Autistic disorder, a pervasive developmental disorder resulting in social, language, or sensorimotor deficits, occurs in approximately seven of 10,000 persons. Early detection and intervention significantly improve outcome, with about one third of autistic persons achieving some degree of independent living. Indications for developmental evaluation include no babbling, pointing, or use of other gestures by 12 months of age, no single words by 16 months of age, no two-word spontaneous phrases by 24 months of age, and loss of previously learned language or social skills at any age. The differential diagnosis includes other psychiatric and pervasive developmental disorders, deafness, and profound hearing loss. Autism is frequently associated with fragile X syndrome and tuberous sclerosis, and may be caused by lead poisoning and metabolic disorders. Common comorbidities include mental retardation, seizure disorder, and psychiatric disorders such as depression and anxiety. Behavior modification programs are helpful and are usually administered by multidisciplinary teams; targeted medication is used to address behavior concerns. Many different treatment approaches can be used, some of which are unproven and have little scientific support. Parents may be encouraged to investigate national resources and local support networks. (Am Fam Physician 2002;66:1667-74,1680. Copyright© 2002 American Academy of Family Physicians.)
ductive movements of hands and fingers, rocking, meaningless vocalizations), self-stimulation, self-injury behaviors, and seizures. Mental retardation is not a diagnostic criterion, but it is frequently present in the moderate to severe range.

**Epidemiology**

In general, pervasive developmental disorders are estimated to occur at a rate of 63 per 10,000 persons.\(^3\) While the reported incidence of autistic disorder ranges from about five per 10,000\(^4\) to 20 per 10,000 persons,\(^5\) a recent meta-analysis reports the median rate for 11 surveys conducted since 1989 to be seven per 10,000 persons.\(^6\) Male-to-female ratios vary with IQ scores from 2:1 in severely handicapped persons to 4:1 in moderately handicapped persons.\(^7\) [Evidence level B, non-randomized studies] The occurrence rate in siblings is suspected to be from 3 to 7 percent, representing a 50- to 100-fold increase in risk.\(^8\)

**Etiology**

No single cause has been identified for the development of autism. Genetic origins are suggested by studies of twins and a higher
incidence of recurrence among siblings.9 In addition, an increased frequency of occurrence is noted in patients with genetic conditions such as fragile X syndrome and tuberous sclerosis.10 Some reports have suggested a possible association with Down syndrome.11

In addition to the implication of neurotransmitters, such as serotonin, in the development and expression of autism,12 many other disorders may result in brain dysfunction. Possible contributing factors in the development of autism include infections, errors in metabolism, immunology, lead poisoning, and fetal alcohol syndrome.13

Concerns have been raised in recent years that immunizations, particularly measles, mumps, and rubella (MMR) vaccine, may precipitate autism. In addition to reports from several parents who first detected autism in their children following an MMR vaccination at 12 to 15 months of age, an anecdotal study14 reported similar suspicions on the part of physicians who provided care for 12 autistic patients. Subsequent studies in the United Kingdom15,16 [reference 16, Evidence level B: epidemiologic study] and the United States17 [Evidence level B: epidemiologic study] have failed to show an association between any vaccine and the development of autism. Information about ongoing studies being conducted by the Centers for Disease Control and Prevention and the National Institutes of Health (NIH) is available at their Web sites (Table 3).

### Recognition and Screening

Indications for formal developmental evaluation include no babbling, pointing, or other gestures by 12 months of age, no single words by 16 months of age, no two-word spontaneous phrases by 24 months of age, and loss of previously learned language or social skills at any age.18 Parental concerns about delayed speech and language development, typically noticed at about 18 months of age, should always be taken seriously.

Including developmental surveillance as a routine part of the well-child examination can enhance recognition of developmental disorders. While the Denver screening tools19 have historically been used in primary care settings for routine developmental surveillance, they lack the sensitivity and specificity necessary for use as screening tools for developmental disorders.20 More specific and sensitive screening surveillance tools, such as the Parents’ Evaluation of Developmental Status (PEDS),21 are available for assessing these conditions. Screening tools that are specific for autism include the Checklist for Autism in Toddlers

<table>
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<th>TABLE 3</th>
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<tr>
<td>Resources for Management of Autism</td>
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| Centers for Disease Control and Prevention  
National Immunization Program  
Web address: www.cdc.gov/nip |
| --- |
| National Institutes of Health, National Institute of  
Child Health and Human Development  
Web address: www.nichd.nih.gov |
| Parents’ Evaluation of Developmental Status (PEDS),  
Ellsworth & Vandermeer Press  
Telephone: 615-226-4460  
Web address: www.pedtest.com/test/peds_manual.html |
| Checklist for Autism in Toddlers (CHAT)  
Web address: www.nas.org.uk/profess/chat.html |
| Pervasive Developmental Disorders Screening  
Test-Stage I (PDDST), Porter Psychiatric Institute  
Telephone: 415-476-7385 |
| Association of University Centers on Disabilities,  
a listing of professionals by state  
Telephone: 301-588-8252  
Web address: www.aucd.org |
| The National Institute of Child Health and Human Development, a listing of research centers investigating treatment strategies for autism  
Web address: www.nichd.nih.gov |
| Autism Society of America  
Telephone: 800-3AUTISM (800-328-8476)  
Web address: www.autism-society.org |
| Center for the Study of Autism  
Web address: www.autism.org |
(CHAT)\(^2\) (Table 4\(^2\)) and the Pervasive Developmental Disorders Screening Test-Stage I (PDDST).\(^4\) For a comprehensive review of available screening tools, the authors recommend an article by Filipek and colleagues.\(^1\)

When an autistic disorder is suspected, referral should be made for further developmental evaluation and cognitive testing. Although there is currently no cure for autism, early diagnosis and initiation of structured multidisciplinary intervention can significantly enhance functioning in later life.\(^2\) Experienced clinicians can reliably diagnose autism in children younger than three years and, frequently, as young as two years. Presently no biologic markers are available to identify patients with autistic disorders. Useful resources for identifying clinicians with expertise in the diagnosis of autism include the University Affiliated Program system and the National Institute of Child Health and Human Development (Table 3).

Once an autistic disorder is suspected, certain medical evaluations should be performed. A family history of limited cognitive abilities or the presence of dysmorphic features may suggest the need for genetic evaluation. Wood's light examination of the skin should be performed to help identify the depigmented macules of tuberous sclerosis. Lead screening and metabolic testing should be considered if there is a history of lethargy, cyclic vomiting, early seizures, dysmorphic features, or mental retardation. Electrophysiologic testing such as electroencephalography and central nervous system imaging studies are warranted to evaluate neurologic features that cannot be explained by the diagnosis of autism alone.\(^4\) Because deafness or profound hearing loss can cause symptoms mimicking autism, a formal hearing evaluation should be given if the diagnosis of autism is being considered.

Autistic disorders should be distinguished from other psychiatric and pervasive developmental disorders. Table 5\(^1\) lists a differential diagnosis for several similar disorders.

### Clinical Course

The typical presenting symptoms of autistic disorder are delayed speech or challenging behavior before three years of age.\(^7\) Although parents frequently see these signs and suspect that something is wrong with their child by 18 months of age, a diagnosis of autism is frequently delayed by two to three years because of reluctance on the part of clinicians and families to incorrectly label a child as autistic.\(^2\) Seventy-five percent of autistic persons have some level of mental retardation.\(^1\) Developmental gains in childhood and adolescence are common, but some persons have behavioral regression during adolescence.

Low IQ scores and failure to develop communicative language by five years of age correlate positively with a poor prognosis for
response to treatment. About one third of autistic persons can achieve some degree of independent living, although fewer than 5 percent go on to become self-sufficient adults. Development of stereotypic behavior, self-injury behavior, and selective attention toward distracting stimuli (e.g., a ticking clock) markedly interfere with structured learning and working environments.

Many autistic persons develop seizures in their first year of life in the form of infantile spasms, a particularly severe form of seizure that is difficult to treat. There is also a significant incidence of first occurrence of seizures during adolescence, and as many as 35 percent may develop seizures by adulthood. Comorbid anxiety is common, as are depression and obsessional behavior.

Management of Autism and Comorbid Conditions

The general goals of treatment for autistic patients are to improve language and social skills, decrease problem behaviors, support parents and families in their adjustment to and education of autistic children, and foster independence. Because autistic children who begin treatment at a young age have significantly better outcomes, early intervention is critical. Public Law 99-457 and the Individuals with Disabilities Education Act mandate referral to the special services departments of local preschool or school systems.

Because no treatment protocol meets the needs of every autistic child, it is helpful to get suggestions from a variety of sources. Organizations available to help families and educators are listed in Table 3.

Neuropharmacologic Treatment

Primary care physicians are commonly asked to address the stereotypic or disruptive behaviors of autistic patients. While numerous medications have been used to treat autistic symptoms, no single medication has been shown to be universally effective. Historically, psychotropic medications have been reserved for use in situations where all attempts at behavior management have failed, and the patients are considered to be harmful to themselves or others.

While use of behavior modification programs is often the primary method of managing challenging behaviors in autistic children, supportive medication use has been found to help reduce behavior problems. Obtaining a correct diagnosis is important before initiating any pharmacologic intervention. For example, attention deficit with or without hyperactivity can coexist with autism and may possibly be managed with the use of methylphenidate (Ritalin) [Evidence level C: consensus opinion] or clonidine (Catapres). It can be difficult to distinguish between the behaviors associated with autism; attention-deficit/hyperactivity disorder; and mania, and an appropriate treatment for one disorder may be ineffective or exacerbate the symptoms of another.
When behavior management programs or the use of supportive medications are unsuccessful in correcting potentially dangerous behavior, the use of sedating medications may be necessary for brief periods while less invasive interventions are attempted. Sedative-hypnotics and neuroleptics such as buspirone (BuSpar), in a dosage of 5 to 20 mg two to three times a day[^32] [Evidence level C: consensus opinion], or risperidone (Risperdal), in a dosage of 0.5 to 2 mg twice a day[^33] are commonly used for this purpose. Benzodiazepines should be used with caution because they can cause disinhibition, resulting in more excitable behavior.[^32]

Objective data collection and outcome monitoring are important because of the variable nature of individual responses to medication. Information should be collected by persons who have regular contact with the patient—family members, support personnel in day care and residential programs, case managers, and physicians. Given the multiple developmental, behavior, and medical problems associated with autism, coordination of services by a multidisciplinary team is highly recommended.[^34] [Evidence level C: consensus opinion]

**ALTERNATIVE THERAPIES**

A number of methods for teaching communication and socialization skills have been developed over the years. One recent example is augmented communication, a method whereby nonverbal persons are assisted in communication by means of a letter board or a computer keyboard. When a facilitator helps the person choose letters, words, etc., the process is referred to as facilitated communication. While augmented communication devices have markedly improved communication potential in some patients, numerous controlled studies have failed to show that facilitated communication is reliably useful.[^13]

Another treatment currently being advocated is auditory integration training (AIT), whereby persons with autism typically spend 10 hours during a two-week period listening to music that has been computer-modified to remove sensitive frequencies and reduce predictable patterns. AIT is said to improve auditory processing capabilities by correcting distortions in hearing and by conditioning patients to focus their attention more appropriately. Unfortunately, this technique also has little supporting scientific documentation.[^35]

Another popular behavior-based intervention is the Lovaas program[^36], sometimes referred to as DTT because of its use of positive reinforcement through a series of intensive discrete trial training sessions. While initial reports suggested a 47 percent recovery rate from autism when preschoolers were treated,[^36] subsequent studies have been unable to document long-term gains.[^37] Studies using similar behavior interventions, however, have been able to document short-term improvements.[^37] Verification of long-term success becomes important in view of the cost of such intensive treatment programs, which typically require extensive one-on-one training with autistic children for 40 hours a week for a minimum of two years—a cost of approximately $40,000 a year.[^38] Controversies about fiscal responsibility are ongoing because some parents feel local school systems should make this level of care available for their children.[^38]

Other interventional strategies include deep pressure therapy, nutritional supplements, and specialty diets[^39-42] [reference 40, Evidence

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Anecdotal reports of success with alternative or complementary interventions are common, but efficacy in most cases remains clinically unproven. Generally speaking, most successful programs have several common components: (1) recognition of the importance of early identification and intervention; (2) use of behavior-oriented strategies; (3) development of social communication; and (4) active involvement of parents and families.

Recently, there has been discussion about a possible role of the gastric hormone secretin as a pharmacologic intervention in the treatment of autism. This information was based on one study of three autistic persons. Unfortunatley, a subsequent study involving 56 autistic persons failed to support the initial findings. At present, most investigators do not see a role for secretin in the treatment of autism, an opinion supported by ongoing research at the NIH.

The authors indicate that they do not have any conflicts of interest. Sources of funding: none reported.

level A: randomized controlled trial (RCT); reference 42, Evidence level C: consensus opinion

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<td>Behavior Modification in the Management of Autism</td>
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<td>Structuring the environment</td>
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<tr>
<td>Providing consistent responses to behaviors</td>
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<tr>
<td>Positive reinforcement—rewarding a desired behavior</td>
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<td>Negative reinforcement—not rewarding an undesirable behavior</td>
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<tr>
<td>Punishment—application of an adverse stimulus to deter an unwanted response</td>
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<tr>
<td>Shaping—reinforcing closer and closer approximations to the desired behavior</td>
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level B: lower quality RCT; reference 43, Evidence level C: consensus opinion [Evidence level B: lower quality RCT] At present, most investigators do not see a role for secretin in the treatment of autism, an opinion supported by ongoing research at the NIH.

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Autism