

# Speak the language of autism



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**A**utism affects 1 in 110 children and 1 in 70 boys in the United States.<sup>1</sup> This represents an astounding 57% increase from 2002 to 2006 and a 600% increase in the past 20 years.<sup>1</sup> The reasons for this drastic increase are being debated in literature. Some experts claim that autism is becoming more common, whereas others attribute the increase to widening diagnostic criteria, greater awareness among parents and healthcare professionals, and improved case finding methods. Autism is one of a group of disorders known as autism spectrum disorders (ASDs). Autism is characterized by impairment in reciprocal social interaction, delayed and/or stereotyped communication, and restricted or repetitive behaviors and interests.<sup>2</sup> The extent of the signs and symptoms vary from mild to severe. Medical professionals previously believed that as many as 70% of those diagnosed with autism also had mental retardation; however, recent studies show that nearly half of children with autism have an average or better intelligence quotient (IQ).<sup>3</sup>

Autism manifests in early childhood (usually between 12 to 24 months) and continues throughout adulthood.<sup>4</sup> Specific underlying causes cannot be identified in the majority of children with autism. Although there are numerous and varied treatment programs available, many controversies surround the etiology and treatment of autism, which can cause confusion among families about the best course of action. Recent research has emphasized the importance of early identification of autism by healthcare providers.<sup>4</sup> Early identification enhances the success of treatment programs and is essential for improving long-term outcomes. The purpose of this article is to explore the epidemiology of autism, discuss contributing factors and diagnostic criteria, investigate various treatments, and suggest future recommendations for practitioners.

### ■ Epidemiology

The epidemiology of autism is difficult to determine. Researchers have proposed several events that may lead to the development of autism, such as a neurodevelopmental error

that occurs before birth, a genetic disturbance, a metabolic disorder that occurs after birth, an autoimmune dysfunction from environmental toxins, or childhood vaccines.<sup>8</sup> Furthermore, diagnostic criteria have been revised in recent years. Early epidemiologic studies suggested a prevalence of 4 to 10 affected individuals per 10,000 children. Recent population-based surveys have shown a much higher prevalence: 40 to 60 cases per 10,000 children.<sup>4</sup> There has been much publicity about the increased prevalence of autism recorded in the last 20 to 30 years. With the discovery of new diagnostic criteria, there are an estimated 4.5 and 9.5 per 1,000 8-year-old children and roughly 560,000 persons between birth and 21 years of age diagnosed with autism.<sup>4</sup> The CDC Autism and Developmental Disabilities Monitoring (ADDM) released data in 2007 that found 1 in 150 8-year-old children in various areas in the United States have been diagnosed with an ASD.<sup>5</sup> (See *ADDM network sites*.)

### ■ Theorized causative factors

#### Genetics

There is strong and convincing evidence that genetic factors may be the primary cause of autism. Numerous studies have been conducted and articles published that support the supposition of a genetic link in ASDs.<sup>6</sup> Many researchers have hypothesized that because epidemiologic studies report a male-to-female ratio of 4:1 of autism, there must be a link to the X chromosome. However, conflicting data between two different research groups has ruled out X linkage as the predominant mode due to the occurrence of male-to-male transmission in a number of families.<sup>4</sup> The rate of occurrence in siblings is 2% to 8% greater than in the general population. Conversely, the percentage is much lower than would be expected for single gene disease. The National Institutes of Health (NIH) concurred that autism was one of the more heritable mental disorders and found that if one identical twin had autism the other did as well in nearly 9 of 10 instances.<sup>7</sup>

Five to ten percent of diagnosed autism cases have a linkage between abnormalities on chromosomes 4 and



7 and ASDs.<sup>6</sup> Population and case reports have found abnormalities on the 15q11-q13, 15q11.2, and 16p11.2 loci.<sup>6</sup> Duplications, inversions, or deletions in this area can account for language delays, ataxia, and mental retardation in 2% to 3% of autistic children.<sup>6</sup> In 2005, scientists isolated a version of a gene located on chromosome 7 that had been associated with autism in families with more than one autistic child.<sup>8</sup> Currently, researchers at the Massachusetts Institute of Technology have pinpointed two genes, PTEN and the serotonin transporter gene in mice, that influence autism-like symptoms such as increased brain size and sociability in mice.<sup>9</sup>

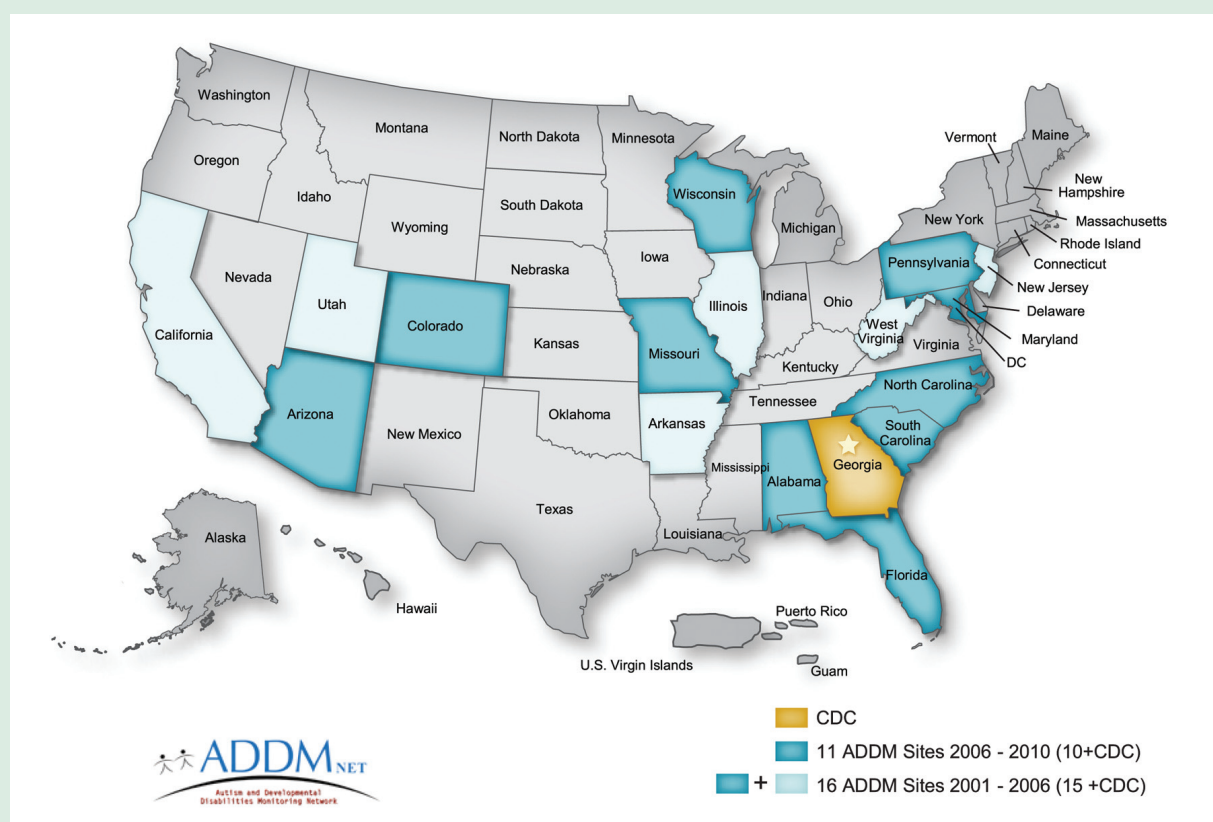
Additional research suggests that autism is a multifactorial disease in which several genes combine and are manipulated by environmental factors to produce clinical symptoms.<sup>10</sup> The University of California examined brain development through the use of magnetic resonance imaging (MRI) in 60 autistic children ages 2 to 16 years and found that 90% of these children in the 2- to 4-year age group had a brain volume larger than average for the

control group and had increased cerebral gray and white matter possibly due to the increased number of neurons (VIP, BDNF, NT-4, CGRP).<sup>10</sup> These findings enabled the researchers to attribute the abnormal neural growth to autism and propose that this may account for the varying degrees of symptoms in children.<sup>10</sup> Furthermore, positron emission tomography studies have shown that children with autism have global and functional abnormalities in serotonin synthesis.<sup>11</sup> Neuropathic studies have further demonstrated a link between autism and a decrease in the number of Purkinje cells. A decrease in Purkinje cells would display changes in a child's language processing, anticipatory and motor planning, mental imagery, and timed sequencing.<sup>11</sup>

### Environment

Researchers have identified environmental factors such as toxic exposures, teratogens, perinatal insults, and prenatal infections that can potentially influence environmentally responsive genes and may make them key participants in the etiology of autism. However, the effects of environmental

### ADDM network sites



Source: CDC. *Autism and Developmental Disabilities Monitoring Network*. <http://www.cdc.gov/ncbddd/autism/states/ADDMMCommunityReport2009.pdf>.

exposures have been shown to depend on the timing and duration of exposure, strength of the toxin, mechanism of action, and distribution in the central nervous system.<sup>12</sup> In one study, a correlation was made between the rise in autism and the decrease in sun exposure during pregnancy. The study showed that since 90% of vitamin D production comes from skin, many pregnant women were getting minimal amounts of vitamin D from their prenatal vitamins.<sup>13</sup> Another study examined the brains of rats that received small amounts of oral vitamin D. The rats had increased brain mass and ventricular lumen size, which is also characteristic of children with autism.<sup>13</sup> CDC data from a study in 14 states linked autism with decreased UVB exposure. New Jersey had the highest prevalence of children with autism, and Alabama had the lowest.<sup>13</sup> Other studies have inversely correlated the consumption of vitamin D-rich fish eaten during pregnancy with the incidence of autism. Results showed that mothers who ate the most vitamin D-rich fish had infants with better cognition than those who did not.<sup>13</sup>

Additional research has implied a possible link between the measles, mumps, and rubella (MMR) vaccination and developing autism. A connection was established between the MMR vaccine and autism due to signs of autism becoming evident around 12 to 18 months of age when the vaccine is routinely administered.<sup>1</sup> However, more than 20 epidemiologic research studies have disproved this association. A report by the Institute of Medicine (IOM) in 2004 concluded that there is no link between autism and the MMR vaccine. In 2006, the CDC along with the National Institute of Child Health and Human Development of the National Institutes of Health assessed data from 351 children with ASDs and 31 normally developing children. The results of the study did not find a link between the MMR vaccine and autism.<sup>5</sup> A study in 2008, published in the Public Library of Sciences, was conducted to confirm whether the measles virus RNA was in the intestinal tissue of a specific group of autistic children. This study concluded that the measles virus RNA was not in the intestinal tissue and that no link between the MMR vaccine and autism was found.<sup>14</sup> Vaccine experts at the American Academy of Pediatrics (AAP) and the CDC agree that the MMR vaccine is not responsible for the rise in the number of children with autism.<sup>5</sup>

### ■ Diagnostic criteria

The *Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition (DSM-IV)* includes autistic disorder in the category of pervasive developmental disorders along with pervasive developmental disorder, not otherwise specified

(PDD-NOS), Asperger disorder, Rett disorder, and childhood disintegrative disorder.<sup>30</sup> Asperger disorder, PDD-NOS, and autistic disorder are often collectively referred to as the ASDs.<sup>15</sup> The grouping of the above disorders suggests that these terms are related, which causes confusion in the ability to differentiate between them, making the definitive diagnosis of autism challenging, especially in children between the ages of 12 and 24 months. There are no lab tests to detect autism and the symptoms are generally broad variations of psychomotor, behavioral, and verbal deficits. However, one of the earliest and most important signs of

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autism is failure to develop joint attention. Joint attention refers to a child's degree of ability in sharing interests, pleasurable experiences, or requests by using gestures or verbal communication in combination with eye contact with another person.<sup>15</sup> The diagnostic criteria for autism require the presence of six symptoms from three categories: impaired reciprocal social interaction, impaired communication, and restricted, repetitive, or stereotyped behaviors.<sup>15</sup> Parents usually begin to express concern about their child's developmental process before age 3; however the average diagnosis is not made until 53 months. There are several diagnostic scales that help validate the identification of autism.

The most widely used and accepted diagnostic tool is the Childhood Autism Rating Scale (CARS). The CARS is a standardized observational instrument that rates children on 15 symptoms divided into four domains: communication, sensory sensitivities, emotional response, and socialization.<sup>17</sup> The diagnostic algorithm classifies children from mild to severe autism, yields a total score to continuously gauge the severity of autism, and has a sensitivity of 0.71 and a specificity of 0.93.<sup>17</sup> Studies have tested the reliability of CARS. In one study of 537 preschool children, the internal consistency was 0.94, and in another study of 431 children it was 0.91.<sup>18</sup> In a 2005 test-retest study of 274 preschool children (ages 2 to 6), the CARS was used to test the degree of agreement between it and the *DSM-IV*.<sup>18</sup> The children in the study were divided and sent to one of three sites for initial developmental-diagnostic assessment or a second opinion. Prior to the study, a 2-day course was administered to educate participating clinicians; reliability was then established for each clinician collecting data. The children were similarly reassessed 3 months later, using the CARS.

The results of the study showed excellent agreement (88%) between the CARS categorical diagnosis based on behavior and the clinical criteria of the *DSM-IV*.<sup>18</sup>

The Autism Observation Scale for Infants (AOSI) is one tool that has been developed to detect and monitor for early signs of autism in infants (ages 6 to 18 months) that have an older sibling with autism.<sup>16</sup> Through direct observation, the AOSI utilizes 18 items to detect and monitor putative signs of autism in infants. Using the AOSI, the examiner engages the infant in play and monitors the infant's responses to a set of semistructured activities that involve eye contact, behavioral reactivity, soothability, social interest, shared effect, and motor control. A test-retest ongoing prospective study was conducted on 32 infants at ages 6 and 12 months age to test the reliability of the AOSI. The reliability for the total scores was fair to good at 0.68 and 0.61, respectively, thus confirming the ability of the scale to detect markers for autism in infants.<sup>16</sup>

Additional studies have shown the significance of clinical judgment by an experienced clinician to be the "gold standard" for the diagnosis of autism.<sup>19</sup> Clinical judgment with the use of the *DSM-IV* criteria and advanced assessment knowledge of autism can solidify an early (under age 2) identification of autism at well-child visits.<sup>12</sup> In a study of 77 children who received diagnostic evaluation between the ages of 16 and 35 months, and then again between 42 and 82 months, clinical judgment proved to be most reliable and accurate over time as compared to the other diagnostic tools.<sup>19</sup> Eighty percent of the children in this study remained within the autism diagnostic category when retested, thus validating the accuracy of the early diagnosis of autism by an experienced clinician.

The Kleinman study in 2008 further compared additional autistic screening tools, such as the Autism Diagnostic Interview-Revised (ADI-R) tool, the Vineland Adaptive Behavior Scale (VABS), the Mullen Scales for Early Learning, the Bayley Scales of Infant Development, and the Autism Diagnostic Observation Schedule (ADOS) for validity and stability in the early diagnosis of autism.<sup>19</sup> The ADI-R tool is a semistructured, clinician-based interview tool intended for caregivers to evaluate a child's communication, social development, play, and restricted, repetitive, and stereotyped behaviors. The ADI-R tool shows a diagnostic stability of 67% and is considered a valid instrument to use in the early diagnosis of autism.<sup>19</sup> The ADOS, which is also based on direct child observation, has been deemed valid with a diagnostic stability of 83%.<sup>19</sup>

### ■ Treatments

Children with autism have challenging behaviors that cause difficulty interacting both communicatively and socially.

The majority of treatment programs focus on behavior modification, but other approaches include dietary changes, parental counseling, speech therapy, special education in schools, medications, and complementary therapies such as acupuncture. However, no one approach is usually successful and many treatments are often attempted in the management of this disorder.

### Behavior modification

Early intervention programs that are individually tailored to a child's strengths and weaknesses can decrease the intensity of aberrant behaviors and enable a child to develop appropriate communicative and social skills as well as handle activities of daily living. There are two behavior modification programs that are used to successfully treat autism. One such behavioral program, applied behavior analysis, is centered on life and play skills as well as academic facets such as vocabulary, reading, and mathematics.<sup>20</sup> This is a highly structured program in which a teacher or therapist works one-on-one to assist the child in mastering particular skills before moving on to the next level. For example, a child must first master the ability to sit on a chair and then once that is mastered, the child must sit on a chair next to a table.<sup>20</sup>

A second program, Lovaas, is an intensive (approximately 40 hours a week of therapy), home-based behavioral program that claims to decrease the severity of autism in 42% of the cases.<sup>20</sup> Research indicates this type of approach needs to be thoroughly evaluated by parents prior to initiating due to the intensity and duration of the treatment. In the United Kingdom, a unique study was conducted involving 141 members of a group called Parents for Early Intervention of Autism in Children (PEACH). The parent(s) integrated the Lovaas style behavioral program in each of their homes and 26.2% noted improvement in their child's behavior as well as described them as being compliant and happy. Barriers that were identified included lack of physical resources in the community; personal, financial requirements; and lack of cooperation in the schools.<sup>21</sup>

### Dietary modifications

Dietary adjustments have been found to be beneficial to some children with autism. One adjustment requires eliminating gluten (wheat) and casein (milk). Case reports by parents and teachers throughout the United States claim that children have been "cured" of their autism, and marked improvements have been seen in language and social skills after removing casein and gluten from their diet. However, there is insufficient empirical data that support these claims.<sup>22</sup>

A randomized, double-blind pilot study was conducted with 15 autistic children ages 2 to 16 to test the effect of a

casein- and gluten-free diet provided by the University of Florida's metabolic kitchen for a 12-week period.<sup>22</sup> Data were collected by qualified individuals, using validated, reliable instruments (for example, the CARS criteria) immediately before the start of the diet, at 6 weeks, and again at 12 weeks. Results indicated no statistical significance, possibly due to the small sample size and large variances in observed behavior within individual groups. There was also no change in the child's baseline after being reevaluated using the CARS criteria. However, parents of seven children involved in the study reported improvements in language and decreased hyperactivity and tantrums.<sup>22</sup>

Another pilot study was conducted with 30 autistic children between the ages 4 and 10 to test the effects of a ketogenic diet on autistic behavior.<sup>23</sup> The ketogenic diet, which distributes energy intake as follows: 30% of energy as medium-chain triglyceride oil, 30% as fresh cream, 11% as saturated fat, 19% as carbohydrates, and 10% as protein, was conducted for 6 months with intervals of 4 weeks on the diet and 2 weeks off.<sup>23</sup> Results demonstrated that 7 children in the study got sick and could not tolerate the diet, 5 children only adhered to it for 2 months, whereas 18 or 60% completed the 6-month duration. After reevaluating the children with the CARS (standard error 0.89; highly significant), improvements were recorded in social behavior, learning, hyperactivity, speech, and cooperation. Furthermore, the children who had the most significant improvements were the ones diagnosed with mild autism and not the more severe forms.<sup>23</sup>

### Complementary therapy

A type of acupuncture, termed scalp acupuncture, is the only complementary therapy shown to be effective in treating language delays associated with mild autism. Scalp acupuncture is specialized and involves needle placement at specific body locations such as the ear, nose, hand, and foot.<sup>24</sup> A 9-month, test-retest study applying scalp acupuncture was conducted with 20 autistic children between the ages of 4 and 7 who had delayed language development. The children were divided into two groups: one group received scalp acupuncture and language therapy, while the second group received only language therapy. The use of scalp acupuncture proved to be highly significant ( $P < 0.001$ ) and the children who received the acupuncture along with language therapy showed significant improvements in cognitive and expressive language skills.<sup>24</sup>

### Pharmacologic approaches

There are no medications specifically designed to treat autism. Instead, pharmacologic interventions include medications that target the symptoms of autism such as repetitive thoughts and behaviors, depression, and anxiety. Medica-

### Counseling

NPs can help increase the overall function in children with autism. Treatment goals may include decreasing maladaptive and repetitive behaviors and helping the family manage stress associated with raising a child with autism. The child's entire family should be involved in the treatment plans. According to the AAP, primary care providers must provide honest and truthful explanations about the disorder to the siblings of a child diagnosed with autism to help them understand why they do not receive as much attention at home. NPs can also help families develop coping strategies such as devising a schedule, communicating effectively, introducing change slowly, planning physical activities, and working with the child's strengths and weaknesses. Unfortunately, many NPs do not have time to effectively treat the whole family, and in these cases, a referral to family counseling has shown to be an effective complement.<sup>12,28</sup>

tions commonly prescribed by healthcare providers for these symptoms are selective serotonin reuptake inhibitors.<sup>25</sup> Fluoxetine has been approved by the FDA for both depression and obsessive compulsive disorder (OCD) in children age 7 and older.<sup>31</sup> Three agents have been approved for only OCD and these include fluvoxamine for children age 8 and older; sertraline for children age 6 and older; and the tricyclic, clomipramine, for children age 10 and older.<sup>31</sup> Yet, despite their effectiveness, some studies have shown that treatment with these medications can have unintentional side effects in adolescents and young adults.

In 2004, the FDA adopted a black box warning on all antidepressant medications in order to alert the public about the increased risk for suicidal thinking and attempts in children and adolescents. A black box is used to alert the public about the potentially harmful side effects such as worsening depression, suicidal thinking or behavior, and unusual changes in behavior like sleeplessness, agitation, or withdrawal from normal social situations.<sup>31</sup>

An atypical antipsychotic, risperidone (Risperdal), has been involved in several clinical trials to also treat the symptoms of autism. One trial indicated that risperidone may be effective in decreasing hyperactivity, obsessive preoccupations, aggressiveness, and impulsivity.<sup>26</sup> The FDA has recently approved the use of risperidone for the treatment of irritability in autistic children between the ages of 5 and 17 but it is only licensed to be used in children displaying serious behavioral problems such as tantrums, aggression, and self-injury, not for the core symptoms of autism.<sup>26</sup> Other pharmacologic treatments may include antipsychotics such as haloperidol to treat serious aggressive behaviors, stimulant medications such as methylphenidate for attention deficit hyperactivity disorder, as well as some benzodiazepines



such as diazepam and lorazepam. However, further studies are needed to determine the long-term side effects, safety, and efficacy of these medications in children with autism.<sup>25</sup>

### ■ Advanced practice implications

The NP's role includes early diagnosis of autism, offering anticipatory guidance and family or parental counseling, preparation for medical procedures, and continuity of care.

#### Early diagnosis

Literature has shown that one of the most important roles of the NP is early diagnosis of autism because it improves the outcome for children.<sup>12</sup> According to the AAP, the NP's ability to note behaviors typical of autism at a 1-year well-child check and a focus on key elements of history, such as parental reports, are important tools in providing proper early diagnosis.<sup>27</sup> It is imperative for the NP to begin to screen children in infancy and at later well-child visits, and provide additional assessment if autism is suspected. The NP can further address parental concerns regarding their child's development, follow up with developmental surveillance and screening, and provide early management strategies such as parental education and support, community services, behavior management, and medical treatments at well-child checks.<sup>12</sup> Finally, the NP can provide referrals for further evaluation and treatment.

#### Anticipatory guidance


NPs and other primary care providers are unable to determine a child's outcome at the time of autism diagnosis; however, they can provide anticipatory guidance in terms of possible trajectory and the child's potential ability for development.<sup>28</sup> Parents can use this information as a guideline for assessing their child's progress over time. NPs should gear anticipatory guidance toward both parents, or guardians, and patients. NPs can teach parents how to manage problematic behaviors and to encourage positive social skills in their child. The NP can further provide parents with local resources such as local mental health agencies, counseling centers, and credible Internet sites. The NP can help older children and adolescents with their behavioral weaknesses and strengths so they may advance socially.<sup>29</sup> The NP's ability to refer a child to counseling and anticipate how a child or adolescent with autism will be treated in society, especially in the classroom, can help the child cope with feelings of loneliness and low self-esteem often caused by ridicule from classmates.<sup>29</sup> (See *Counseling*.)

#### Preparation for procedures and continuity of care

The AAP has provided recommendations for primary NPs who treat children with autism. These include monitoring

all areas of development at each well-child visit, genetic counseling for families, referrals to outside therapies, and providing comprehensive care and age-appropriate interventions to the child.<sup>27</sup> Research indicates the importance of continuity of care for children with autism; therefore, NPs should avoid rapid caregiver changes and opt for more gradual methods of transferring care, such as introducing new people, places, and medical procedures over time.<sup>12</sup> A gradual method will help decrease a child's anxiety and is less disruptive to the daily routine.

### ■ Multidisciplinary teams a must

A new case of autism is diagnosed almost every 20 minutes; 67 children are diagnosed every day.<sup>5</sup> Autism can cause profound behavioral, communicative, and psychomotor deficits. Although symptoms are often subtle in infants, as children begins to interact socially with others, the signs of autism become increasingly apparent. There is no definitive treatment for autism and research is ongoing in hopes of finding a cure. Early diagnosis of autism is the key to initiating an intervention program that will enhance outcomes. Diagnosis of autism requires a comprehensive evaluation by a multidisciplinary team. Primary care NPs should be able to recognize the clinical signs of autism, analyze the disorder using diagnostic tools, and provide management and referral options for families with autistic children. The NP must also stay current in autistic research for the purpose of gaining knowledge about current treatments as well as to remain informed about changes in causes and diagnostic criteria for autism. Awareness of the existing research will help APNs guide families toward effective interventions. 

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