# Primary Care for Children with Autism

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The earliest sign of autism in children is the delayed attainment of social skill milestones, including joint attention, social orienting, and pretend play. Language impairment is a common, but less specific, sign of autism. Repetitive behaviors and restricted interests may not be noted until after social skill and communication impairments are exhibited. Physicians should perform developmental surveillance at all well-child visits, and the American Academy of Pedi-

atrics recommends administering an autism-specific screening tool at the 18- and 24-month visits. A referral for comprehensive diagnostic evaluation is appropriate if concerns arise from surveillance, screening, or parental observations. The goals of long-term management are to maximize functional independence and community engagement, minimize maladaptive behaviors, and provide family and caregiver support. Physicians play an important role in coordinating care through an interdisciplinary team; referring families for specialized services; and treating children's associated conditions, including sleep disturbances, gastrointestinal problems, anxiety, and hyperactivity. Autism is a lifelong condition, but early recognition, diagnosis, and treatment can improve the prognosis, whereas associated medical conditions, psychiatric conditions, and intellectual disability can worsen the prognosis. (Am Fam Physician. 2010;81(4):453-460, 461. Copyright © 2010 American Academy of Family Physicians.)



▶ Patient information: A handout on helping a child with autism, written by the authors of this article, is provided on page 461.

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amily physicians play a critical role in the identification and long-term management of autism in children. Autism spectrum disorders (ASDs) include autistic disorder; pervasive developmental disorder, not otherwise specified (PDD-NOS); and Asperger syndrome. The current prevalence of ASDs (approximately one in 110 children) has been rising steadily, with an increase of 57 percent between 2002 and 2006.1 It is unclear whether this represents a true increase in disease prevalence. Potential explanations for this rise in prevalence include increased societal awareness of ASDs, changing diagnostic criteria, and better access to educational services. Autism is four times more common in boys than in girls, a consistent finding that remains unexplained.2

ASDs share three core features: delays in social interaction, impairments in language, and restricted and repetitive behaviors. Diagnostic criteria have been developed for autistic disorder (Table 13) and Asperger syndrome. PDD-NOS is a subthreshold diagnostic term for children with similar impairments that do not meet the full criteria for autistic disorder or Asperger syndrome. Children with Asperger syndrome demonstrate relatively normal expressive language and cognitive skills. This review focuses on autistic disorder and PDD-NOS, which are collectively referred to as autism. Table 2 lists associated conditions often present in children with autism that may have a negative impact on overall function.<sup>1,4-10</sup>

#### Etiology

The role of genetics in ASDs is suggested by a recurrence rate of autism in siblings of affected children that is approximately 10-fold higher than in the general population, and by studies showing a high concordance of ASDs in monozygotic twins.2 Autism susceptibility genes have been demonstrated by whole genome screens of families with multiple children with an ASD, by cytogenetic studies of affected children, and by the evaluation of candidate genes involved in brain development.<sup>2,11</sup> The role of prenatal or

SORT: KEY RECOMMENDATIONS FOR PRACTICE		
Clinical recommendation	Evidence rating	References
An autism-specific screening tool should be administered to all children at the 18- and 24-month office visits.	С	17, 18
Early intensive behavioral therapy can improve cognitive, language, and adaptive skill outcomes in children with autism.	В	15, 31, 32
Treating associated medical and psychiatric conditions can improve overall functioning in children with autism.	А	29, 36-39

A = consistent, good-quality patient-oriented evidence; B = inconsistent or limited-quality patient-oriented evidence; C = consensus, disease-oriented evidence, usual practice, expert opinion, or case series. For information about the SORT evidence rating system, go to http://www.aafp.org/afpsort.xml.

## **Table 1. Diagnostic Criteria for Autistic Disorder**

- A. A total of six (or more) items from (1), (2), and (3), with at least two from (1), and one each from (2) and (3):
  - 1. Qualitative impairment in social interaction, as manifested by at least two of the following:
    - Marked impairment in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction
    - b. Failure to develop peer relationships appropriate to developmental level
    - A lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g., by a lack of showing, bringing, or pointing out objects of interest)
    - d. Lack of social or emotional reciprocity
  - 2. Qualitative impairments in communication as manifested by at least one of the following:
    - a. Delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime)
    - b. In individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others
    - c. Stereotyped and repetitive use of language or idiosyncratic language
    - d. Lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level
  - Restricted repetitive and stereotyped patterns of behavior, interests, and activities, as manifested by at least one of the following:
     a. Encompassing preoccupation with one or more stereotyped and
    - restricted patterns of interest that is abnormal either in intensity or focus
    - b. Apparently inflexible adherence to specific, nonfunctional routines or rituals
    - c. Stereotyped and repetitive motor mannerisms (e.g., hand or finger flapping or twisting, or complex whole-body movements)
    - d. Persistent preoccupation with parts of objects
- B. Delays or abnormal functioning in at least one of the following areas, with onset prior to age three years: (1) social interaction; (2) language as used in social communication; or (3) symbolic or imaginative play.
- C. The disturbance is not better accounted for by Rett's disorder or childhood disintegrative disorder.

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postnatal environmental exposures in altering the expression of autism genes remains unclear. Epidemiologic studies have not demonstrated an association between autism and the measles, mumps, and rubella vaccine<sup>12</sup> or exposure to thimerosal in vaccines.<sup>13</sup>

In a small percentage of children, autism is a feature of an underlying medical condition. For example, a boy with autism, intellectual disability, macrocephaly, and large pinnae should be tested for fragile X syndrome, which is known to account for approximately 1 percent of all cases of autism. Heferral to a medical geneticist should be considered when a child with autism has intellectual disability, dysmorphic physical findings, or a family history of ASD or intellectual disability.<sup>2</sup>

## **Surveillance and Screening**

Evidence shows that early treatment is beneficial; therefore, early diagnosis of autism is critical.<sup>15</sup> Physicians who routinely perform developmental surveillance and use appropriate screening tools increase the chances of an early diagnosis.<sup>16</sup> Surveillance entails asking parents at every well-child visit about developmental or behavioral concerns, observing for early signs of autism, and documenting a family history of ASDs. Screening involves administering an autismspecific test to all children at their 18- and 24-month office visits, not just to children who demonstrate autistic behaviors.<sup>17</sup> The Modified Checklist for Autism in Toddlers (M-CHAT; *Online Figure A*) is a screening ■ tool validated for use in children at these ages and is designed for a parent or caregiver to complete. It may be downloaded at http:// www.mchatscreen.com. The M-CHAT is a two-step screening instrument consisting of a 23-question checklist and a structured follow-up interview that is designed to identify false-positive results from the checklist.<sup>18</sup> The M-CHAT has been shown to identify a considerable number of children with ASDs who were missed by surveillance alone.18 Other primary care screening tools are also available.17,18 Although the American Academy of Pediatrics (AAP) recommends using an autism-specific screening test at 18 and 24 months of age,17 it should not take the

Condition	Features
Epilepsy	Prevalence ranges from 11 to 39 percent <sup>4</sup>
	Increased risk in girls and persons with intellectual disability
Gastrointestinal	Primarily diarrhea and constipation
problems	Associated with daytime behavioral problems <sup>5</sup>
Insomnia	Very common, associated with daytime behavioral problems <sup>6</sup>
	Includes circadian rhythm disturbance <sup>6</sup> and periodic limb movements of sleep <sup>7</sup>
Intellectual disability (formerly mental retardation)	Prevalence of 41 percent <sup>1</sup>
Motor impairments	Includes hypotonia, apraxia (motor planning), clumsiness, toe walking, gross motor delay <sup>8</sup>
Psychiatric conditions	High prevalence of anxiety, attention-deficit/ hyperactivity disorder, depression <sup>9</sup>
Sensory processing disorder	Differences in the perceptions of sights, sounds, textures, smells, and pain <sup>10</sup>

place of surveillance. Careful clinical observation may uncover subtle social deficits that parents do not report.

#### **DELAYED SOCIAL SKILL MILESTONES**

In addition to inquiring about a family history of ASDs, surveillance involves probing for early signs of autism (Table 319). Delayed attainment of social skill milestones is the earliest and most specific sign of autism. 20 The three milestones (joint attention, social orienting, and pretend play) can be quickly evaluated during an office visit. Joint attention is a child's inherent desire to share experiences with others. For example, if the physician points at a toy across the room and exclaims, "Look!", a typically developing 12- to 15-month-old child will shift his or her gaze first to the object and then back to the physician. By the 18-month visit, the child may spontaneously point at the toy and look back at his or her caregiver while smiling. This is known as declarative pointing, which serves the social purpose of experience sharing. In contrast, a child who points to an object to obtain it, known as imperative pointing, is not exhibiting joint attention because the pointing does not serve a social function. Likewise, a 24-month-old child who brings a toy to his or her father and smiles is engaging in joint attention, whereas a child who brings a jar of bubbles to his or her mother so that she will open it is not exhibiting joint attention. A lack of joint attention should prompt further evaluation.

Social orienting, or orienting to name, is another easily evaluated social skill milestone that, when absent, should prompt consideration of autism.<sup>21</sup> A typically developing 12-month-old child will turn and look in response to

hearing his or her name, whereas a child with autism may rarely or only fleetingly look, even after repeated attempts. A lack of appropriate pretend play skills is another feature of autism that can be observed in the office.<sup>22</sup> For example, a typically developing 18-month-old child will speak jargon into a parent's cell phone, whereas a child with autism may push the buttons repeatedly but not imitate the manner in which it should be used.

#### LANGUAGE IMPAIRMENT

Delayed or odd use of language is a common, but less specific, early sign of autism. Infants who do not babble (e.g., single syllables, monotone voice) by six months of age or speak jargon (e.g., multiple syllables with inflection) by nine months of age may be exhibiting early signs of autism, although autism should also be strongly considered in children 18 to

24 months of age with speech delay.<sup>17</sup> Children with autism have a diminished intrinsic drive to communicate. Unlike children with simple expressive language delay (late talkers) or those with mixed receptive and expressive language disorders, children with autism do not use pointing, gesturing, or facial expressions to compensate for their lack of spoken language. Speech, when present, is often repeated from what was just uttered to the child or heard on television (i.e., echolalia). Children with autism may also have difficulty understanding simple commands or identifying body parts. These early language deficits lead to trouble initiating and sustaining conversations. When conversations do occur, they may be one-sided or inappropriately focused on an area of intense interest. In approximately 25 percent of children with autism, there is a history of regression in language or other developmental area between 15 and 24 months of age.<sup>23</sup>

## RESTRICTED INTERESTS AND REPETITIVE BEHAVIORS

Compared with early social and language impairments, restricted interests and repetitive behaviors are less prominent and more variable in young children. Repetitive behaviors represent a continuum and may be exhibited by typically developing children. However, stereotypic movements (e.g., hand flapping), repetitive use of objects, and difficulty with changes in routine are more common and intense in children with autism.<sup>24</sup> One useful tool for physicians is the ASD Video Glossary (http://www.autismspeaks.org/video/glossary.php), which offers a side-by-side comparison of typically developing children and those with autism.

Age	Social skill probes*	Language probes	Restricted interests or repetitive behavior probes
Nine months	(–) Turning and making eye contact when hearing his or her name called (social orienting)	(-) Babbling (-) Taking turns vocalizing back and forth (-) Developing more varied vocalizations (-) Waving "bye-bye" or raising arms to be lifted (-) Responding to caregiver's voice as well as environmental sounds (+) Making unusual or high-pitched sounds	<ul> <li>(+) Carrying an unusual comfort item (e.g., hard items such as pens, stick rocks)</li> <li>(+) Demonstrating unusual or intense attachments, stereotypic movement self-injurious behaviors, or unusually severe temper tantrums with transitions or for no apparent reasor</li> <li>(+) Engaging in repetitive behaviors, such as lining objects in a row</li> </ul>
12 months	Repeat social orienting probes from nine months  (–) Turning and looking if the caregiver or physician points at an object across the room and asks the child to look (joint attention)  (–) Pointing to request an object (imperative pointing)	Repeat language probes from nine months	Repeat restricted interests or repetitive behavior probes from nine months
15 months	Repeat joint attention and imperative pointing probes from 12 months	<ul><li>(+) Repeating only what he or she hears (echolalia)</li><li>(+) Demonstrating regression in language milestones</li></ul>	Repeat restricted interests or repetitiv behavior probes from nine months
18 months	<ul><li>(–) Pointing for experience sharing (declarative pointing)</li><li>(–) Engaging in pretend play</li></ul>	Repeat language probes from 15 months	Repeat restricted interests or repetitive behavior probes from nine months
24 months	(–) Engaging in declarative pointing and showing of objects (joint attention)	Repeat language probes from 15 months	Repeat restricted interests or repetitive behavior probes from nine months

In addition to using surveillance and routine screening, a physician practice that wishes to improve its early detection of autism must develop a comprehensive plan for administering, scoring, and interpreting screening tools. Practices must also become familiar with community referral sources. Several online resources are available to help practices implement a program of developmental surveillance and screening, including two from the AAP (http://www.medicalhomeinfo.org/Screening/ and http://www.dbpeds.org/screening/).

## **Referral and Diagnosis**

Physicians should refer children for a diagnostic evaluation as soon as concerns are raised by surveillance or screening test results. Families of children with autism often feel that their early concerns were ignored, which led to excessive delays in diagnosis.<sup>25</sup> Prompt, simultaneous referrals to an audiologist, a multidisciplinary

autism team, an early intervention program (for children younger than three years), or the special education department of the local school district (for children three years and older) will prevent unnecessary delays in the diagnostic and treatment process.<sup>17</sup> Parents should be given appropriate information to prepare them for ongoing evaluations (*Table 4*).

Ideally, an interdisciplinary assessment team (*Table 5*) will evaluate a child using history, observation, and diagnostic tools to apply criteria from the *Diagnostic and Statistical Manual of Mental Disorders*, 4th ed. The diagnostic evaluation should also include a functional assessment and a review of associated conditions.

#### **Long-term Management**

Children with ASDs should have a medical home that provides accessible, continuous, comprehensive, family-centered, coordinated, compassionate, and culturally

Table 4. Online Resources for Autism Spectrum Disorders			
Organization	Description	Web site	
American Academy of Family Physicians	Information for families, includes links to other organizations	http://familydoctor.org/634.xml	
American Academy of Pediatrics	Information for families, includes links to community and professional autism-related resources	http://www.aap.org/healthtopics/autism.cfm	
Association for Science in Autism Treatment	Scientific information about autism and autism treatment, reviews of complementary and alternative medicine therapies	http://www.asatonline.org	
Autism Society of America	National autism organization with chapters throughout the United States	http://www.autism-society.org	
Autism Speaks	Autism advocacy organization with information and support for families, physicians, and researchers  Video glossary with side-by-side examples of children with typical development and children with an autism spectrum disorder	http://www.autismspeaks.org http://www.autismspeaks.org/video/glossary.php	
Centers for Disease Control and Prevention	Autism awareness campaign with materials for physicians and families Information on autism-specific screening and diagnostic tools	http://www.cdc.gov/ncbddd/autism/actearly/ http://www.cdc.gov/ncbddd/autism/screening. html	
Healing Thresholds	Updates on the scientific evidence of various autism- specific therapies	http://autism.healingthresholds.com	

effective care.26 Some parents of children with autism report dissatisfaction with the service provided by their child's physician, and physicians report major barriers and low self-perceived competency in providing these services.<sup>27</sup> In addition to listening to family members and understanding their needs, physicians should be knowledgeable about ASDs and community resources. The goals of long-term management are to increase independent functioning, improve community engagement, and provide family and caregiver support. A successful long-term management plan requires coordinating the efforts of educators, therapists, physicians, and mental health professionals. The AAP has published guidelines, created a toolkit, and developed a Web site (http://www. medicalhomeinfo.org/health/autism.html) to help physicians better meet the needs of families of children with autism.<sup>28-30</sup>

#### Treatment

Behavioral therapy using several approaches is the primary management strategy for behavioral deficits and excesses (*Table* 6<sup>31-33</sup>). It can be provided by an early intervention program, a school special education program, or by therapists in private practice. Intensive behavioral therapy (i.e., at least

## Table 5. Interdisciplinary Assessment Team for Children with Autism

Team member	Role	
Audiologist	Evaluates for hearing loss as etiology for developmental delay	
Developmental pediatrician,	Performs medical evaluation	
child neurologist, physician	Identifies and treats associated conditions	
Geneticist and genetic counselor	Performs evaluation when an underlying medica condition or genetic syndrome is suggested by family history, examination, or clinical course	
	Counsels family on recurrence risk	
Occupational therapist	Evaluates for fine and gross motor deficits	
	Evaluates for sensory processing deficits	
	Develops plan for treatment	
Psychiatrist	Evaluates and treats associated psychiatric conditions and maladaptive behaviors	
Psychologist	Administers cognitive or developmental testing	
	Administers diagnostic tools	
	Identifies associated psychiatric conditions and develops behavioral treatment plan	
Social worker	Identifies family needs	
	Refers family to formal and informal support agencies and organizations	
Speech-language pathologist	Evaluates for expressive, receptive, and pragmatic language deficits	
	Develops plan for treatment	

NOTE: To facilitate recollection of developmental milestones and behavior, parents should review baby books, records, and video recordings of their child's early years before attending a diagnostic evaluation.

Approach	Description	Evidence	Examples
Applied behavior analysis	Uses principles of operant conditioning Goals are to increase and maintain desirable behaviors and reduce maladaptive behaviors Simple skills are mastered and systematically built into more complex skills	Significant improvements in standardized measures of intelligence, language, and adaptive skills <sup>31,32</sup>	Discrete trial training Natural environment training Pivotal response training Verbal behavior training
Developmental	Relies on theories of child development Intervention is directed at establishing strong interpersonal relationships	Not supported by any controlled trials	Developmental, individual differences, relationship-based approach Relationship development intervention
Structured teaching	Eclectic elements of both developmental and behavioral methods Environments are organized with clear, concrete, visual information that is highly organized and structured	Significant improvement on standardized measure of imitation, fine motor, gross motor, and nonverbal conceptual skills <sup>33</sup>	Treatment and Education of Autistic and related Communication-handicapped Children (TEACCH Autism Program)

25 hours per week) initiated at a young age is more likely to lead to improved cognitive, language, and adaptive skill outcomes. 15,31,32 Although access to comprehensive treatment programs is currently limited, advocacy by parents and physicians has started to increase public awareness and improve access. A number of states have passed laws mandating insurance coverage for autism treatment. A recent publication entitled First 100 Days Kit (http://www.autismspeaks.org/docs/family\_services\_docs/100\_day\_kit.pdf) can help families arrange and advocate for effective early treatment.

Medication may also be used to treat behavioral and psychiatric conditions commonly associated with autism. A number of randomized controlled studies have documented the effectiveness of psychotropic medications for various maladaptive behaviors in children with autism (*Table 7* <sup>34-44</sup>).

## **Management of Associated Conditions**

Physicians should be aware of medical and psychiatric conditions associated with autism, especially sleep problems, gastrointestinal symptoms, and maladaptive behaviors. Treatment can improve overall child and family functioning. For example, treating constipation, which is common in children with ASDs, can enhance toilet training and overall behavior. <sup>45</sup> Children with ASDs often exhibit delayed sleep onset or frequent night awakenings, which can contribute to maladaptive behaviors and family distress. <sup>6</sup> Melatonin is safe and is often effective for children with autism and sleep difficulties. <sup>44</sup>

Treatment of maladaptive behaviors usually requires assistance from other members of the interdisciplinary assessment team (*Table 5*). Psychotropic medications are sometimes prescribed to treat associated behavioral

and psychiatric conditions (*Table 7*<sup>34-44</sup>). Before prescribing medications, the physician should rule out a medical cause for a new-onset maladaptive behavior. For example, a child who begins banging his head should be evaluated for evidence of a dental abscess, headaches, sinusitis, otitis media, or other potential causes of pain. Psychotropic medications may be indicated if all treatable medical conditions have been addressed, if behavioral modification is unsuccessful, and if the behavior inhibits attainment of functional goals.<sup>28</sup> Although physicians may choose to refer patients to a child psychiatrist or developmental pediatrician for the initial choice of medications, effective comanagement requires communication with the prescribing physician and awareness of potential adverse effects and drug interactions.

## **Family-Centered Care**

Autism affects everyone in the child's family. Caregivers report increased stress,<sup>46</sup> as well as financial hardships.<sup>47</sup> Delivery of family-centered services can improve patient and family outcomes.<sup>48</sup> Physicians should respect parents and patients as partners, listening and acting on their specific concerns. Longer well-child visits may allow assessment of the health and well-being of the entire family. A referral to a family-to-family network, such as Family Voices (http://www.familyvoices.org), can help parents make informed decisions, advocate for their children, and build partnerships with other families.

## Complementary and Alternative Approaches

Although many complementary and alternative medicine (CAM) therapies lack proven effectiveness,<sup>49</sup> physicians should recognize that most families of children with ASDs are likely to try at least one CAM approach.<sup>25</sup> Parents of

Table 7. Medication Options for Target Symptoms Often Associated with Autism

Target symptoms	Medication class	Examples*
Aggression, explosive outbursts, irritability,	Atypical antipsychotic	Aripiprazole (Abilify), <sup>35</sup> risperidone (Risperdal)† <sup>36</sup>
self-injury	SSRI	Fluvoxamine <sup>37</sup>
Anxiety	SSRI	Fluoxetine (Prozac)38
Behavioral rigidity,	Atypical antipsychotic	Risperidone† <sup>36</sup>
obsessive-compulsive symptoms, repetitive behavior	SSRI	Fluoxetine, <sup>39</sup> fluvoxamine <sup>37</sup>
Hyperactivity, impulsivity,	Alpha-2 agonist	Clonidine (Catapres), <sup>40</sup> guanfacine (Tenex) <sup>41</sup>
inattention	Selective norepinephrine reuptake inhibitor	Atomoxetine (Strattera) <sup>42</sup>
	Stimulant	Methylphenidate (Ritalin)43
Sleep dysfunction	Endogenous neurohormone	Melatonin <sup>44</sup>

SSRI = selective serotonin reuptake inhibitor.

Information from references 34 through 44.

children with autism who are interested in CAM are often disappointed with their physicians' lack of knowledge or negative attitudes about CAM. Physicians can foster a trusting relationship by asking, in a nonjudgmental way, about CAM and listening to the parents' perceptions regarding the benefits of a particular treatment. Once trust is established, the physician can help families distinguish validated treatment approaches from treatments that have been proven ineffective or those that are unproven and potentially harmful.<sup>28</sup> Several Web sites can assist physicians with staying up to date on new treatments (*Table 4*).

## **Prognosis**

Longitudinal research suggests that up to one half of adults with autism who have average or near-average cognitive ability can achieve a generally high level of independence in work and home life. Although interest in developing social relationships often increases as persons with autism age, relatively few adults marry or develop truly reciprocal relationships. Many adults with a child-hood diagnosis of autism remain impaired, especially those with poorer cognitive functioning. Associated psychiatric conditions—including anxiety, mood, psychotic, and attention disorders—are a challenge for many adults with autism. However, treating these associated conditions can improve overall functioning. Additionally, miscommunication, peer pressure to engage in unlawful

activity, obsessional behavior, reactions to bullying, and misunderstanding of social proprieties increase this population's risk of encounters with law enforcement.<sup>51</sup>

Improvements in service quality and availability are likely to produce better outcomes for children diagnosed today. Communicative phrase speech, early acquisition of joint attention skills, early intervention, and a childhood IQ of 70 or higher are important factors associated with obtaining a high level of independence in adulthood. <sup>15,52</sup> With earlier recognition and diagnosis, more intensive treatment, and increased acceptance and social support for families, physicians caring for children with autism can now be more optimistic regarding their prognosis.

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<sup>\*—</sup>At least one randomized controlled trial supporting effectiveness in patients with autism spectrum disorders.

<sup>†—</sup>Aripiprazole and risperidone are the only current medications with U.S. Food and Drug Administration—approved labeling specific to autism (for the symptomatic treatment of irritability, including aggressive behavior, deliberate self-injury, and tantrums in children and adolescents with autism).

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