Screening of Developmental Delay and Autism Spectrum Disorders

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Key Points

- The national prevalence of developmental disabilities is increasing. Early recognition of and intervention for ASD and/or other developmental problems is paramount in order to maximize the developmental progress.
- Autism spectrum disorder (ASD) is defined by impairments in social communication and social interaction skills, restricted or repetitive patterns of behavior, interests, and activities as well as sensory processing differences.
- The American Academy of Pediatrics (AAP) recommends that primary care physicians screen for ASD routinely at 18 months and 24 months of age in addition to regular developmental screening.

Developmental Delay (DD)

Definition and Prevalence

- Developmental delay is any condition in which a child is not achieving developmental milestones in the anticipated time frame. Developmental disabilities are a broad group of conditions associated with early and lasting impairments in physical, linguistic, intellectual, and/or behavioral skills.[1]
- Nationally, the prevalence of developmental delays and disabilities in children is increasing.

Approximately 1 in 6 children in the United States are reported as having a developmental disability based on the National Health Interview Survey data 2006-2008.[1]

Screening and Surveillance

- The AAP guidelines classify "surveillance" as an ongoing process of identifying children who may be at risk for developmental disorders, and "screening" as the use of standardized tools to further define or determine that risk.
- Traditional developmental surveillance only identifies 30% of children with delays. Utilization of standardized developmental screening tools increases this to 80-90%,[2] and is associated with quicker referral and access to developmental services.[3]
- In 2006, the AAP published recommendations for screening for developmental disorders in general pediatric practice. These recommendations, which have since been incorporated into the Bright Futures Guidelines, are as follows:[2, 4]
    - Developmental surveillance should be completed at each visit by specifically asking the parents about any possible developmental concerns.
    - A standardized developmental screening tool should be administered to all children, regardless of risk status or parental concern, during routine health supervision visits at the following intervals: 9-12 months, 18 months, and 24-30 months.
    - Choice of the specific screening tool to use at these intervals is left to the discretion of individual clinicians. Commonly used tools include the Ages and Stages Questionnaire (ASQ) and the Parents' Evaluation of Developmental Status (PEDS).
- Several barriers to implementing developmental screening practices have been recognized, including sociocultural factors as well as logistical challenges. Research supports consideration of family culture, engaging allied health professionals and Family Navigators, and close communication between physicians and early intervention services can help to overcome these barriers and optimize screening completion rates.[5,6] Several resources exist to assist physicians with implementation and optimization of developmental screening practices.[4, 7, 8]

Diagnostic Evaluation

- In any child with developmental delays, an audiology evaluation and referral to Early Intervention (First Connections, ages 0-36 months) or Early Childhood Services (through the local educational cooperative, ages 3-5 years). Not all these children require assessment by a developmental pediatrician unless concerns are present for ASD or possible organic etiology to the delay.
- It is important to note that specific developmental delays (i.e., speech-language delays) can and should be addressed with early intervention services prior to, or in absence of, a more specific or overarching diagnoses. Developmental therapies should not be deferred during the diagnostic process.
- Children with developmental delays require close monitoring of their developmental trajectory to assess if they are "closing the gap" between their current developmental skills and those expected for age. If global delays persist, serial cognitive assessments may establish a clinical diagnosis of intellectual disability.
- The AAP recommends that all patients with intellectual disability (ID)/global developmental delay (GDD) undergo a comprehensive medical diagnostic evaluation to include, in addition to a complete history (including 3-generation family history) and physical exam, genetic evaluation.[9]
    - If a specific diagnosis is suspected from the clinical presentation, appropriate confirmatory studies should be undertaken.
    - If no specific diagnosis is strongly suspected, the AAP recommends chromosomal

microarray and fragile X genetic testing in all patients (regardless of gender) and as well as consideration of specific metabolic testing for treatable inborn errors of metabolism.
- A brain MRI should be considered in the context of macrocephaly, microcephaly or abnormal findings on neurological examination.
- If no diagnosis is established, additional work-up may be considered in conjunction with other specialists as directed by the patient centered medical home.

Autism Spectrum Disorder (ASD)

Definition and Prevalence

- The overall estimated prevalence of ASD is 16.8 per 1,000 children, or 1 out of every 59. Estimated prevalence is four times higher among boys than girls. Although there is not believed to be a true difference in prevalence based on ethnicity, non-Hispanic white children are more likely to be identified with ASD compared to African American or Hispanic children. [10]
  Current prevalence estimates are based on 2014 data from Autism and Developmental Disabilities Monitoring Network (ADDM), a CDC surveillance network of 11 sites nationally, including Arkansas.[10]
- According to the 5th edition of the Diagnostic and Statistical Manual (DSM-5), autism spectrum disorder (ASD) is defined by the following:[11]
  - **Persistent deficits in social communication and social interaction across multiple contexts**, as manifested by (all of) the following, currently or by history:
    - Deficits in social-emotional reciprocity
    - Deficits in nonverbal communicative behaviors used for social interaction
    - Deficits in developing, maintaining, and understanding relationships
  - **Restricted, repetitive patterns of behavior, interests, or activities**, as manifested by at least two of the following, currently or by history
    - Stereotyped or repetitive motor movements, use of objects, or speech
    - Insistence on sameness, inflexible adherence to routines, or ritualized patterns or verbal nonverbal behavior
    - Highly restricted, fixated interests that are abnormal in intensity or focus
  - **Hyper- or hypo-reactivity to sensory input or unusual interests in sensory aspects of the environment**
- The definition of autism was changed substantially in 2013 with the publication of the 5th edition of the Diagnostic and Statistical Manual (DSM-5). Key differences between the DSM-5 and it's predecessor, the DSM-IV, published in 1994, with regard to autism are as follows.[12]
  - Previously (per DSM-IV) there were three diagnoses (Asperger's Disorder or Syndrome, autistic disorder, and pervasive developmental disorder, not otherwise specified [PDD-NOS]) under the "umbrella" of the autism spectrum. Currently (per DSM-5), there is only one categorical diagnosis of **autism spectrum disorder (ASD)**. This diagnoses encompasses all individuals who meet the criteria above, regardless of their level of intellectual or linguistic functioning.
  - Currently (per DSM-5), when clinicians make a diagnosis of ASD, they are asked to additionally specify whether the diagnosis is associated with either language or cognitive impairment. The complete diagnosis with this specification might read, for example, "autism spectrum disorder with language impairment, without cognitive impairment."
  - Currently (per DSM-5), when clinicians make a diagnosis of ASD, they are asked to designate a level of required support for both the social-communication domain and the restricted interest/repetitive behaviors domain. Levels of support range from 1,

requiring a relatively modest support, to 3, requiring significant support.
- If an individual was diagnosed with any autism spectrum disorder included in DSM-IV (Asperger disorder, autistic disorder, or PDD-NOS), the DSM-V indicates that that individual should be given the diagnosis of autism spectrum disorder (ASD).
- Any autism diagnoses given after the publication of the current criteria (DSM-5, 2013) should be given as autism spectrum disorder (ASD) with the specifications and support levels as described above.

Screening and Surveillance

- In 2007, the AAP published recommendations specifically for screening and identifying Autism Spectrum Disorders (ASD) and reaffirmed those recommendations in September 2010.[13]
- Despite the AAP recommendations, universal screening for ASD has not been widely accepted among all community practices. A multidisciplinary, international panel of experts in the field of ASD convened a workgroup in 2010 to review evidence for these guidelines and address barriers to implementing them. Their review of the evidence supports the usefulness of ASD-specific screening at 18 and 24 months, noting that siblings of children with established diagnoses of autism deserve intensified surveillance due to their increased risk of ASD and other developmental disorders.[14]
- In 2016, The U.S. Preventive Services Task Force (USPSTF) published a position statement that insufficient data exist regarding harms and benefits of universal screening for autism spectrum disorder to recommend it at this time.[15]
- The AAP released a statement in opposition of the USPSTF position and reaffirmed it's recommendations for universal screening based on "strong evidence… on the benefit of formal screening… applied to all children – not only those who exhibit overt symptoms, or those an individual clinician judges would benefit." [16]
- Evidence supports the long-term stability of a diagnosis of ASD made at or after 24 months. Support for the long-term stability of diagnoses of ASD made earlier than 24 months is less conclusive, although some data exist to support reliable diagnoses of at least some children prior to 24 months of age.[14]
- Early intervention for children with ASD can lead to significant improvements in IQ, language, and symptom severity. The recent addition of several large randomized controlled trials focused on very young children (less than 3) have added support for early identification, diagnosis, and intervention.[14]

Current AAP Recommendations for ASD Surveillance

Current AAP recommendations for ASD surveillance are as follows:[13]

- The AAP recommends surveillance for ASD at each well child visit with formal evaluation or screening depending upon presence of risk factors and child age.
- Risk factors include sibling with ASD, parental concern, other caregiver concern (i.e., childcare providers), and medical provider concern.
- If the child has a single risk factor and is <18 months of age, an evaluation of communication skills is recommended as few ASD-specific screening tools have been tested in very young children.
- For children >18 months of age with a single risk factor, administering an ASD screening tool is recommended regardless of their exactage.
- For children with multiple risk factors, referral for formal developmental evaluation, audiology

evaluation, and early intervention services are recommended.

Current AAP Recommendations for ASD Screening

Current AAP recommendations for ASD screening are as follows:[13]

- All children, regardless of risk factors, clinical or family concern, should be screen for ASD at

18 and 24 months of life.
- All children with positive screens should be referred for a comprehensive ASD evaluation and, simultaneously, to early intervention services.

Available Screening Tools

- Available screening tools for ASD are divided into 2 levels. Although titles and terminology of some screening tools still contain DSM-IV nomenclature, the assessments remain clinically relevant even after transition to DSM-5.[14]

LEVEL 1 (Parent Questionnaires)

- Checklist for Autism in Toddlers (CHAT) – one of the first screening tools developed, this is no longer recommended for use due to it's low sensitivity (only 18% when 18-month cohorts are followed for 6 years).
- **Modified Checklist for Autism in Toddlers (M-CHAT)** – currently the most widely used and recommended, recent review found strong evidence for use in children 16-30 months of age. When combined with a follow-up interview, positive predictive value (PPV) is estimated at 0.57. When screen-positive cases are evaluated for other significant developmental delays, the PPV of an M-CHAT plus a follow-up interview has been demonstrated as >0.90 across multiple studies. The MCHAT is available free online. It can be completed by the parent or administered in an interview.
- Modified Checklist for Autism in Toddlers – Revised with Follow-Up (M-CHAT-R/F) – recent revision to M-CHAT with simplified wording and omission of several items found to be less reliable. Early data are promising with PPV = 0.946 if other DDs are excluded.
- First Year Inventory (FYI) – a screening tool specific to children at age 12 months. Initial data are promising (PPV = 0.31, NPV = 0.99) but more data are needed to develop reliable screening tools in very young children.

LEVEL 2 (interactive observational assessments)

- These screening tools require higher level of expertise to administer but could be performed by community providers after brief training.
- Established level two screeners include the Childhood Autism Rating Scale (CARS), the Gilliam Autism Rating Scale (GARS), and the Australian Scale for Asperger Syndrome.
- Promising new level two screeners include the Screening Tool for Autism In Two-Year Olds (STAT), the Systematic Observation for Red Flags (SORF), and the Baby and Infant Screen for Children with aUtism Traits (BISCUIT).

Diagnostic Evaluation

Initial Evaluation and Referral

- In any child with developmental delays, an audiology evaluation and Early Intervention or Early Childhood referral is recommended. Not all these children require assessment by a developmental pediatrician unless concerns are present for an ASD or possible organic etiology to the delay.[13,17]
- Any child with a positive screen on an ASD tool or clinical features such as developmental regression should be referred to a developmental specialist for further Reported regression in multiple developmental domains should lead to a more urgent evaluation.[17]

Formal Assessment

- A multidisciplinary 'team' evaluation is considered gold standard for evaluation and diagnosis of ASD, although it may not be available or indicated at a single appointment. This commonly consists of a subspecialty physician, psychologist and speech-language pathologist but may also include other disciplines such as occupational therapists, social workers, or others.
- Includes extensive medical, developmental, and behavioral history focusing on diagnostic features of ASD. Historical information is sought from caregivers who observe the child in social situations across contexts, such as parents, teachers, and therapy providers.
- Includes developmental or psycho-educational and speech-language testing. If these assessments have not been completed recently, or if the child is extremely young, these may not be conducted during the formal evaluation.[17]

Etiological Evaluation

- Recent technological advances have widened the scope of tools available to clinicians and families considering further medical work-up for potential etiologies of ASD. The AAP, along with the American College of Medical Genetics (ACMG) recommend that laboratory investigation be directed by the specific clinical presentation of the child.[17, 18] The AAP specifically notes that recommendations for laboratory work up of ID/GDD (see above), may drive clinical decision-making in a child with both ID/GDD and ASD, whereas diagnostic yield for children with isolated or "essential" ASD is significantly lower.[17]
- ACMG guidelines (2013) recommend that genetic testing should be discussed with, and genetic counseling offered to, all families of children diagnosed with ASD. They recommend a step-wise approach to evaluation based on the individual clinical situation, including a 3-generation family history and physical examination for dysmorphic features, Fragile X screening for all males with ASD, MECP2 sequencing for all females with ASD, and chromosomal microarray for all individuals with ASD regardless of gender. Second tier evaluation, if deemed necessary, should be directed specifically by the clinical presentation.[17]
    - Further work-up or referral to a neurologist or geneticist depends on patient complexity or presence of atypical symptoms. Neuroimaging and EEGs are not routinely recommended upon diagnosis of an ASD unless the child's history or physical exam is concerning features, including history of developmental regression, concern for possible seizures, microcephaly, tonal abnormalities, or focal neurological findings.[17, 18]

This guideline was developed to improve health care access in Arkansas and to aid health care providers in making decisions about appropriate patient care. The needs of the individual patient, resources available, and limitations unique to the institution or type of practice may warrant variations.

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