



SCREENING AND DIAGNOSIS OF AUTISM

This is a summary of the American Academy of Neurology (AAN) and Child Neurology Society (CNS) guideline on screening and diagnosis for autism. This practice parameter reviews the available empirical evidence and gives specific recommendations for the identification of children with autism. This approach requires a dual process: 1) routine developmental surveillance and screening specifically for autism to be performed on all children to first identify those at risk for any type of atypical development, and to identify those specifically at risk for autism; and 2) to diagnose and evaluate autism, to differentiate autism from other developmental disorders.

LEVEL ONE: Routine developmental surveillance screening specifically for autism

Good Evidence Supports

1. Developmental surveillance should be performed at all well-child visits from infancy through school-age, and at any age thereafter if concerns are raised about social acceptance, learning, or behavior (Level* B).
2. Recommended developmental screening tools include the Ages and Stages Questionnaire, the BRIGANCE® Screens, the Child Development Inventories, and the Parents' Evaluations of Developmental Status (Level B).
3. Because of the lack of sensitivity and specificity, the Denver-II (DDST-II) and the Revised Denver Pre-Screening Developmental Questionnaire (R-DPDQ) are not recommended for appropriate primary-care developmental surveillance (Level B).
4. Further developmental evaluation is required whenever a child fails to meet any of the following milestones (Level B): babbling by 12 months; gesturing (e.g., pointing, waving bye-bye) by 12 months; single words by 16 months; two-word spontaneous (not just echolalic) phrases by 24 months; loss of any language or social skills at any age.
5. Siblings of children with autism should be carefully monitored for acquisition of social, communication, and play skills, and the occurrence of maladaptive behaviors. Screening should be performed not only for autism-related symptoms but also for language delays, learning difficulties, social problems, and anxiety or depressive symptoms (Level B).
6. Screening specifically for autism should be performed on all children failing routine developmental surveillance procedures using one of the validated instruments—the CHAT or the Autism Screening Questionnaire (Level B).
7. Laboratory investigations recommended for any child with developmental delay and/or autism include audiologic assessment and lead screening (Level B). Early referral for a formal audiologic assessment should include behavioral audiometric measures, assessment of middle ear function, and electrophysiologic procedures using experienced pediatric audiologists with current audiologic testing methods and technologies (Level B). Lead screening should be performed in any child with developmental delay and pica. Additional periodic screening should be considered if the pica persists (Level B).

LEVEL TWO: Diagnosis and evaluation of autism

Strong Evidence Supports

1. Genetic testing in children with autism, specifically high resolution chromosome studies (karyotype) and DNA analysis for FraX, should be performed in the presence of mental retardation (or if mental retardation cannot be excluded), if there is a family history of FraX or undiagnosed mental retardation, or if dysmorphic features are present (Level A). However, there is little likelihood of positive karyotype or FraX testing in the presence of high-functioning autism.
2. Selective metabolic testing (Level A) should be initiated by the presence of suggestive clinical and physical findings such as the following: if lethargy, cyclic vomiting, or early seizures are evident; the presence of dysmorphic or coarse features; evidence of mental retardation or if mental retardation cannot be ruled out; or if occurrence or adequacy of newborn screening at birth is questionable.

Good Evidence Supports

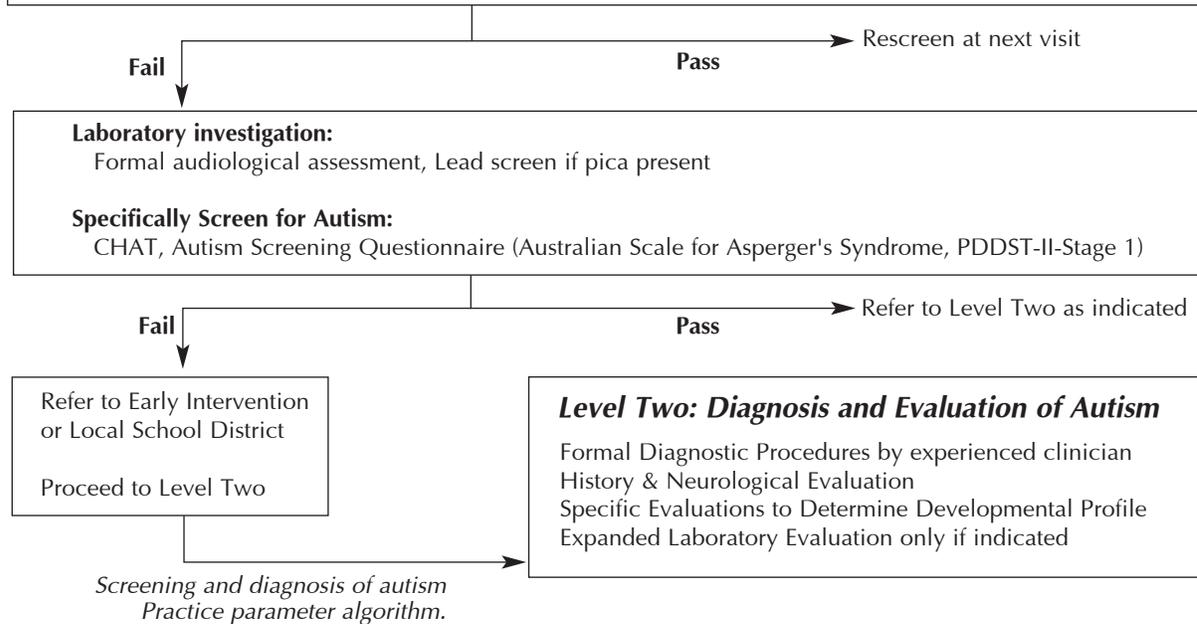
1. There is inadequate evidence at the present time to recommend an EEG study in all individuals with autism. Indications for an adequate sleep-deprived EEG with appropriate sampling of slow wave sleep include (Level B) clinical seizures or suspicion of subclinical seizures, and a history of regression (clinically significant loss of social and communicative function) at any age, but especially in toddlers and preschoolers.
2. Recording of event-related potentials and magnetoencephalography are research tools at the present time, without evidence of routine clinical utility (Level B).
3. There is no clinical evidence to support the role of routine clinical neuroimaging in the diagnostic evaluation of autism, even in the presence of megalencephaly (Level B).
4. There is inadequate supporting evidence for hair analysis, celiac antibodies, allergy testing (particularly food allergies for gluten, casein, candida, and other molds), immunologic or neurochemical abnormalities, micronutrients such as vitamin levels, intestinal permeability studies, stool analysis, urinary peptides, mitochondrial disorders (including lactate and pyruvate), thyroid function tests, or erythrocyte glutathione peroxidase studies (Level B).

LEVEL ONE: Routine Developmental Surveillance

by all providers at every well-child visit e.g., PEDS, ASQ, CDIs, or BRIGANCE®

Absolute Indications for Immediate Evaluation:

No babbling, or pointing or other gesture by 12 months
No single words by 16 months
No 2-word spontaneous (not echolalic) phrases by 24 months
ANY loss of ANY language or social skills at ANY age



Visit www.aan.com/professionals/practice/index.cfm to view the entire guideline and additional AAN child neurology guidelines.

This guideline summary is evidence-based. The AAN uses the following definitions for the level of recommendation and classification of evidence. *Definitions for strength of the recommendations: **Level A:** Established as effective, ineffective or harmful, (or established as useful/predictive or not useful/predictive) for the given condition in the specified population. **Level B:** Probably effective, ineffective or harmful (or probably useful predictive or not useful/predictive) for the given condition in the specified population. **Level C:** Possibly effective, ineffective or harmful (or possibly useful predictive or not useful/predictive) for the given condition in the specified population. **Level U:** Data inadequate or conflicting. Given current knowledge, treatment (test, predictor) is unproven.

This is an educational service of the American Academy of Neurology. It is designed to provide members with evidence-based guideline recommendations to assist with decision-making in patient care. It is based on an assessment of current scientific and clinical information, and is not intended to exclude any reasonable alternative methodologies. The AAN recognizes that specific patient care decisions are the prerogative of the patient and the physician caring for the patient, based on the circumstances involved. Physicians are encouraged to carefully review the full AAN guidelines so they understand all recommendations associated with care of these patients.

Copies of this summary and a companion patient version are available at www.aan.com/professionals/practice/index.cfm or through AAN Member Services at (800) 879-1960.



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