

Developmental Delay: Timely Identification and Assessment

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This paper outlines the prevalence of developmental delay in children and discusses the recent literature regarding the benefits of early identification and evidence based strategies for developmental surveillance and screening. We describe a systematic approach to the child with developmental delay and the optimal methodology for arriving at the etiologic basis for the delay. A review of the most up-to-date and relevant literature was completed using Pub Med, online databases, and texts. The medical evaluation with specific emphasis on the most recent recommendations for genetic, laboratory and imaging studies is described. The American Academy of Pediatrics algorithm for developmental surveillance and screening is discussed with consideration for the importance of culturally relevant screening tools across populations. In addition, specific screening tools are briefly discussed that may prove beneficial to the primary care provider as he/she implements routine surveillance and screening.

The value of early identification of children with developmental delays has been well documented(1-7). Pediatricians, unfortunately, frequently postpone referring eligible children and their families for early intervention services, and even more experienced clinicians have demonstrated difficulty in the identification of children with mild developmental delays, who are typically the children most amenable to early intervention(5,8). As a result, there has been increasing emphasis on the use of appropriate developmental surveillance and screening for children.

Developmental delay occurs when a child exhibits a significant delay in the acquisition of milestones or skills, in one or more domains of development (*i.e.*, gross motor, fine motor, speech/language, cognitive, personal/social, or activities of daily living). A significant delay has been traditionally defined as discrepancy of 25 percent or more from the expected rate, or a discrepancy of 1.5 to 2 standard deviations from the norm. Global developmental delay is defined as a delay in two or more developmental domains. In addition to delays in development, physicians should also recognize deviations in development. Deviance occurs when a

child develops milestones or skills outside of the typical acquisition sequence. An example of this can be seen in conditions such as cerebral palsy, in which the infant rolls over early secondary to increased extensor tone. Developmental dissociations may also occur. Dissociations arise when a child has widely differing rates of development in different developmental domains. For example, children with autism often have typical gross motor development but significantly delayed language development, therefore language development has dissociated from gross motor development. Finally, developmental regression must be considered. Regression is when a child loses previously acquired skills or milestones, and although less common than the other patterns, should cause the greatest concern since it is often associated with serious neurological and inherited metabolic disorders.

PREVALENCE AND SIGNIFICANCE

As estimated by the World Health Organization (WHO), about 5% of the world's children 14 years of age and under have some type of moderate to severe disability(9). In the United States, developmental and/or behavioral disorders occur in 16-18% of children under 18 years of age(10,11). Other reported

childhood disability prevalence includes Jamaica-15%, Pakistan-15%, and Bangladesh-8%(12). In India, sources have found prevalence of 1.5-2.5% of developmental delay in children under 2 years of age(13,14). These impairments impact not only the child and the family, but also the society, in terms of the cost of providing health care, educational support, and treatment services(15). Evidence supports that early treatment of developmental disorders leads to improved outcomes for children and reduced costs to society(15,16). However, studies in the US have shown only about 1/3 of children are identified prior to school entrance, and as a result, miss out on the proven long term benefits of early intervention(17-19).

STRATEGIES FOR EARLY DETECTION

In order to improve the identification of children with developmental delays so that early intervention can be provided in a timely manner, a significant emphasis has been placed on the routine use of developmental surveillance and screening. Developmental surveillance is defined as a flexible, longitudinal, continuous process through which potential risk factors for developmental and behavioral disorders can be identified(20-22). There are five components to surveillance: eliciting and attending to the parents' concerns about their child's development, documenting and maintaining a developmental history, making accurate observations of the child, identifying risk and protective factors, and maintaining an accurate record of documentation of the surveillance process and findings(23).

Several studies confirm that asking parents about their concerns regarding their child's development, learning, or behavior can provide quality information towards assessing child development(24,25). In addition, it gives the physician an opportunity to educate parents on age appropriate developmental and behavioral milestones. Maintaining a routine developmental history at each visit allows improved identification of delays, dissociations, deviancy and regression. As development is a continuous process, having a detailed history provides the framework needed for early identification of delays.

Screening is defined as a brief, formal, standardized evaluation that aids in the early identification of patients at risk for a developmental and/or behavioral disorder(23). The ideal screening method should use a standardized and validated tool with established psychometric qualities, be easy to perform and interpret, be inexpensive to administer, and have good sensitivity and specificity(26). Furthermore, this tool should be norm referenced and standardized on a population which is representative of the group to be tested. The American Academy of Pediatrics (AAP) describes "good" screening tools as those with sensitivity and specificity in the 70-80% range(23). Screening tools can assist in identifying at-risk children; however, they do not provide diagnoses. When a child passes a screening test it provides an opportunity to promote developmentally appropriate activities and discuss age appropriate milestones. Children who fail a screening test need close follow-up and additional assessment. Additional assessment and early intervention referral should not be delayed by what has typically been called a "wait and see" approach. Early treatment of both developmental and behavioral problems is less costly than treatment for long standing, fully developed disorders and improves the quality of life for both the child and family. Referral for an in-depth diagnostic evaluation by a developmental-behavioral specialist and referral for interventions (i.e. speech and language therapy, occupational therapy, physical therapy, special educational services etc.) do not require a diagnosis.

In 2006, the AAP released a policy statement and algorithm for developmental surveillance and screening in children from birth to 3 years of age(23). The policy statement recommends developmental surveillance at each health maintenance visit in childhood, with the administration of a standardized developmental screening tool for those who have concerns by surveillance(23). In addition, it is recommended that a standardized developmental screening tool should be used routinely at the 9, 18, and 24-30 month health maintenance checks, regardless of surveillance results. If there are concerns by surveillance that do not yield concerns by developmental screen, the child should have early

follow-up visits. However, if the developmental screen is concerning, the child should be referred for early intervention, with developmental and medical evaluations planned. The primary care provider should be the medical home for these children, creating a management plan for children with developmental concerns. The original policy statement did not specifically address older children, but screening at the 4 year or 5 year preventive visit was subsequently recommended for early detection of academic/learning problems(27).

DEVELOPMENTAL SCREENING TOOLS

Finding an ideal screening tool that is easily administered, cost effective, demonstrates strong psychometric qualities, and is culturally relevant remains a challenge. In an effort to assist primary care providers in the US, the AAP has provided a list of screening tools to choose from in a table format in the policy statement. Several of these tools have been validated in other languages. However, the key is finding a tool that meets the ideal qualities described above.

There are a variety of screening tests to choose from, many of which are completed by parents and require only a short period of time to administer and score. These questionnaire screening forms are convenient, as there are no directly administered test items and scoring requires minimal training. For example, the Parents' Evaluation of Developmental Status (PEDS) is a parent interview form that provides an algorithm to guide a need for referral, more screening, or continued surveillance(28). The PEDS has open ended questions to parents, such as "Do you have any concerns about how your child understands what you say?" It takes under 10 minutes to complete and has been translated into over 10 different languages. Another example, the Ages and Stages Questionnaire (ASQ), is a parent-completed questionnaire that may be used as a general developmental screening tool, evaluating five developmental domains: communication, gross motor, fine motor, problem-solving, and personal adaptive skills, for children 4 to 60 months old(29). It relies on specific questions to parents, such as, "Does your baby laugh?" The ASQ is estimated to take under 15 minutes. At this time, neither of these

parent completed screens is available in Indian languages. However, consideration of translation and licensing to establish validity may be of value to increase the availability of these kinds of screening tools, particularly to the nonprofessional health care worker such as the Anganwadi workers.

One of the most well-known and frequently used screening tests is the Denver II, formerly the Denver Developmental Screening Test (DDST). However, in review of the psychometric qualities, it is a more appropriate surveillance tool that can provide a "growth chart" of milestone acquisitions(30). As a screening tool, its specificity of only 43% increases the risk of false positives, which may lead to the over identification of children(31). As such, it can be used to aid in making skilled observations for developmental surveillance, but should not be used for applications beyond its intended purpose.

Several directly administered screening tests have been developed in India. One of the key unifying factors in these screening tests is the minimal training required, which allows for ease of administration by house-to-house child development workers. The Baroda Development Screening Test for Infants was developed from the Bayley Scales of Infant Development and normed on Indian children up to 30 months of age(32). It has motor and cognitive items and provides an age equivalent and a developmental quotient. It was designed to be a test easily administered by health workers for door-to-door surveys, as well as in clinical practice. The Developmental Assessment Tool for Anganwadis (DATA) is another screening test designed for identifying toddlers aged 1.6 to 3 years attending Anganwadis (government sponsored preschool centers in India) and administered by Anganwadi workers, at risk for or with developmental delays(33). The DATA evaluates motor, cognitive, personal-social and language skills. Another screening tool, the Trivandrum Developmental Screening Chart (TDSC) was developed from the Bayley Scales (using Baroda Norms). It is a 17 item screening tool for children up to 24 months of age, requiring minimal training for administration(34). The TDSC can be done in 5 minutes and covers mental and motor developmental milestones. The Disability Screening Schedule (DSS) is a broad

based screen for the identification of major disabilities in children under 6 years of age(35). The authors of the DSS designed it to be distinct from among others as a one-time screening instrument for all major disabilities. It was also created to be easily administered with minimal training.

Another consideration at health maintenance visits are behavioral problems. Children with developmental disorders are at increased risk for these problems, and developmental disorders may first present as a behavioral problem. For example, temper tantrums or disruptive behavior may be a manifestation of language delay. The use of behavioral, social, or emotional screening tools should also be considered in the context of developmental surveillance and screening.

Autism is another specific developmental-behavioral disorder that has been a subject of increasing awareness and concern. Surveillance and screening for autism spectrum disorders (ASD) is also an important part of health maintenance visits. ASD characterized by deficits in social interaction, communication, and restricted, repetitive, stereotyped behaviors, have an estimated prevalence of 20 per 10,000 by meta-analysis(36). Considering these core deficits, children with autism may often present with parental concerns of delayed speech or overall delayed development. Early intervention for autism has been shown to be beneficial(37). Screening should be considered for this specific developmental disorder and has been recommended at 18 and 24 months of age(37). A validated autism screen widely used in the US is the Modified Checklist for Autism in Toddlers (M-CHAT), a 23-item parent completed questionnaire designed to screen children between 16 to 30 months of age. It is available in a number of languages with the validation of these translations underway.

UTILIZATION AND BARRIERS

Even with existing guidelines, one US study found that 71% of general pediatricians almost always used their clinical judgment without using a standardized screening tool in evaluating a child with developmental delay and only 23% used a standardized screening tool, despite the fact that only 30% of

children with developmental disorders are detected prior to 5 years of age(38, 39). Some barriers cited for this low utilization rate include the lack of time to administer a screen, lack of training in the use of a screen, lack of access to assessment and treatment, and inadequate compensation. Furthermore, barriers such as the schedule of recommended health maintenance and irregular adherence of families to the recommended schedule also influence developmental surveillance and screening. The time and cost it takes to administer developmental screening may be addressed by utilizing a parent concern questionnaire(40,41). Most of these questionnaires may be filled out by the parent prior to the visit and easily scored by staff, saving the time needed to administer a screen by the physician. Furthermore, the use of these developmental screening tools is important to pick up concerns in children who may not be seen as frequently as recommended.

Prior to surveillance and screening, it is important to explain to parents the importance of monitoring development along with the goals of surveillance and screening. Once surveillance and screening are initiated, it is imperative to discuss all findings with parents, preferably personally. If a child passes a screen, praise and reassurance should be provided to the parents. However, if a child fails a screen, it should be explained to the parent that a more comprehensive evaluation is required. In the discussion of a failed screen, it is important to emphasize screening tools are not intended to diagnose a developmental disability, but are instead used as guides to further assessment of developmental delays.

Referral to early intervention services, including early childhood education, physical, occupational, and/or speech therapies, should not be delayed when a child fails developmental screening. It has been demonstrated that early intervention services produce improved outcomes for children and society(15,16). For example, for those who participate in these services, higher rates of high school completion, lower rates of juvenile arrests, and lower rates of grade retention have been seen(16). Furthermore, early intervention programs have been shown to reduce the cost of public resources for health, educational, and public assistance ser-

vices(15). In view of the parental anxiety likely to be generated, in spite of reassurances, the primary care provider should play the key role of arranging referrals for early intervention services and further subspecialty consultation, in addition to providing ongoing support for the parents.

When concerns for potential developmental and behavioral problems are present either by surveillance or screening, a detailed medical history and physical examination is an essential part of decision making. This should include reviewing results of the newborn metabolic screen, the most recent vision and audiologic screening, as well as environmental screening (e.g. lead testing).

Past medical history is important for eliciting risk factors including biological (e.g., prematurity), genetic (e.g., Down syndrome), environmental (e.g., lead exposure) and psychosocial factors (e.g., maternal education, family income, marital status etc.).

Protective factors should also be documented and may include a supportive family structure, opportunities to interact with other children in a safe environment, and consistent expectations with age appropriate limitations. A developmental history reviewing the acquisition of developmental milestones should be taken, evaluating gross motor, fine motor, expressive and receptive language, as well as social skills. Finally, family history should include reviewing for developmental delays, learning disabilities, hyperactivity, and other behavioral and psychiatric problems.

The physical examination should include, but not be limited to, evaluating growth parameters, including head circumference, dysmorphology, and a complete neurologic examination(42-44). In evaluating growth parameters, careful attention should be paid to head circumference, looking for macrocephaly, micro-cephaly, or an increased growth velocity. Dymor-phologic examination should look at both minor and major anomalies that might explain the etiology of the developmental delay. A neurologic examination should review strength, tone, symmetry and evaluate for the presence or absence of primitive reflexes.

LABORATORY TESTS AND CONSULTATIONS

The recommended laboratory and imaging studies and consultations recommended by the AAP, the American Academy of Neurology, and the American College of Medical Genetics include cytogenetic studies, DNA testing for Fragile X syndrome, and microarray-based chromosome analysis(42-44). In the child with global developmental delay, 3-4% of the time, an abnormality may be found on standard chromosome analysis(44). For children with an autism spectrum disorder, an abnormal chromosome analysis occurs about 7% of the time(45).

Fragile X syndrome is the most common genetic cause of intellectual disability, and therefore warrants attention in the laboratory work-up of developmental delay(44). Fragile X is phenotypically characterized by intellectual disability, with physical characteristics such as a long jaw, high forehead, long ears, hyperextensible joints, and in males, enlarged testes. Males are more frequently affected than females, and females may show fewer clinical symptoms. The American College of Medical Genetics and the American Academy of Neurology advise the consideration of fragile X testing in the work-up of developmental delay, taking into account cognitive ability, family history, and clinical presentation (44).

Microarray analysis based on comparative genomic hybridization (array CGH) is a more recent method of identifying submicroscopic chromosomal abnormalities where the copy numbers of preselected segments of DNA of the patient is compared with control DNA, allowing detection of deletions and duplications. Limitations of the microarray include the inability to detect balanced rearrangements such as translocations and inversions or single nucleotide changes. Identification of a chromosomal abnormality is important in diagnosis to provide family with an explanation for their child's delay. Furthermore, it allows for genetic risk estimation and counseling regarding future pregnancies of parents, as well as providing awareness of potential medical issues that may require attention in the future.

Neuroimaging with an MRI may be useful in the evaluation of a child with developmental delay;

however, the necessity of imaging varies among the literature(42-44,46). It must be taken into account that most children will require sedation to immobilize them during the imaging study. The yield of neuroimaging is greater in patients with macrocephaly, microcephaly, or abnormal neurologic signs. Other tests that may be considered rely on pertinent history and physical exam findings. An EEG should be obtained if there are clinical features that raise suspicion for epilepsy(44). Metabolic screening should be reserved for those with pertinent history (including where universal newborn screening is not done) or physical findings, as the yield for patients with isolated developmental delay is less than 1%(42-44). Furthermore, if hearing and vision screenings are not current, the child with developmental delay should be referred for formal audiologic and ophthalmologic assessment.

One of the most rewarding experiences primary care providers have and cherish, the opportunity of watching children grow and develop, comes along with the responsibility of recognizing those children who have developmental delays and behavioral problems. Considering the prevalence of developmental delays, the primary care provider must be vigilant in identifying those children who require further evaluation and referral. Early identification leads to early treatment and ultimately, improved long-term outcomes. It is necessary to listen to parents' concerns with regular surveillance, integrate routine screening with health maintenance visits, and refer early, not only to an appropriate medical specialist, such as a developmental and behavioral pediatrician, child neurologist or medical geneticist, but also to early intervention services and therapies which have proven effective, independent of the medical diagnosis. By adopting these practices, one can ensure an optimal and effective system in approaching children with developmental or behavioral concerns and improving their future prospects.

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