



# The Pediatrician's Role in the Diagnosis and Management of Autistic Spectrum Disorder in Children (RE060018)

## AMERICAN ACADEMY OF PEDIATRICS

### [Technical Report: The Pediatrician's Role in the Diagnosis and Management of Autistic Spectrum Disorder in Children](#)

Committee on Children With Disabilities

**ABSTRACT.** Primary care physicians have the opportunity, especially within the context of the medical home, to be the first point of contact when parents have concerns about their child's development or behavior. The goal of this policy statement is to help the pediatrician recognize the early symptoms of autism and participate in its diagnosis and management. This statement and the accompanying technical report will serve to familiarize the pediatrician with currently accepted criteria defining the spectrum of autism, strategies used in making a diagnosis, and conventional and alternative interventions.

**ABBREVIATIONS.** ASD, autistic spectrum disorder; PDD-NOS, pervasive developmental disorder-not otherwise specified; MMR, measles-mumps-rubella; *DSM-IV*, *Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition*; CHAT, Checklist for Autism in Toddlers.

Autism is not a specific disease but rather a disorder of brain development with a strong genetic basis. Children with autism demonstrate behaviors and skills that span a broad continuum extending from very mild peculiarities to severe developmental challenges. It is now known to be a very heterogeneous disorder, with milder forms being more common than the classic form. Although clinical patterns vary depending on severity, all children with autism demonstrate some degree of qualitative impairment in reciprocal social interaction, qualitative impairment of communication, and restricted, repetitive, and stereotypic patterns of behaviors, interests, and activities.<sup>1</sup> Because of qualitative and quantitative variations in symptoms, autism is often referred to as autistic spectrum disorder (ASD).<sup>2-5</sup> This term encompasses the classic autistic disorder and other pervasive developmental disorders. This statement focuses on autistic disorder and its milder variants, including Asperger syndrome and pervasive developmental disorder-not otherwise specified (PDD-NOS).

Recent evidence that the prevalence of diagnosed ASD may be increasing and that early diagnosis and intervention are likely associated with better long-term outcomes<sup>6-15</sup> has made it imperative that pediatricians increase their fund of knowledge regarding the disorder. Earlier studies estimated the prevalence of autism to be 4 to 5 in 10 000 persons.<sup>16</sup> Most of the more recent studies have revealed that a conservative estimate is approximately 1 in 1000 children for autistic disorder and 2 or more in 1000 children for ASD.<sup>17,18</sup> The majority of studies of autistic disorder conducted through 1998 showed a prevalence of less than 1 per 1000. There have been a few recent studies that have shown higher rates.<sup>19-</sup>

<sup>23</sup> These studies with higher rates have been in communities where intense case finding was used to try to identify every possibly affected child in the area. Currently, there are no data available for a large US population; however, even if the conservative rates apply, pediatricians can now expect to care for at least 1 child with ASD. The apparent increase may represent a combination of several factors, including changing criteria with inclusion of milder forms in the spectrum of autism, a higher public and professional recognition of the disorder, and a true rise in prevalence.

Although a group of investigators in the United Kingdom<sup>24</sup> has hypothesized that administration of measles-mumps-rubella (MMR) vaccine was associated with an increased risk of ASD, this hypothesis has not been substantiated by more in-depth research.<sup>25-30</sup> In addition, it is imperative that health professionals and the public realize that congenital rubella can cause autism and that measles and mumps can cause significant disability, including encephalitis.<sup>31,32</sup>

## **STATEMENT OF THE PROBLEM**

The pediatrician is faced with the challenging task of suspecting an ASD diagnosis as early as possible and implementing a timely treatment plan to achieve the best outcome for the child and family. Early diagnosis of ASD is challenging in the context of primary care visits, because there is no pathognomonic sign or laboratory test to detect it. Thus, the physician must make the diagnosis on the basis of the presence or absence of a constellation of symptoms. ASD is a phenomenologic rather than an etiologic disorder (eg, trisomy 21 in Down syndrome), making the diagnosis more challenging. Pediatricians must rely on parent report, clinical judgment, and the ability to recognize criteria-based behaviors that define ASD.

Pediatricians are now seeing more children with ASD in their offices; thus, they need to increase their fund of knowledge and comfort level in caring for these children. Families are calling on their pediatricians to guide them through the plethora of behavioral, educational, psychopharmacologic, and alternative treatment options available to them. Early diagnosis is imperative to ensure prompt referral to an appropriate early intervention program.

Research has demonstrated that the recurrence rate for isolated ASD in subsequent siblings ranges from 3% to 7%.<sup>33-35</sup> This represents a recurrence risk approximately 50 times the baseline. Thus, early diagnosis is also important to ensure timely genetic counseling before the conception of subsequent siblings.

## **NEW INFORMATION**

Substantial progress has been made during the past 20 years in the early diagnosis of ASD, detection of underlying etiologic neurologic and genetic conditions,<sup>3,4</sup> and development of behavioral, educational,<sup>36</sup> and psychopharmacologic<sup>37</sup> interventions. There also has been an increase in alternative therapies for children with ASD with which the pediatrician should be familiar.<sup>38,39</sup>

Early diagnosis is dependent on listening carefully to parents' concerns about their child's development and behavior. Current research has revealed that parents are usually correct in their concerns about their child's development.<sup>40-43</sup> Any concerns should be valued and should lead to additional investigation by the primary care pediatrician, a child neurologist, a developmental pediatrician or other qualified specialist, or preferably, a team of specialists.

Aberrant social skill development is a hallmark for ASD. Early social skill deficits may include abnormal eye contact, aloofness, failure to orient to name, failure to use gestures to point or show, lack of interactive play, and lack of interest in peers, among others. In general, parents infrequently raise concerns about social skill deficits; therefore, when they do, the concerns are serious red flags and ASD should be considered. Combined language and social delays and regression in language or social milestones are even bigger red flags for ASD and should prompt additional evaluation immediately. More commonly, parents of children later diagnosed with ASD express subtle concerns about speech delays and unusual behavior problems.<sup>3,44</sup> Speech delay has many additional causes (including hearing loss and cognitive deficits) and is the most common developmental concern voiced by parents of children between 1 and 3 years old.<sup>45,46</sup>

In contrast, lack of a developmental concern does not imply typical development. All children should be formally monitored for developmental progress at every well-child care visit. Developmental surveillance is an important function of the pediatrician in the context of the medical home<sup>47,48</sup> and should include social-emotional milestones in addition to the more traditional motor, cognitive, and language ones. Parents may complete a standardized developmental questionnaire or an objective screening tool may be used during the visit.<sup>3,49</sup> Any concerns should prompt the pediatrician to perform a more comprehensive standardized test. Additionally, because of the relatively high familial recurrence rates, a younger sibling of a child with known isolated ASD deserves a high level of surveillance whether or not parents have concerns. If the pediatrician is unfamiliar with or unable to perform developmental testing, the child for whom there is a concern and/or the sibling should promptly be referred to a specialist or, preferably, a team of specialists.

When considering the diagnosis of ASD, physicians refer to the *Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition (DSM-IV)*<sup>1</sup> to determine whether or not a child meets the criteria-based diagnosis for a disorder within the spectrum. The definition and criteria for ASD have been broadened to include milder forms. The newest criteria (Table 1 in the accompanying technical report<sup>2</sup>) can be found in the *DSM-IV*<sup>1</sup> or the *DSM for Primary Care, Child and Adolescent Version*<sup>50</sup> published by the Academy. These criteria were established for children 3 years and older and may be difficult to apply to younger children. If a child meets some but not all criteria, the diagnosis of PDD-NOS may apply.

Alternative tools have been developed to aid physicians in screening for ASD in younger children. These relatively new instruments, although promising, need additional validation to assess their sensitivity and specificity across various ethnic populations. The Checklist for Autism in Toddlers (CHAT; Fig 1 in accompanying technical report<sup>2</sup>) was developed in England for use with 18-month-old children and has been used to screen more than 16 000 toddlers.<sup>51-53</sup> Although it has a high specificity, its relatively low sensitivity is a concern. A modified version of the CHAT is being developed to address this issue. The Pervasive Developmental Disorder Screening Test is a parent-completed survey of early signs and symptoms of ASD, but less is known about its sensitivity and specificity.<sup>54</sup> When ASD is suspected on the basis of clinical symptoms or results of these or similar screening tools, a comprehensive assessment (including autism-specific tools<sup>2-4,36</sup>) should be performed by a specialist or, preferably, a multidisciplinary team of specialists with expertise in ASD to make the definitive diagnosis and search for possible etiologic disorders.

Most children with idiopathic ASD have a normal physical appearance except for the presence of macrocephaly in 25%.<sup>55,56</sup> Thus, the physical examination, including a detailed neurologic examination, may be more helpful in determining the presence or absence of an associated medical condition. A

recognizable etiologic disorder occurs in less than 25% of cases of ASD.<sup>57</sup> Although now rare because of widespread immunization, congenital rubella is associated with autistic behaviors, severe mental retardation, microcephaly, congenital heart defects, and vision and hearing impairments.<sup>31,32</sup>

Dysmorphic characteristics of more common etiologic conditions include the long face, large ears, and large testes (postpubertally) associated with fragile X syndrome; the hypopigmented ash leaf macules, facial angiofibromas, and seizures associated with tuberous sclerosis; and the ataxic gait and broad mouth with persistent large smile associated with Angelman syndrome.

In addition to making the clinical diagnosis of ASD, the pediatrician is faced with deciding which diagnostic tests are indicated to determine the cause of ASD, determine whether there are comorbid disorders, and rule out disorders included in the differential diagnosis. Some measure of the child's overall level of cognitive functioning and adaptive skills is necessary, especially if there is a concern about comorbid mental retardation. An audiologic evaluation and a comprehensive speech and language evaluation should be done in any child who has language delays whether or not autistic features are present. Other tests to consider include a lead screening, amino acid screening to detect phenylketonuria, DNA analysis to detect fragile X syndrome, high-resolution chromosome analysis, and prolonged sleep-deprived electroencephalography (in children who have symptoms of developmental regression or clinical seizures or when there is a high suspicion of subclinical seizures).<sup>3,4</sup> Computed tomography or magnetic resonance imaging is not routinely indicated and probably will not be helpful in the child with ASD who has isolated macrocephaly and no localizing signs.<sup>3,4,58,59</sup> The need for diagnostic studies must be evaluated on the basis of specific signs in the individual child and the possible contribution the results will make to genetic counseling and management rather than using a "shotgun" approach.

To summarize diagnostic challenges facing the pediatrician, there must be a high index of suspicion, especially when parents have concerns about their child's language and social development, and extra attention should be given to subsequent siblings of children with isolated ASD. Pediatricians with adequate training and experience are encouraged to use autism-specific diagnostic tools to make the definitive diagnosis and additional diagnostic tools to search for an etiologic or comorbid disorder. The importance of early diagnosis cannot be overemphasized. If a primary care physician is uncomfortable with making the diagnosis, the child should be promptly referred to a specialist or, preferably, a team of specialists with expertise in the diagnosis of ASD. Primary care physicians seeking assistance with an etiologic diagnosis may additionally request a genetics consultation.

The challenge for the pediatrician does not end when the diagnosis of ASD has been made. Unlike some disorders for which there are specific treatment protocols based on abundant research evidence, presently there are no treatment guidelines for ASD published in the general pediatric literature to assist the primary care physician. However, reviews have recently been published in early intervention<sup>36</sup> and child psychiatry literature.<sup>37</sup> Currently accepted strategies are to improve the overall functional status of the child by enrolling the child in an appropriate and intensive early intervention program that promotes development of communication, social, adaptive, behavioral, and academic skills; decrease maladaptive and repetitive behaviors through use of behavioral and sometimes pharmacologic strategies; and help the family manage the stress associated with raising a child with autism, particularly by providing information about community resources, respite care, and parent support organizations.

Early diagnosis resulting in early, appropriate, and consistent intervention has also been shown to be associated with improved long-term outcomes.<sup>6-15</sup> Although there is growing agreement among experts that early and sustained intensive behavioral and educational interventions may improve overall outcomes, there is less agreement regarding the relative effectiveness of specific intervention strategies or the degree to which they should be delivered.

Intervention strategies should be tailored to the child's developmental and behavioral needs and to the family's coping style and resources. Although the menu of services may vary among children, all children with ASD should be cared for in the context of the medical home. Early management strategies include the following:

1. Parental education and support. National and regional parent support organizations, such as the Autism Society of America (web site available at: <http://www.autism-society.org> [includes additional links to other organizations]), can be a source of information and of great benefit to families. However, parents should be cautioned about the possibility of misinformation, especially from Internet sources, regarding etiology (eg, the alleged association with MMR vaccine) and miracle cures.
2. Early intervention (children younger than 3 years). Appropriate interventions in this age group usually take place in homes or child care centers and are individualized. They include behavioral methods, early developmental education, communication, occupational and physical therapy, highly structured social play interventions, and extensive parent training.
3. School-based special education (children older than 3 years). Educational interventions should be individualized and take into account the child's specific strengths and deficits. Programs vary somewhat in philosophy, curricula, and implementation but should always provide the child with structure and methods that will facilitate social skills, functional communication, and learning. Speech and occupational therapy and use of typically developing peers as role models and playmates are usually included in these programs.
4. Behavior management. Behavioral training, including communication development, has been shown to be effective in reducing problem behaviors and improving adaptation.<sup>60-62</sup> The overall goal of the approach is to reinforce desirable behaviors and reduce undesirable ones using behavioral psychologic theory.
5. Medical treatment. Although children with ASD have the same health care needs as children without disabilities, they are at greater risk of concurring psychiatric problems.<sup>63</sup> In addition, newer and safer medications have been developed and shown to be beneficial in the treatment of certain target behaviors associated with ASD.<sup>37,64-68</sup> Whenever possible, a specific psychiatric diagnosis (eg, anxiety, obsessive-compulsive disorder) should be made to guide treatment. This might best be facilitated by a referral to a pediatric psychiatrist or a developmental pediatrician.
6. Community services. Raising a child with ASD can impose a great deal of stress on the entire family. The degree to which the family needs community support depends on their available resources (ie, extended family, neighbors, friends, and spiritual community). The number of agencies serving children with ASD has increased, and they can provide valuable support services, such as respite, to families.
7. Alternative therapies. Because ASD is a chronic condition for which presently there is no medical cure, it has become the focus of several unconventional treatments. However, rigorously controlled randomized studies are still scarce and scientific evidence is lacking for alternative interventions.

More detailed descriptions of these interventions can be found in the accompanying technical report.<sup>2</sup> Pediatricians who provide a medical home to children with ASD should become aware of interventions available in their own communities and how to access them. If the pediatrician is not comfortable with managing and coordinating the care of a child with ASD, the child should be referred to an experienced professional or team of professionals.

## CONCLUSIONS

Our understanding of the spectrum, etiology, diagnosis, and management of ASD in children has

changed dramatically throughout the past 2 decades. Early diagnosis has become increasingly important as recent studies have shown improved outcomes with implementation of early, consistent, and appropriate intervention strategies that have been individually tailored to the needs of the child and parents.<sup>6-15</sup> Most pediatricians will have the opportunity to provide a medical home and coordinate systems of care for children with ASD.

## RECOMMENDATIONS

1. Pediatricians should listen carefully to parents when discussing their child's development. They are reliable sources of information, and their concerns should be valued and addressed immediately.
2. In the context of the medical home, pediatricians should monitor all areas of development at each well-child visit. They should be especially vigilant when there are deficits in language and social skill development.
3. Pediatricians should consider using screening and diagnostic tools specific for ASD. Such tools should be ethnoculturally and linguistically appropriate. If a pediatrician feels unable to do so, the child should be promptly referred to a specialist or, preferably, a multidisciplinary team of specialists with expertise in ASD.
4. Any child who has language delays should be referred for an audiologic and a comprehensive speech and language evaluation. If the child is uncooperative, diagnostic otoacoustic emissions or sedated brainstem auditory evoked responses should be obtained.
5. Pediatricians should continue to promote immunizations for all children. Continued high immunization rates are crucial in preventing an increase in life-threatening infectious diseases. Parents should be reassured that at the present time, there is no scientific evidence to support claims that MMR vaccine or any combination of vaccines cause ASD. A decision to not vaccinate places children and communities at risk.
6. Lead screening is indicated in the presence of risk factors, particularly pica, even in an older child. DNA analysis, high-resolution chromosome analysis, and referral to a geneticist should be considered in the presence of dysmorphic features, family history of fragile X syndrome, or mental retardation of undetermined etiology.<sup>3,4</sup> Electroencephalography and a neurology referral are indicated in children with suspected seizures or those who have symptoms of regression. Decisions to pursue additional investigation (eg, neuroimaging) or consultation for a coexisting etiologic diagnosis (eg, phenylketonuria, tuberous sclerosis, etc) should be made on the basis of the history and physical examination, including an assessment for focal neurologic signs.<sup>3,4</sup>
7. Once the diagnosis of ASD is made, the family and caregivers should be provided with current literature and information regarding parent support groups, specific autism intervention programs, and other available community services.
8. Families should receive genetic counseling appropriate to the etiologic diagnosis. Parents of a child with apparently isolated ASD should be counseled regarding the increased recurrence risk (3% -7%) in subsequent children. When following a younger sibling of a child with known ASD, pediatricians should demonstrate a high level of vigilance and monitor the child closely for any developmental or behavioral concern.
9. Any child with a suspected delay or symptoms of ASD should be given the opportunity to enroll in an age-appropriate early intervention program or school program immediately, even before a definitive diagnosis is available. Because these programs are federally mandated<sup>69,70</sup> (and fully implemented in most states), children with delayed or deviant development are entitled to them. Although criteria may vary slightly among states, eligibility for these programs is based on the presence of a delay, not on a categoric diagnosis.
10. Because many parents of children with ASD pursue alternative therapies, pediatricians are encouraged to become familiar with the more popular ones and approach the issue objectively and

compassionately.<sup>38,39</sup>

11. Pediatricians should provide comprehensive care of the child with ASD in the context of a medical home. This includes provision of medical interventions and coordination of care with appropriate educational, rehabilitation, social, and subspecialty pediatric services.
12. In the event of an untimely death, physicians should encourage parents of a child with ASD to consent to tissue donation to support ASD research endeavors. To advance this effort, national autism organizations have established a centralized brain bank. For more information, one may call: 1-800-BRAIN-BANK.

## IMPLEMENTATION

Implementation of the above strategies depends on educating and empowering pediatricians to recognize the wide spectrum of symptoms that ASD now comprises and use standardized developmental and ASD-specific screening and diagnostic tools. Implementation also depends on reimbursement policies that allow additional well-child visits during toddler and preschool years and adequate time to use these tools. Reimbursement mechanisms must take into account efforts needed to provide comprehensive management and coordination of care in the context of the medical home.

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The recommendations in this statement do not indicate an exclusive course of treatment or serve as a standard of medical care. Variations, taking into account individual circumstances, may be appropriate.

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