasis on prospective studies with systematic measurement procedures using up-to-date diagnostic criteria.

MAIN MESSAGE Autistic spectrum disorders are characterized by impairments in social interaction and verbal and nonverbal communication, and by preferences for repetitive interests and behaviours. Early signs that distinguish ASD from other atypical patterns of development include poor use of eye gaze, lack of gestures to direct other people's attention (particularly to show things of interest), diminished social responsiveness, and lack of age-appropriate play with toys (especially imaginative use of toys). Careful attention to parents' concerns and specific inquiry into and observation of how children interact, communicate, and play will help ensure that early signs are detected during regular health maintenance visits.

CONCLUSION Family physicians have an important role in early identification of children with ASD. Early diagnosis of these disorders is essential to ensure timely access to interventions known to improve outcomes for these children.

RÉSUMÉ

OBJECTIF Passer en revue les données existantes sur les signes précurseurs de troubles autistiques et sur les distinctions permettant d'exclure d'autres modèles atypiques de développement. Décrire une approche de surveillance du développement que peuvent utiliser les médecins de famille pour assurer le dépistage des enfants autistiques aussitôt que possible.

QUALITÉ DES DONNÉES Une recension a été effectuée dans MEDLINE, de janvier 1966 à juillet 2000, à l'aide des termes MeSH en anglais pour trouble/diagnostic de l'autisme ET diagnostic, différentiel ET (nourrisson OU enfant, préscolaire). La sélection des articles s'est fondée sur leur pertinence à la surveillance du développement dans les soins de première ligne et sur la conception expérimentale, insistant sur les études prospectives ayant des procédures de mesure systématiques fondées sur les critères de diagnostic les plus récents.

PRINCIPAL MESSAGE Les troubles autistiques se caractérisent par des déficiences dans l'interaction sociale et dans la communication verbale et non verbale, et par la préférence accordée aux intérêts et aux comportements répétitifs. Au nombre des premiers signes qui distinguent l'autisme d'autres modèles atypiques de développement figurent une utilisation médiocre du contact oculaire, le manque de gestes pour diriger l'attention d'autrui (en particulier pour désigner des choses d'intérêt), une réceptivité sociale diminuée et le manque de jeu approprié à l'âge avec des jouets (en particulier, l'usage imaginaire de jouets). Une attention précise aux préoccupations des parents et des questions spécifiques concernant la façon dont l'enfant interagit, communique et joue ainsi qu'une observation de ces comportements aideront à assurer le dépistage des premiers signes durant les visites médicales régulières.

CONCLUSION Les médecins de famille ont un rôle important à jouer dans l'identification précoce des enfants autistiques. Un diagnostic sans délai de ces troubles est essentiel pour assurer l'accès opportun aux interventions connues pour aider à améliorer les résultats chez ces enfants.

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Cet article a fait l'objet d'une évaluation externe.

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Autism is the prototype of a group of disorders referred to by the International Classification of Diseases, 10th edition (ICD-10)¹ and the *Diagnostic and Statistical Manual of Mental Disorders*, 4th edition (DSM-IV)² as pervasive developmental disorders (PDD). Autism and PDD are characterized by severe impairments in social interaction and communication, and a preference for repetitive, solitary activities.

The range of diagnostic subtypes within autism and PDD has also been referred to as "autistic spectrum disorders" (ASD), a term that might be more easily understood and accepted by families than PDD.³ For many years, ASD have been regarded as rare and untreatable conditions⁴; however, recent evidence strongly refutes this point of view on both counts. Prevalence rates of autism and of the full range of ASD are estimated to be at least 1/1000^{5,6} and at least 2/1000,⁷ respectively. Several studies have demonstrated that early, intensive intervention leads to marked improvements in long-term outcomes for children with these disorders.^{8,9}

Primary care providers have a key role in early identification of children with ASD.¹⁰ Currently, many children are diagnosed too late to receive the full benefit of early intervention. Most parents first note symptoms before their children are 2 years old.^{11,12} The average delay between parents' first request for help and referral for consultation, however, is longer than 1 year.¹² Autism is finally diagnosed on average at about 4.5 years old.^{12,13}

Quality of evidence

Evidence on early signs of ASD is based on retrospective reports, analysis of home videos recorded before diagnosis, prospective studies of high-risk groups and population-based samples, and one recent systematic review.¹⁰ MEDLINE was searched from January 1966 to July 2000 using the MeSH terms autistic disorder/diagnosis AND diagnosis, differential AND (infant OR child, preschool). Articles were selected based on relevance to developmental surveillance in primary care and experimental design, with emphasis on prospective studies with systematic measurement procedures using current diagnostic criteria.

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Opportunities for early identification: key questions

At what age can the earliest signs of ASD be detected? It is currently uncommon for ASD to be diagnosed in children younger than 3 years. Yet one well-designed population-based study suggests that early signs can be detected as young as 18 months.¹⁴

Can ASD be distinguished from other atypical patterns of development at an early age?

Several prospective studies have demonstrated that experienced clinicians can reliably make the diagnosis in 2-year-old children referred by primary care physicians¹⁵⁻¹⁷ and that specific features distinguish these children from others who have isolated language or general cognitive delays.

Can these early diagnostic features be detected by family physicians during regular health maintenance visits?

Although this question has not been systematically evaluated, evidence indicates that early signs can be detected by primary care providers, even during brief interactions.¹⁴ Overall, although much remains to be learned, there is compelling evidence that clinical signs of ASD can be observed by 18 months to 2 years old and that these diagnoses can be distinguished from other atypical patterns of development even at this early age.

Diagnostic criteria

Autistic spectrum disorders are defined by impairments in three core areas of development: social interaction, communication, and behaviours and interests. Specific criteria are listed in ICD-10¹ and DSM-IV² and are summarized by questions listed in **Table 1**.

Distinguishing ASD from other atypical patterns of development

Impairments in communication. Speech delay is one of the most common early concerns of parents of children with ASD. Speech delay is not specific to ASD; in fact, children who are late talkers are more likely to have isolated language delays or general cognitive delays than to have ASD.¹⁸ Speech delay also occurs when there is severe-to-profound hearing loss. Children with language delays and hearing impairments, however, are generally able to communicate effectively using nonverbal means, such as eye contact, facial expressions, and gestures such as pointing.

Children with cognitive handicaps might have delays in nonverbal communication in line with their overall delays. By the time children reach a

**Table 1. Diagnostic criteria for autism:
Key clinical questions**

Does the child show impairments in social interaction?

- Does the child use eye contact, facial expression, and other body language to initiate and maintain social interaction?
- Does the child interact in a reciprocal fashion? (Can the child both initiate and respond to social interaction, does the child show awareness of how other people feel?)
- Does the child make an effort to show other people things that interest him or her and want other people to know when he or she is having fun?
- Does the child interact with peers in the expected way for someone of his or her developmental stage? (Does the child show interest and approach other children in a friendly way, does the child seem to enjoy the interaction or only the toy or activity?)

Does the child have impairments in verbal and nonverbal communication?

- Is the child able to communicate (eg, make requests) either using words or using specific gestures such as pointing? (Taking parent's hand is not considered a gesture)
- Are there unusual features to the child's speech? (eg, echolalia—repeating back what others have said; pronoun reversal—referring to himself or herself as “you”; odd intonation)
- Can the child participate in a two-way conversation on a topic that someone else chooses? (For children with sufficient speech)
- Does the child spontaneously imitate what other people are doing and incorporate this into spontaneous, imaginative play?

Does the child show repetitive, stereotyped interests and behaviours?

- Is the child fascinated by unusual topics or items? Is this fascination so intense that it is difficult to get the child interested in other things?
 - Is the child distressed by minor changes in daily routines or how his or her personal things are arranged? Does the family go to unusual lengths to avoid changing things?
 - Does the child play with toys in a conventional way or does he or she become preoccupied with a certain aspect (eg, spinning the wheels or lining up toy cars rather than driving around)?
 - Does the child have any unusual motor behaviours (eg, hand flapping or finger flicking)?
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developmental level of 12 months, however, they are usually able to use eye gaze and simple gestures to direct other people's attention very effectively.¹⁹ Young children with ASD frequently lack these other means of communicating and cannot compensate for their lack of words. A profound lack of nonverbal communication should be considered highly indicative of

a diagnosis of autism, even in children who are globally delayed.²⁰ As children with ASD begin to speak, they might have unusual speech patterns, such as echolalia (repeating back what someone else has said) and pronoun reversal (eg, referring to themselves as “you” instead of “I”).

Children with specific language delays might occasionally repeat words or use rote phrases, but this tends to be a passing phase and does not represent most of what they say, as is often the case with children with ASD. Some children with ASD, however, have neither speech delays nor unusual speech patterns. For example, Asperger disorder, a diagnosis within ASD, is defined by impairments in social interaction and stereotyped interests and behaviours, but essentially normal early language skills.²¹ Even so, children with this disorder lack social chit-chat (ie, making comments simply to be friendly). Children with ASD sometimes also obsess on specific topics of interest and lack the ability to shift from these topics to have a flexible two-way conversation.

Impairments in social interaction. Contrary to traditional stereotypes, children with ASD can be very affectionate and enjoy attention and physical play with their parents and other people they know. What they often lack is an understanding of another person's point of view and the need to reciprocate in play and other social interactions. They might be unable to effectively get someone else's attention, for example, by making eye contact and smiling. Whereas typically developing toddlers are constantly seeking their parent's attention and drawing them into their play, children with ASD might be quite content to play on their own and even resist the attempts of their parents to join in.

Although children with ASD frequently show interest in other children, they might react negatively when approached by other children and fail to imitate or join into play activities. Basic deficits in early social behaviours (eg, reciprocal smiling, social responsiveness, imitation, and wanting to show things of interest) appear to be highly specific for ASD.^{9,22,23}

Interests and behaviours. Intense narrow interests and stereotyped behaviour patterns are often prominent in older children with ASD but are less frequent and less specific for diagnosis in children younger than 3 years.^{13,15,24} Many children with ASD from a very early age show intense fascination (often of a sensory nature, such as watching dust twinkle in the sunlight), resistance to change, and motor mannerisms (such as

Table 2. Signs of autism in preschoolers

- Failure to achieve language milestones:
- Babbling by 11 months
 - Simple gestures (eg, waving) by 12 months
 - Single words by 16 months
 - Two-word phrases (a noun and a verb) by 24 months
- Loss of any language or social skills at any age
- Lack of response to name being called
- Rarely makes eye contact when interacting with other people
- Does not point to show things in which he or she is interested
- Does not smile socially
- More interested in looking at objects than at people's faces
- Does not make attempts to get parents' attention
- Does not respond to or avoids parents' attempts to play, even if relaxed
- Avoids or ignores other children when they approach
- Has odd or repetitive ways of moving his or her hands or fingers
- Lacks interest in toys or plays with them in an unusual way (eg, lining up, spinning, opening or closing parts rather than using the toy as a whole)
- Has compulsions and rituals (has to perform activities in a special way or sequence)

hand flapping). Stereotyped behaviours and sensory interests can also be observed in children with severe cognitive disabilities, however. One area of behaviour that might help identify autism at an early age is a child's interest in and use of toys. A lack of imaginative play, in combination with specific deficits in social-communication skills, is highly predictive of a later diagnosis of autism.

Baron-Cohen et al^{14,25} developed a Checklist for Autism in Toddlers (CHAT) based on skills that can be observed in typically developing children by the age of 15 months. They assessed 16 000 18-month-olds in the community and found that the absence of three key skills identified toddlers likely to be diagnosed with autism: pretend play (even as simple as offering a sip from an empty toy teacup), pointing to show (rather than just to request), and gaze monitoring (following someone else's shift in gaze). In only 12 of the 16 000 children were these three skills all absent, and 10 of these 12 were subsequently diagnosed with autism.^{14,26}

Table 3. Information sources on treatment options for children with autism spectrum disorders

ORGANIZATION	ADDRESS	WEBSITE
Autism Treatment Services of Canada	404-94th Ave SE, Calgary, AB T2J 0E8	www.autism.ca
The Geneva Centre	250 Davisville Ave, Toronto, ON M4S 1H2	www.autism.net
Autism Society of America	7910 Woodmont Ave, Suite 300, Bethesda, MD 20814-3015	www.autism-society.org
Online Asperger Syndrome Information and Support (OASIS)	(website only; affiliated with University of Delaware)	www.udel.edu/bkirby/asperger/

Implications for clinical practice

It was initially hoped that the CHAT, with its high specificity and positive predictive value, could be adopted as a universal screen. Recent data on the CHAT, however, indicate that, while it can aid primary care physicians in identifying children at risk, it is not sufficiently sensitive to stand alone as a screening tool.

Baird and colleagues²⁷ followed up the original population screened by the CHAT to the age of 7 years, checking for additional cases of autism by review of medical and educational records. They found that the CHAT, administered at 18 months, detected only 38 % of children who went on to be diagnosed with autism (or other ASD). Other checklists have been designed to screen for early signs of ASD (eg, PDD Screening Test²⁸), but they are still being evaluated for predictive validity. At this point, no single questionnaire or observational measure can be used to reliably and sensitively identify all young children with autism.

A more multifaceted approach will be required to substantially lower the average age at which ASD are first identified. Questionnaires such as the CHAT might be used as aids, but family physicians' clinical judgment and expertise will also be needed to identify specific features that warrant referring children for a formal diagnostic assessment for autism.

The Child Neurology Society and the American Academy of Neurology recently published Practice Parameters for the Diagnosis and Evaluation of Autism.¹⁰ As part of developing these parameters, a multidisciplinary consensus panel reviewed existing literature and developed a model of community care. The first and most critical level of this model of care involves routine developmental surveillance by primary care providers. Key elements of family physicians' role are

familiarity with the signs and symptoms of autism; careful attention to parents' concerns about their children's early language and social skills; specific inquiry into how children interact, communicate, and play; attention to these aspects of children's functioning during office visits; and prompt referral of children failing to achieve key communication milestones or showing other signs indicative of risk of ASD (**Table 2**).

Timely access to specialized assessment and treatment services is also critical for early diagnosis and intervention for preschool children with ASD. It is important to note that, while not all children who show these signs have ASD, many who do not will have other conditions, such as specific language or general cognitive delays, and will still benefit from early identification and access to intervention.²⁶

Intervention services for children with autism vary by communities; it is important that physicians learn about local services available to young children with autism and other developmental disorders. Children with autism benefit from intensive intervention designed to systematically teach communication, social, and play skills.^{8,9} A critical review of treatment options for autism is beyond the scope of this paper, but several websites containing useful information and resources are listed in **Table 3**.

Conclusion

Autistic spectrum disorders are characterized by impairments in social interaction and verbal and non-verbal communication, and a pattern of repetitive interests and behaviours. Most children with ASD first present with deficits in basic social and communication skills in the first 2 years of life. Early signs that distinguish ASD from other atypical patterns of development include poor use of eye gaze and gestures to direct other people's attention, diminished social responsiveness, and lack of age-appropriate play.

Family physicians have an important role in early identification of children with ASD. Careful attention to parents' concerns, and specific inquiry into and observation of how children interact, communicate, and play will help ensure that these early signs are detected during regular health maintenance visits. Early diagnosis of these disorders is essential to ensure timely access to interventions known to improve outcomes for these children. ❖

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Editor's key points

- Clinical signs of autistic spectrum disorders (ASD) can be observed as early as 18 to 24 months; these disorders can be distinguished from other atypical patterns of development even at this early age.
- No single questionnaire or observational measure can be used to reliably and sensitively identify all young children with autism.
- Physicians should suspect these disorders when children lack language, gestures, and other body movements to direct attention, do not try to interact socially, and have no imaginative play with toys.
- Early diagnosis of ASD is essential to ensure timely therapeutic intervention known to improve outcomes for these children.

Points de repère du rédacteur

- Il est possible de distinguer le trouble autistique des autres profils de développement atypique dès l'âge de 18 à 24 mois.
- Il n'existe pas actuellement de questionnaire ou d'outil de dépistage du trouble autistique offrant une sensibilité et une spécificité suffisante pour être utilisé de façon universelle chez les enfants.
- Le médecin doit envisager ce diagnostic chez le jeune enfant qui présente un retard de langage, une faible utilisation du langage non verbal, un manque d'intérêt pour les interactions sociales et pour les jouets.
- Le diagnostic précoce de l'autisme est essentiel car l'intervention thérapeutique, si elle est débutée tôt, améliore le pronostic de ces enfants.

Competing interests

None declared

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References

1. World Health Organization. *The ICD-10 classification of mental and behavioral disorders—clinical descriptions and diagnostic guidelines*. Geneva, Switz: World Health Organization; 1992.
2. American Psychiatric Association. *Diagnostic and statistical manual of mental disorders*. 4th ed. Washington, DC: American Psychiatric Association; 1994.
3. Wing L. Autistic spectrum disorders. *BMJ* 1996;312:327-8.
4. Lotter V. Follow-up studies. In: Rutter M, Schopler E, editors. *Autism: a re-appraisal of concepts and treatment*. New York, NY: Plenum Press; 1978. p. 475-95.
5. Gillberg C, Wing L. Autism: not an extremely rare disorder. *Acta Psychiatr Scand* 1999;99:399-406.
6. Fombonne E. The epidemiology of autism: a review. *Psychol Med* 1999;29:769-86.
7. Kadesjo B, Gillberg C, Hagberg B. Brief report: autism and Asperger syndrome in seven-year-old children: a total population study. *J Autism Dev Disord* 1999;29:327-31.

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8. Rogers SJ. Brief report: early intervention in autism. *J Autism Dev Disord* 1996;26:243-6.
9. Dawson G, Osterling J. Early intervention in autism: effectiveness and common elements of current approaches. In: Guralnick MJ, editor. *The effectiveness of early intervention*. Baltimore, Md: Brooks; 1997. p. 307-26.
10. Filipek PA, Accardo PJ, Ashwal S, Baranek GT, Cook EH Jr, Dawson G, et al. The screening and diagnosis of autistic spectrum disorders. *Neurology* 2000;55:468-79.
11. Rogers SJ, DiLalla DL. Age of symptom onset in young children with pervasive developmental disorders. *J Am Acad Child Adolesc Psychiatry* 1990;29:863-72.
12. Howlin P, Moore A. Diagnosis in autism: a survey of over 1200 patients in the UK. *Autism* 1997;1:135-62.
13. Siegel B, Pliner C, Eschler J, Elliott GR. How children with autism are diagnosed: difficulties in identification of children with multiple developmental delays. *J Dev Behav Pediatr* 1988;9:199-204.
14. Baron-Cohen S, Cox A, Baird G, Swettenham J, Nightingale N, Morgan K, et al. Psychological markers in the detection of autism in infancy in a large population. *Br J Psychiatry* 1996;168:158-63.
15. Stone WL, Lee EB, Ashford L, Brissie J, Hepburn SL, Coonrod EE, Weiss BH. Can autism be diagnosed accurately in children under 3 years? *J Child Psychol Psychiatry* 1999;40:219-26.
16. Lord C. Follow-up of two-year-olds referred for possible autism. *J Child Psychol Psychiatry* 1995;36:1365-82.
17. Gillberg C, Ehlers S, Schaumann H, Jakobsson G, Dahlgren SO, Lindblom R, et al. Autism under age 3 years: a clinical study of 28 cases referred for autistic symptoms in infancy. *J Child Psychol Psychiatry* 1990;31:921-34.
18. Bauer S. Autism and the pervasive developmental disorders: part 1. *Pediatr Rev* 1995;16:168-77.
19. Carpenter M, Nagell K, Tomasello M. Social cognition, joint attention, and communicative competence from 9 to 15 months of age. *Monogr Soc Res Child Dev* 1998;63:1-143.
20. Sigman M, Ruskin E, Arbeile S, Corona R, Dissanayake C, Espinosa M, et al. Continuity and change in the social competence of children with autism, Down syndrome, and developmental delays. *Monogr Soc Res Child Dev* 1999;64:1-114.
21. Szatmari P, Archer L, Fisman S, Streiner DL, Wilson F. Asperger's syndrome and autism: differences in behavior, cognition, and adaptive functioning. *J Am Acad Child Adolesc Psychiatry* 1995;34:1662-71.
22. Osterling J, Dawson G. Early recognition of children with autism: a study of first birthday home videotapes. *J Autism Dev Disord* 1994;24:247-57.
23. Mars AE, Mauk JE, Dowrick PW. Symptoms of pervasive developmental disorders as observed in prediagnostic home videos of infants and toddlers. *J Pediatr* 1998;132:500-4.
24. Stone WL, Hoffman EL, Lewis SE, Ousley OY. Early recognition of autism. Parental reports vs clinical observation. *Arch Pediatr Adolesc Med* 1994;148:174-9.
25. Baron-Cohen S, Allen J, Gillberg C. Can autism be detected at 18 months? The needle, the haystack, and the CHAT. *Br J Psychiatry* 1992;161:839-43.
26. Cox A, Klein K, Charman T, Baird G, Baron-Cohen S, Swettenham J, et al. Autism spectrum disorders at 20 and 42 months of age: stability of clinical and ADIR diagnosis. *J Child Psychol Psychiatry* 1999;40:719-32.
27. Baird G, Charman T, Baron-Cohen S, Cox A, Swettenham J, Wheelwright S, et al. A screening instrument for autism at 18 months of age: a 6-year follow-up study. *J Am Acad Child Adolesc Psychiatry* 2000;39:694-702.
28. Siegel B. Early screening and diagnosis in autism spectrum disorders: the Pervasive Developmental Disorders Screening Test (PDDST). *Proceedings of the National Institutes of Health State of the Science in Autism: Screening and Diagnosis Working Conference*; 1998 June 15-17; Bethesda, Md. Bethesda, Md: National Institutes of Health; 1998.