

# Autism Screening and Diagnostic Evaluation: CAN Consensus Statement

By the Cure Autism Now Foundation  
(CAN) Consensus Group\*

## EDUCATIONAL OBJECTIVES

- Discuss the usefulness of a simple screening tool for autism and pervasive developmental disorders.
- Identify appropriate neurodevelopmental evaluations in children with language delay or other indicia of autism.
- Discuss appropriate laboratory studies for children with suspected autism.

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## ABSTRACT

*Autism and related disorders occur in as many as one in 500 children. Recognizing the need for uniform recommendations to help guide the initial diagnostic evaluation and referrals in this area, an initial working group of the Cure Autism Now Foundation promulgated the following consensus guidelines for autism screening and diagnostic referral. Although these recommendations were formulated using the best information currently available, they are likely to be modified as the results of additional studies become available; however, they provide a useful tool for clinicians involved in the management of children with autism and related developmental abnormalities.*

## INTRODUCTION

Autism is a neuropsychiatric syndrome characterized by onset prior to age 3 of severe abnormalities of reciprocal social relatedness; communication deficits (including deficits in language); and restricted, stereotyped patterns of interest and behaviors.<sup>1-3</sup> Although autism has a wide spectrum of clinical presentations, it is distinct from other childhood neuropsychiatric disorders and primary forms of mental retardation.<sup>3</sup> Autism presents a clinical challenge. Children with autism and related disorders, pervasive developmental disorders (PDDs), are often not diagnosed until several years after initially presenting for medical or behavioral evaluation. This is due in part to the lack of uniform guidelines for surveillance, diagnostic evaluation, and follow-up of children with autism and related disorders.

Early diagnosis, however, may be essential to successful intervention in children with autism or other developmental language delay. Advances in developmental neurobiology and psychology suggest that the first few years of life represent a critical period for the development of many perceptual and higher-order

processing systems in the human brain.<sup>4-8</sup> Recent evidence also indicates that early identification and subsequent treatment may portend a better prognosis.<sup>9-12</sup> Recognizing the need for early diagnosis and treatment, Cure Autism Now (CAN) convened this initial working group to produce the following consensus outline for screening, workup, and referral.

Because few controlled trials have been conducted, in many areas this consensus is based on clinical experience and judgment. Therefore, these recommendations are likely to be modified as new data becomes available. Nevertheless, they represent the consensus of a group of neurologists, pediatricians, psychologists, and psychiatrists who are experienced in the diagnosis, evaluation, and treatment of children with autism, using the best information currently available.

This consensus statement is intended as a brief outline and not a comprehensive guide. Detailed practice guidelines for children with PDD are forthcoming from the American Academy of Child and Adolescent Psychiatry (personal communication, F Volkmar and D. Cohen, January, 1998). General detailed guidelines for the workup of infants and toddlers with psychiatric disorders are available from that same organization.<sup>13</sup>

## INITIAL SCREENING

Autism and related disorders occur in between one and 500 and one in 1,000 children.<sup>12,14-18</sup> Many pediatricians are uncertain how to proceed with the workup of the 1 to 2-year-old child who looks normal, but in whom the parents complain of delayed language or social behavior relative to other children. Parents may even report subtle regression. Because timely identification may lead to early intervention and better outcomes,<sup>9-12</sup> a standard screening device that can help distinguish between potentially autistic and nonautistic behaviors, and identify most children at risk, would prove beneficial.

The Checklist for Autism in Toddlers (CHAT) successfully identifies autism or PDD in may

children.<sup>19-20</sup> In Britain, this 3-minute screening tool has been shown to predict 90% of children who will develop autism, PDD, Asperger's syndrome, or other developmental delay syndromes.<sup>20</sup> Therefore, until a better screening tool is developed, the consensus panel recommends that all children (especially those with symptoms or parental concerns suggestive of PDD) be screened by their pediatricians at 18 months of age using the CHAT.

A normal CHAT, however, does not rule out a developmental disorder. Suspicion by the parent or clinician of poorly developed verbal or nonverbal communication skills should always trigger appropriate referral for formal speech, language, and developmental evaluation. Abnormal performance on the CHAT requires further assessment for possible PDD, but may be found in many children who develop normally or who do not have autism or another PDD.

#### **NEURODEVELOPMENTAL REFERRAL**

Abnormal CHAT or language delay in a child of any age should trigger referral to an appropriate specialist for neurodevelopmental and hearing evaluation. In addition, language or other developmental regression should trigger a prompt and comprehensive neurologic evaluation.

Neurodevelopmental specialists may be certified in a variety of disciplines, including child psychiatry, pediatric neurology, developmental pediatrics, neuropsychology, or child psychology. Any such specialist, however, must be experienced in the evaluation and therapy of children with language delay. The initial specialist evaluation should include, but need not be limited to: **1)** neurologic examination with detailed medical exam, mental status evaluation, and thorough family history; **2)** speech and language evaluation; and **3)** developmental history and direct evaluation using a reliable diagnostic instrument to distinguish children with autism or another PDD from children with other developmental difficulties – for example, the Autism Diagnostic Interview or the Autism Diagnostic Observation Schedule. Evaluation of the child's social and emotional developmental levels by a professional who is trained in assessment in the presence of communication impairment and behavioral problems common in autism is also recommended.

#### **AUDIOLOGICAL EVALUATION**

The hearing evaluation should be behavioral in focus and include pure formal tone audiometry performed by an experienced pediatric audiologist. Brainstem auditory evoked potentials are necessary only if the initial test is equivocal, suboptimal, or suggests central nervous system abnormality.

#### **LABORATORY ANALYSES**

If autism is suspected following these initial evaluations and the etiology remains unclear, the following lab studies are recommended to be carried out under the supervision of the specialist.

#### **Electroencephalography**

A significant number of children with autism and related disorders may have abnormal electroencephalograms (EEGs), and many have epileptiform activity and/or epilepsy.<sup>12,21-24</sup> An extended EEG including all four stages of sleep is recommended, especially in children with regression, those with poor phonology, or those who are nonverbal. Whether the recording is conducted for 4 hours, overnight, or for a complete 24-hour period is not currently specified, since the optimal time length is not known. But, some children will require overnight or 24-hour EEGs to obtain all stages of sleep.

For children in whom clinical or subclinical seizure activity is suspected, one or more 24-hour EEGs are recommended, since longer recordings are more likely to detect abnormalities that may be clinically relevant. In children who exhibit good phonology or who lack other clinical indicators of seizure activity, recommendations for the level of clinical aggressiveness, remain to be empirically determined.

#### **Metabolic Screening**

Metabolic screening should be conducted if believed appropriate in the clinical judgment of the specialist. Metabolic tests should be considered specifically in patients with PDD, atypical findings on physical examination, or hypotonia. The following may be considered in the metabolic assessment: 1) quantitative amino acids; 2) urine organic acids; 3) uric acid (24-hour urine); 4) thyroid studies; 5) lactate, pyruvate and carnitine; and 6) lead levels. Unfortunately, the utility of these tests in large populations of infants with suspected autism or language delay is unknown.

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## **Neuroimaging**

Although several studies have demonstrated the possibility of subtle neuroanatomic anomalies associated with autism, no protocols to date have yielded findings of sufficient magnitude to provide diagnostic specificity on a case-by-case basis. In the clinical experience of this consensus group, treatable conditions masquerading as autism are unusual.<sup>13,18</sup> Therefore, the use of neuroanatomic imaging studies such as magnetic resonance imaging in the primary diagnostic workup of children with autism is likely to benefit only those for whom neurologic examination, EEG, or other clinical indicators suggest a focal lesion. For example, 3% to 9% of children with the triad of epilepsy, retardation, and autism may have tuberous sclerosis, which can otherwise remain undiagnosed at early stages.<sup>2,25</sup> In these children, and in any with features suggestive of focal brain lesions, imaging is indicated.

Although valuable research tools, functional brain imaging techniques such as single photon emission tomography, positron emission tomography, magnetoencephalography, or magnetic resonance spectroscopy are of uncertain utility in the diagnosis or treatment of suspected autism. These modalities may be useful in specific circumstances, such as the localization of seizure foci.

## **Karyotype and DNA Testing for Fragile X**

Population-based studies suggest that between 5% and 12% of children with autism have underlying medical or genetic conditions.<sup>25</sup> Both karyotype and fragile X chromosome testing should therefore be considered,<sup>14,26</sup> depending on the degree of clinical suspicion. These tests are especially recommended for genetic counseling purposes in families who are planning to have additional children. Karyotyping should be at high resolution with particular attention paid to possible duplications of 15q11-13.<sup>27</sup>

## **Other Laboratory Tests**

Children with autism may have an increased incidence of certain infections, such as otitis media.<sup>28,29</sup> Altered immune parameters have also been demonstrated in some cases.<sup>30,31</sup> In addition, a higher incidence of allergies has been reported in this population,<sup>30,31</sup> and one study has suggested an increase in gastrointesti-

nal disturbances.<sup>32</sup> Given the poor communication skills of children with autism, the treating physician should maintain a high index of suspicion for infection or other medical problems, particularly during periods of behavioral regression or exacerbation.

## **LONGITUDINAL FOLLOW-UP**

Longitudinal follow-up may be conducted by a neurodevelopmentalist in a specialty clinic for the purposes of evaluating and modifying therapies. Many children with autism exhibit some combination of abnormal sleep patterns, mood disorders, aggression, self-abuse, compulsive behaviors, or abnormal attention spans.<sup>2,12,25</sup> Psychiatric evaluation can therefore be a critical element in the diagnosis and longitudinal follow-up of children with autism and related disorders.

Psychological, developmental testing and EEG monitoring (in patients with seizures or epileptiform activity) should be repeated as clinically indicated, and may be useful tools in the monitoring of patient response to therapy. However, the role of EEG and the significance of epileptiform and nonepileptiform abnormalities in patients with suspected or diagnosed autism are currently uncertain. There is a need for controlled studies to address these issues.

## **CONCLUSIONS**

Autism is as common as many other childhood medical disorders such as diabetes and leukemia. However, detection of the autistic child is often delayed significantly beyond the time of first suspicion by parents. The CHAT is a rapid and convenient tool that should be used to screen all 18-month-old children for autistic spectrum disorders. Children with a positive CHAT should be referred to a neurodevelopmental specialist with expertise in autism for further workup and longitudinal evaluation.

A normal CHAT, however, does not rule out a developmental disorder, and suspicion of poorly developed verbal or nonverbal communication skills by parents or the clinician should always trigger referral for formal speech, language, and developmental evaluation. Longitudinal studies ascertaining the efficacy of the CHAT in diverse clinical settings are needed, as are investigations of the utility of other screening instruments for children with language, social, and adaptive behavioral delays. Detailed neurologic, medical, psychological, speech, and language evaluations should be made a part of every autism workup.

This consensus statement highlights the large

gaps in our current knowledge regarding the appropriate evaluation of children with autism and related disorders (PDD). At this point, management depends largely upon the expert judgment of clinicians experienced in autism, language delay, and pediatric neurology and psychiatry, rather than data gathered from controlled clinical trials. It is the hope of this consensus group that this deficiency in trial data will soon be rectified.

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