



EVALUATION OF THE CHILD WITH GLOBAL DEVELOPMENTAL DELAY

This is a summary of the American Academy of Neurology (AAN) and Child Neurology Society (CNS) guideline on diagnosis of the child with global developmental delay. Global developmental delay is a subset of developmental disabilities defined as significant delay in two or more of the following developmental domains: gross/fine motor, speech/language, cognition, social/personal, and activities of daily living. Those deficits are evident in comparison with the skills attainment of chronological peers. Significant delay is defined as performance two standard deviations or more below the mean on age-appropriate, standardized norm-referenced testing.

This practice parameter reviewed available evidence concerning the value of diagnostic testing in the initial evaluation of a young child with a global developmental delay that is static, nonprogressive, and has no clear etiology. Based on this evidence, below are specific recommendations for each testing modality.

EVIDENCE FOR EVALUATIONS OF THE CHILD WITH GLOBAL DEVELOPMENTAL DELAY

Metabolic screening

(serum amino acids; urine organic acids, acylcarnitines, mucopolysaccharides; serum glucose, bicarbonate, lactate, pyruvate, ammonia, creatine kinase)

- Routine metabolic screening for inborn errors of metabolism is not indicated in the initial evaluation of a child with global developmental delay, provided universal newborn screening was performed and the results are available for review.
- If newborn screening was not performed, if it is uncertain whether a patient had testing, or if the results are unavailable, metabolic screening should be obtained in a child with global developmental delay.
- Metabolic testing may be pursued in the context of historical or physical examination findings that are suggestive of a specific etiology (Level B*; Class II and III** evidence).

Cytogenetic testing

- Routine cytogenetic testing is indicated in the evaluation of the child with developmental delay even in the absence of dysmorphic features or clinical features suggestive of a specific syndrome (Level B; Class II and III evidence).

Fragile X Testing

- Testing for the Fragile X mutation, particularly in the presence of a family history of developmental delay, may be considered in the evaluation of the child with global developmental delay. Clinical preselection may narrow the focus of who should be tested without sacrificing diagnostic yield.
- Although screening for Fragile X is more commonly done in males because of the higher incidence and greater severity, females are frequently affected and may also be considered for testing (Level B; Class II and Class III evidence).

Rett syndrome testing

- The diagnosis of Rett syndrome should be considered in females with unexplained moderate to severe mental retardation. If clinically indicated, testing for the *MECP2* gene deletion may be obtained. Insufficient evidence exists to recommend testing of females with milder clinical phenotypes or males with moderate or severe developmental delay (Level B; Class II and Class III evidence).

Molecular techniques

- In children with unexplained moderate or severe developmental delay, additional testing using newer molecular techniques (e.g. FISH, microsatellite markers) to assess for subtelomeric chromosomal rearrangements may be considered (Level B; Class II and Class III evidence).

Lead screening

- Screening of children with developmental delay for lead toxicity may be targeted to those with known identifiable risk factors for excessive environmental lead exposure as per established current guidelines (Level B; Class II evidence).

Thyroid screening

- In the setting of existing newborn screening programs for congenital hypothyroidism, screening of children with developmental delay with thyroid function studies is not indicated unless there are systemic features suggestive of thyroid dysfunction (Level B; Class II evidence).

EEG

- An EEG can be obtained when a child with global developmental delay has a history or examination features suggesting the presence of epilepsy or a specific epileptic syndrome (Level C; Class III and IV evidence).
- Data are insufficient to permit making a recommendation regarding the role of EEG in a child with global developmental delay in whom there is no clinical evidence of epilepsy (Level U; Class III and IV evidence).

Neuroimaging

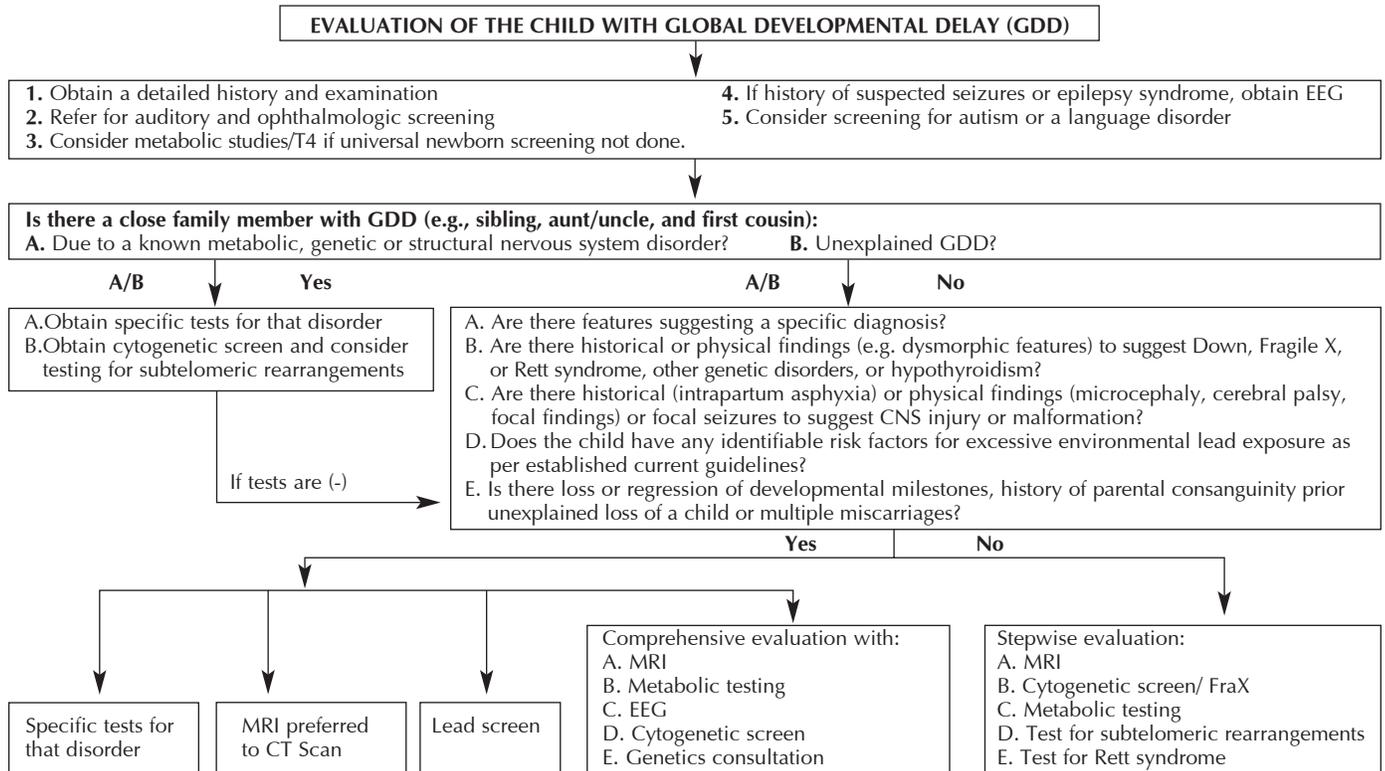
- Neuroimaging is recommended as part of the diagnostic evaluation of the child with global developmental delay (Level B; Class III evidence).
- As the presence of physical findings (e.g., microcephaly, focal motor findings) increases the yield of making a specific neuroimaging diagnosis, physicians can more readily consider obtaining a scan in this population (Level C; Class III evidence).
- If available, MRI should be obtained in preference to CT scanning when a clinical decision has been made that neuroimaging is indicated (Level C; Class III evidence).

Hearing and vision testing

- Children with global developmental delay may undergo appropriate vision and audiometric assessment at the time of their diagnosis (Level C; Class III evidence).
- Vision assessment can include vision screening and a full ophthalmologic examination (visual acuity, extra-ocular movements, fundoscopic) (Level C; Class III evidence).

- Audiometric assessment can include behavioral audiometry or brainstem auditory evoked response testing when feasible (Level C; Class III evidence).
- Early evidence from screening studies suggest that transient evoked otoacoustic emissions should offer an alternative when audiometry is not feasible (Level A; Class I & II evidence).

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This guideline summary is evidence-based. The AAN uses the following definitions for the level of recommendation and classification of evidence. ***Recommendation Level:** "Level" refers to the strength of the practice recommendation based on the reviewed literature. **Level A:** Established as effective, ineffective or harmful or as useful/predictive or not useful/predictive; **Level B:** Probably effective, ineffective or harmful or as useful/predictive or not useful/predictive; **Level C:** Possibly effective, ineffective or harmful or as useful/predictive or not useful/predictive; **Level U:** Data inadequate or conflicting; treatment, test or predictor unproven. ****Class of Evidence:** "Class" refers to the quality of research methods employed in the reviewed literature **Class I:** A statistical, population-based sample of patients studied at a uniform point in time (usually early) during the course of the condition. All patients undergo the intervention of interest. The outcome, if not objective, is determined in an evaluation that is masked to the patients' clinical presentations; **Class II:** A statistical, non-referral-clinic-based sample of patients studied at a uniform point in time (usually early) during the course of the condition. Most (>80%) patients undergo the intervention of interest. The outcome, if not objective, is determined in an evaluation that is masked to the patients' clinical presentations; **Class III:** A selected, referral-clinic-based sample of patients studied during the course of the condition. Some patients undergo the intervention of interest. The outcome, if not objective, is determined in an evaluation by someone other than the treating physician; **Class IV:** Expert opinion, case reports, or any study not meeting criteria for Class I to III.

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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