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 Pediatrics 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.)
### 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:
   a. Marked impairment in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body posture, and gestures to regulate social interaction
   b. Failure to develop peer relationships appropriate to developmental level
   c. 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 imaginative play appropriate to developmental level

3. 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.

Features who points to an object to obtain it, known as imperata-  

tion, 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. A typically develop-

ing 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. 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 inflec-


tion) 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. 

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 expres-

sive 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. 

**RESTRICTED INTERESTS AND REPETITIVE BEHAVIORS** 

Compared with early social and language impairments, restricted interests and repetitive behaviors are less prom-

inent 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. 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. 

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**Table 2. Conditions Associated with Autism**

<table>
<thead>
<tr>
<th>Condition</th>
<th>Features</th>
</tr>
</thead>
<tbody>
<tr>
<td>Epilepsy</td>
<td>Prevalence ranges from 11 to 39 percent&lt;sup&gt;4&lt;/sup&gt;</td>
</tr>
<tr>
<td></td>
<td>Increased risk in girls and persons with intellectual disability</td>
</tr>
<tr>
<td>Gastrointestinal problems</td>
<td>Primarily diarrhea and constipation</td>
</tr>
<tr>
<td></td>
<td>Associated with daytime behavioral problems&lt;sup&gt;1&lt;/sup&gt;</td>
</tr>
<tr>
<td>Insomnia</td>
<td>Very common, associated with daytime behavioral problems&lt;sup&gt;6&lt;/sup&gt;</td>
</tr>
<tr>
<td></td>
<td>Includes circadian rhythm disturbance&lt;sup&gt;6&lt;/sup&gt; and periodic limb movements of sleep&lt;sup&gt;6&lt;/sup&gt;</td>
</tr>
<tr>
<td>Intellectual disability</td>
<td>Prevalence of 41 percent&lt;sup&gt;1&lt;/sup&gt;</td>
</tr>
<tr>
<td>(formerly mental retardation)</td>
<td></td>
</tr>
<tr>
<td>Motor impairments</td>
<td>Includes hypotonia, apraxia (motor planning), clumsiness, toe walking, gross motor delay&lt;sup&gt;8&lt;/sup&gt;</td>
</tr>
<tr>
<td>Psychiatric conditions</td>
<td>High prevalence of anxiety, attention-deficit/hyperactivity disorder&lt;sup&gt;9&lt;/sup&gt;</td>
</tr>
<tr>
<td>Sensory processing disorder</td>
<td>Differences in the perceptions of sights, sounds, textures, smells, and pain&lt;sup&gt;19&lt;/sup&gt;</td>
</tr>
</tbody>
</table>

Information from references 1 and 4 through 10.
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.25 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.17 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

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**Table 3. Clinical Probes for Autism Surveillance**

<table>
<thead>
<tr>
<th>Age</th>
<th>Social skill probes*</th>
<th>Language probes</th>
<th>Restricted interests or repetitive behavior probes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Nine months</td>
<td>(–) Turning and making eye contact when hearing his or her name called (social orienting)</td>
<td>(–) Babbling</td>
<td>(–) Carrying an unusual comfort item (e.g., hard items such as pens, sticks, rocks)</td>
</tr>
<tr>
<td></td>
<td>(–) Taking turns vocalizing back and forth</td>
<td>(–) Taking turns vocalizing back and forth</td>
<td>(–) Demonstrating unusual or intense attachments, stereotypic movements, self-injurious behaviors, or unusually severe temper tantrums with transitions or for no apparent reason</td>
</tr>
<tr>
<td></td>
<td>(–) Developing more varied vocalizations</td>
<td>(–) Waving “bye-bye” or raising arms to be lifted</td>
<td>(–) Engaging in repetitive behaviors, such as lining objects in a row</td>
</tr>
<tr>
<td></td>
<td>(–) Responding to caregiver’s voice as well as environmental sounds</td>
<td>(–) Waving “bye-bye” or raising arms to be lifted</td>
<td>(–) Engaging in repetitive behaviors, such as lining objects in a row</td>
</tr>
<tr>
<td></td>
<td>(+) Making unusual or high-pitched sounds</td>
<td>(+) Making unusual or high-pitched sounds</td>
<td>(–) Engaging in repetitive behaviors, such as lining objects in a row</td>
</tr>
</tbody>
</table>

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*—Delayed attainment of social skill milestones is the earliest and most specific sign of autism.
(+)—The presence of this behavior suggests autism.
(–)—The absence of this behavior suggests autism.

Information from reference 19.
effective care. 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. 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 tool kit, 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.

Table 4. Online Resources for Autism Spectrum Disorders

<table>
<thead>
<tr>
<th>Organization</th>
<th>Description</th>
<th>Web site</th>
</tr>
</thead>
<tbody>
<tr>
<td>American Academy of Family Physicians</td>
<td>Information for families, includes links to other organizations</td>
<td><a href="http://familydoctor.org/634.xml">http://familydoctor.org/634.xml</a></td>
</tr>
<tr>
<td>American Academy of Pediatrics</td>
<td>Information for families, includes links to community and professional autism-related resources</td>
<td><a href="http://www.aap.org/healthtopics/autism.cfm">http://www.aap.org/healthtopics/autism.cfm</a></td>
</tr>
<tr>
<td>Association for Science in Autism Treatment</td>
<td>Scientific information about autism and autism treatment, reviews of complementary and alternative medicine therapies</td>
<td><a href="http://www.asatonline.org">http://www.asatonline.org</a></td>
</tr>
<tr>
<td>Autism Society of America</td>
<td>National autism organization with chapters throughout the United States</td>
<td><a href="http://www.autism-society.org">http://www.autism-society.org</a></td>
</tr>
<tr>
<td>Autism Speaks</td>
<td>Autism advocacy organization with information and support for families, physicians, and researchers</td>
<td><a href="http://www.autismspeaks.org">http://www.autismspeaks.org</a></td>
</tr>
<tr>
<td>Centers for Disease Control and Prevention</td>
<td>Autism awareness campaign with materials for physicians and families</td>
<td><a href="http://www.cdc.gov/ncbddd/autism/actearly/">http://www.cdc.gov/ncbddd/autism/actearly/</a></td>
</tr>
<tr>
<td></td>
<td>Information on autism-specific screening and diagnostic tools</td>
<td><a href="http://www.cdc.gov/ncbddd/autism/screening.html">http://www.cdc.gov/ncbddd/autism/screening.html</a></td>
</tr>
<tr>
<td>Healing Thresholds</td>
<td>Updates on the scientific evidence of various autism-specific therapies</td>
<td><a href="http://autism.healingthresholds.com">http://autism.healingthresholds.com</a></td>
</tr>
</tbody>
</table>

Table 5. Interdisciplinary Assessment Team for Children with Autism

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

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.

Treatment

Behavioral therapy using several approaches is the primary management strategy for behavioral deficits and excesses (Table 6). 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...
25 hours per week) initiated at a young age is more likely to lead to improved cognitive, language, and adaptive skill outcomes.\textsuperscript{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\textsuperscript{34-44}).

### 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.\textsuperscript{45} Children with ASDs often exhibit delayed sleep onset or frequent night awakenings, which can contribute to maladaptive behaviors and family distress.\textsuperscript{6} Melatonin is safe and is often effective for children with autism and sleep difficulties.\textsuperscript{44}

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\textsuperscript{34-44}). 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.\textsuperscript{28} 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,\textsuperscript{46} as well as financial hardships.\textsuperscript{47} Delivery of family-centered services can improve patient and family outcomes.\textsuperscript{48} 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,\textsuperscript{49} physicians should recognize that most families of children with ASDs are likely to try at least one CAM approach.\textsuperscript{25} Parents of

<table>
<thead>
<tr>
<th>Approach</th>
<th>Description</th>
<th>Evidence</th>
<th>Examples</th>
</tr>
</thead>
<tbody>
<tr>
<td>Applied behavior analysis</td>
<td>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.</td>
<td>Significant improvements in standardized measures of intelligence, language, and adaptive skills\textsuperscript{31,32}</td>
<td>Discrete trial training, Natural environment training, Pivotal response training, Verbal behavior training.</td>
</tr>
<tr>
<td>Developmental</td>
<td>Relies on theories of child development. Intervention is directed at establishing strong interpersonal relationships.</td>
<td>Not supported by any controlled trials.</td>
<td>Developmental, individual differences, relationship-based approach, Relationship development intervention.</td>
</tr>
<tr>
<td>Structured teaching</td>
<td>Eclectic elements of both developmental and behavioral methods. Environments are organized with clear, concrete, visual information that is highly organized and structured.</td>
<td>Significant improvement on standardized measure of imitation, fine motor, gross motor, and nonverbal conceptual skills\textsuperscript{53}</td>
<td>Treatment and Education of Autistic and related Communication-handicapped Children (TEACCH Autism Program).</td>
</tr>
</tbody>
</table>

Information from references 31 through 33.
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. 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 childhood 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, obsessive behavior, reactions to bullying, and misunderstanding of social proprieties increase this population’s risk of encounters with law enforcement.

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. 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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### REFERENCES


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

<table>
<thead>
<tr>
<th>Target symptoms</th>
<th>Medication class</th>
<th>Examples*</th>
</tr>
</thead>
<tbody>
<tr>
<td>Aggression, explosive outbursts, irritability, self-injury</td>
<td>Atypical antipsychotic</td>
<td>Aripiprazole (Abilify)†, risperidone (Risperdal)†</td>
</tr>
<tr>
<td></td>
<td>SSRI</td>
<td>Fluoxetine (Prozac)‡</td>
</tr>
<tr>
<td>Anxiety</td>
<td>SSRI</td>
<td>Fluoxetine (Prozac)‡</td>
</tr>
<tr>
<td>Behavioral rigidity, obsessive-compulsive symptoms, repetitive behavior</td>
<td>Atypical antipsychotic</td>
<td>Risperidone (Risperdal)†</td>
</tr>
<tr>
<td></td>
<td>SSRI</td>
<td>Fluoxetine, fluvoxamine‡</td>
</tr>
<tr>
<td>Hyperactivity, impulsivity, inattention</td>
<td>Alpha-2 agonist</td>
<td>Clonidine (Catapres)‡</td>
</tr>
<tr>
<td></td>
<td>Selective norepinephrine reuptake inhibitor</td>
<td>Atomoxetine (Strattera)‡</td>
</tr>
<tr>
<td>Sleep dysfunction</td>
<td>Stimulant</td>
<td>Methylphenidate (Ritalin)‡</td>
</tr>
<tr>
<td></td>
<td>Endogenous neurohormone</td>
<td>Melatonin‡</td>
</tr>
</tbody>
</table>

SSRI = selective serotonin reuptake inhibitor.

*—At least one randomized controlled trial supporting effectiveness in patients with autism spectrum disorders.

†—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).

Information from references 34 through 44.
Autism