DIAGNOSTIC APPROACH TO DEVELOPMENTAL DELAY

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Developmental delay is a common problem in child health and, as such, is a frequent reason for referral of a child for specialty evaluation by a developmental pediatrician or pediatric neurologist. Developmental delays are a group of related, etiologically heterogeneous, chronic disorders that share as an essential feature a documented disturbance in one or more of the recognized developmental domains: motor (gross or fine), speech/language, cognitive, social, and activities of daily living. Usually, the disturbance needs to be significant, that is, a performance of ≥ 2 standard deviations below the mean on an age-appropriate, norm-referenced, standardized developmental assessment. When more than one domain is affected, a global developmental delay exists. When a single domain (motor or speech) is affected, a gross motor delay or a developmental language disorder (developmental dysphasia, specific language impairment) exists. The autistic spectrum disorders (ie, autism syndrome and pervasive developmental disorder) are characterized by a qualitative as well as a quantitative distortion in the acquisition of developmental skills, particularly relating to the social and language domains, with associated, often prominent, behavioral disturbances.

The specialty evaluation of the young child (ie, < 5 years of age) with a suspected developmental delay has a multitude of aims and objectives. These include the following: (1) confirming and classifying the suspected delay, (2) searching for a possible underlying etiology, (3) arranging for the provision of appropriate rehabilitation service interventions, (4) counseling the family regarding the diagnosis, and (5) managing any associated medical or behavioral conditions (eg, spasticity, epilepsy, attentional difficulties, sleep disturbances) that may detract from the child’s full actualization of his or her intrinsic developmental potential.

This chapter describes the elements of the specialty evaluation of the young child with a suspected developmental delay.

History

Much time and effort need to be directed to a careful and detailed history in this particular clinical setting. This begins with a comprehensive family history. The number of children and siblings of the parents; their respective health, developmental, and school (if applicable) attainment status; and the referred child’s place in the birth order needs to be ascertained. Open-ended questions regarding the health and developmental status and school attainment of other family members are helpful. Specific examples of neurologic disorders (eg, epilepsy, convulsive disorders, mental retardation) may need to be suggested so parents don’t inadvertently fail to recall important relevant conditions. The possibility of parental consanguinity has to be probed as well as any familial neonatal or infantile deaths or maternal pregnancy losses. Ethnic heritage and geographic origin of the family may, in some instances, provide a clue to etiology.

Details regarding the mother’s pregnancy and prenatal care with the affected child should be established. Adverse
prenatal events, such as per-vaginal bleeding, gestational diabetes, intercurrent infections, or medical conditions, should be discussed. Documentation of maternal prescription medication, tobacco, alcohol, or illicit drug use is necessary. The timing of labor (premature or term), its spontaneous or induced onset, duration, mode of presentation, and means of delivery must be ascertained. Suggestions of difficulties during the delivery process, such as meconium staining or abnormalities in fetal heart monitoring, are relevant. The reason for cesarean section, if any, is important to note. Birth weight, Apgar scores (especially beyond 5 minutes, if early scores are of concern), and the duration of the infant’s postnatal hospital stay are important objective parameters. The neonatal course should be probed for any clinical findings, such as seizures, encephalopathy, or feeding difficulties, that suggest early concerns that may lead to a focus on a prenatal or perinatal etiology.

Subsequent to birth, the child’s medical history should be ascertained. Hospital admissions, surgical procedures, possible chronic medical conditions, and current medication use need to be documented. Parental socioeconomic (ie, educational attainment, occupation), marital, and custody status, together with existing child-care arrangements, are relevant to establishing a proper social and familial context for the child under evaluation. The provision, either previously or currently, of any special services, particularly those of a rehabilitative nature, should be ascertained and relevant reports from these services obtained.

With the above historical background established, the evaluator can then turn to specific developmental issues. The age of initial parental concern and the precise domain (ie, motor, language, social) of original concern should be elicited. Developmental progress in each of the domains should be established. Key sequential motor skills include rolling over, sitting, crawling, pulling up to stand, cruising, and walking independently. Key progressive language skills include cooing, babbling, saying initial words, speaking two- and three-word phrases, and correctly using pronouns, plurals, and subject-verb-object sentence structure. Parental recall may often be sketchy and frustratingly elusive, so it may be useful to obtain from the caregiver an idea of the child’s capabilities at specific ages (eg, first or second birthday). If the parents have other children, they may phrase their recall for the child of concern by comparing the child’s pace of skill acquisition with those of their other children. The possible loss of skills or developmental regression should be specifically ascertained, as this may be an important clue to a possible neurodegenerative or metabolic disorder. Current developmental performance in each domain should be ascertained. For the older child, performance of activities of daily living, such as toileting, dressing, feeding, and self-hygiene, is a good barometer of overall developmental attainment. For the older child with motor concerns, copying and writing skills, running, riding a tricycle, and going up and down stairs are a good index of status. For the language-impaired child, assessment of comprehension and a grasp of body parts, colors, shapes, objects and their uses, and simple analogies provide a good idea of linguistic attainment. The possible presence of any autistic features, such as poor eye contact, emotional inappropriateness, desire for sameness, repetitive behaviors, lack of pretend play and appropriate social interaction should be specifically sought. Given their frequency among children with developmental delays, the possible presence of coexisting paroxysmal behaviors suggestive of epilepsy, disruptive sleep disturbances, ongoing behavioral concerns, and feeding difficulties also should be specifically searched for.

**Physical Examination**

The physical examination is an integral part of the diagnostic evaluation of the delayed child. The examination may alternatively suggest a specific syndrome due to the observation of a constellation of particular findings, confirm a diagnostic suggestion apparent on history taking, or document findings that suggest an increased likelihood of an etiologic yield on subsequent laboratory testing.

Much of the neurodevelopmental examination takes place by observation during the extended history-taking described above. Therefore, a child-friendly environment is necessary, with provisions made for the availability of age-appropriate playthings, including blocks, picture books, crayons, paper, simple puzzles, stuffed animals or dolls, and balls. Through the child’s interaction with this environment, the manipulation and use of toys, expression of curiosity in the surroundings, grasp of language, imaginative play skills, and personal and social interactions can be assessed by the observer in a detached and nonthreatening manner. This playtime can take place while the young child sits on the lap of a reassuring caregiver or, for the older ambulatory child, in a child-sized chair or at a table set up in the examining room. Before proceeding to the assessment’s necessary formal aspects, it is crucial that the evaluator establish a rapport with the young child. The reassuring physical proximity between child and caregiver should be maintained when at all possible. The child needs to know what to expect by being told what will happen next. The sequence of the examination needs to be fluid and adaptable to immediate opportunities and, if possible, direct physical manipulation of a body part by the examiner deferred to the end.

This observation of spontaneously expressed developmental skills is complemented by a more formal developmental assessment that permits accurate documentation of the child’s current developmental status in all relevant...
domains. Fine motor skills are best assessed by the use of blocks and pen/paper tasks and gross motor skills by observation, gait analysis, and ball playing. Language is assessed by the child’s ability to identify pictures, body parts, colors, and shapes and to explore concepts and analogies. Spontaneous speech and storytelling by the child provides a means of assessing vocabulary and grammatical and semantic capabilities. Comprehension skills can be tested by following progressively more complex commands conveyed by the examiner or caregiver. Cognitive skills can be evaluated by the child’s grasp of puzzles and such concepts as small/big, short/long, open/close, and under/over, as well as simple counting skills. Activities of daily living are best assessed by questioning the caregiver; however, observation of how the child dresses and undresses may be instructive in the examining room.

Particularly relevant aspects of the general physical examination include current height and weight measurements and an understanding of their evolution over time and a careful search for possible dysmorphic features. Dysmorphology needs to be considered within the context of ethnic and familial variation. The skin must be examined in detail to search for possible stigmata of a neurocutaneous disorder, such as café-au-lait spots or hypopigmented macules and an underlying myelodysplasia that may manifest itself overtly simply by a cutaneous abnormality over the spine. Storage disorders commonly manifest by palpable hepatosplenomegaly in addition to a coarsening of facies.

An essential element of the neurologic examination is obtaining a head circumference (occipitofrontal), which is then plotted to yield an age-appropriate percentile. Documented microcephaly (< 2%) or macrocephaly (> 98%) mandates the obtaining of prior measurements and plotting these values over time. In addition in the setting of microcephaly or macrocephaly, parental head circumference measurements should be obtained and plotted.

Formal neurologic assessment includes a search for nystagmus, facial paresis, excessive drooling, dysphagia, or dysarthria. Visual fields should be ascertained by confrontation as well as pupillary responses to light. Office screening for a primary sensory impairment (either visual or auditory) as well as a funduscopic examination is necessary, which may be limited because of a patient’s age and lack of cooperation. Motor examination focuses on the detection of any asymmetries or lateralizing features with respect to bulk, strength, tone, stretch reflexes, and planar responses. The quality of limb movements also should be appreciated and any dyskinesias (eg, dystonia, athetosis, chorea, tremor, dysmetria) documented. Gait should be assessed if the child is ambulatory and described in detail if abnormal. Arising from a squatting position or a supine posture on the floor (Gowers’ sign) is a good test for proximal weakness. Running down an adjacent hallway, ascending or descending stairs, standing on one foot and hopping (if possible), copying simple figures, and catching/throwing/kicking a ball provide a good assessment of dexterity, coordination, and motor planning skills.

**Laboratory Investigations**

Laboratory investigations in this setting are directed at establishing a possible etiology underlying the child’s specific developmental delay. This etiology may or may not be apparent at the end of the clinical assessment. Thus, laboratory testing may be directed by findings on history or examination (ie, confirmatory) or undertaken on a screening basis to detect a previously unsuspected etiology. Laboratory testing needs to be selective, as extensive nondirected testing is neither feasible nor justified on the basis of medical indications, invasiveness, or costs.

Determining an underlying etiology is important for many reasons. It imparts an understanding of the pathogenesis to the family, answering their “need to know why.” It also may have implications with respect to an accurate estimation of recurrence risk if genetic factors are found to be at play, which may subsequently involve parental testing. It also allows for more accurate prognostication of what can be expected for the child. A specific etiologic diagnosis may also modify ongoing medical management or programmatic follow-up (eg, tuberous sclerosis, neurofibromatosis). A diagnosis also empowers family members by providing them with sufficient information to act on their child’s behalf and brings closure to the first stages of dealing with having a child with developmental delay.

Etiologic yield and appropriate laboratory testing is highly dependent on the specific subtype of childhood developmental delay under consideration. Recent retrospective and prospective studies have shown that more often than not an underlying etiology can be determined in the situation of global developmental delay. Three-quarters of etiologies in cases of global developmental delay are accounted for by four diagnostic categories: (1) cerebral dysgenesis, (2) hypoxic-ischemic encephalopathy, (3) antenatal toxin exposure (ie, alcohol or drugs), and (4) chromosomal abnormalities (including fragile X). Clues to the successful etiologic determination on the basis of history and physical examination include documentation of antenatal toxin exposure, microcephaly, and focal motor findings. The presence of any autistic features is a negative predictor of successful etiologic yield. The yield of karyotyping is the same whether it is done for screening or when a condition is suspected because of dysmorphic features. In contrast to karyotyping, neuroimaging is three times more likely to identify an etiology if one is suspected clinically (ie, microcephaly, focal findings).
Routine metabolic testing for an extensive list of inborn errors of metabolism cannot be justified. However, given the implications of their ascertainment, for reasons of treatment, prognosis, and recurrence risk, diagnostic vigilance for these disorders must be maintained. Clinical situations that should prompt careful consideration for metabolic testing include family history, parental consanguinity, documented developmental regression, episodic decompensation, suggestive dysmorphism, involvement of nonectodermal derived organ systems, and suggestion of white matter involvement.

In the setting of a child with a global developmental delay, the following recommended testing has been put forward. Lead testing should only be done on those children with identifiable risk factors for excessive lead exposure. Similarly, testing for thyroid hormone status should target those in whom newborn screening was not undertaken or in whom specific systemic features of hypothyroidism are present. Metabolic screening is indicated in the context of no prior newborn universal screening or historical or physical examination findings that are suggestive of a possible metabolic etiology. These findings include parental consanguinity, prior fetal or child loss, history of episodic decompensation, developmental regression, dysmorphic features, or hepatosplenomegaly. Karyotyping (high resolution) is recommended routinely even if there are no dysmorphic features, as well as fragile X mental retardation (FMR-1) molecular genotyping. Fluorescent in situ hybridization (FISH) studies are reserved for those situations in which the delay is unexplained or a specific syndromic diagnosis is suspected. Obtaining an electroencephalogram (EEG) is not useful from a diagnostic point of view and should be pursued only if there is a suggestion of seizures or a specific epilepsy syndrome. Routine neuroimaging is recommended, with magnetic resonance imaging preferable to computed tomography, especially in the context of documented physical findings. All children with a global developmental delay should undergo hearing and vision assessment.

With respect to the child with a single-domain developmental delay, there is a marked dichotomy to etiologic yield. The child with restricted motor delay is more likely than not to have an underlying etiology that can be determined. This is especially so if physical findings, such as weakness, asymmetry, and tone or reflex changes are demonstrable. Once again, specific laboratory testing has to be directed by findings on history or physical examination. These findings, if present, should allow for classification of underlying etiology into either a central or peripheral origin. If central, imaging of the brain is indicated. If peripheral, studies targeting muscle/nerve integrity should be pursued (ie, creatinine kinase, serum acid-base status and lactate levels, electromyography/nerve conduction studies). The situation is markedly different among children with a developmental language disorder. An underlying etiology is rarely appreciated (<5% of cases), despite careful evaluation. Laboratory testing in this clinical situation can be restricted routinely to detailed audiometric assessment and perhaps EEG if there is a suggestion of a possible acquired epileptic aphasia (Landau-Kleffner syndrome). Clinical features that are suggestive of this include objective loss of previously acquired language skills and behavioral disturbances. EEG should include, when feasible, a sleep study.

With reference to the child with an autistic spectrum disorder, recommended testing presently consists of karyotyping and an FMR-1 molecular genotype, especially if there is coexisting mental retardation, a positive family history, or suggestive dysmorphic features. Similarly, as for global developmental delay, metabolic testing should be undertaken only if there are suggestive clinical or physical examination features. At present, an EEG is not routinely recommended in the setting of an autistic spectrum disorder, but only if seizures are suspected or if there is regression such as a significant loss of social and communication function, which raises the possibility of Landau-Kleffner syndrome. At present, routine neuroimaging is not indicated for autistic spectrum disorder, even in the setting of macrocephaly.

Other Health Professionals

The diagnostic evaluation of the delayed child by a developmental pediatrician or child neurologist is not a solo or stand-alone effort. Other health professionals with disparate yet complementary expertise need to be consulted to ensure a complete and thorough evaluation and to ensure that appropriate services are provided to the affected child. The high frequency of primary sensory impairments in this population requires careful consideration of audiometric and ophthalmologic assessments. The developmental concerns profiled following history and physical examination will mandate referrals, where appropriate, to occupational therapy (fine motor, activities of daily living), physical therapy (gross motor), speech-language pathology (language), or psychology (cognition, social, behavioral). These professionals possess the skills to apply standardized assessments that objectively document the child’s developmental deficits in a way that is far more rigorous than what can be done in the standard office examination described above. Furthermore, these professionals assume responsibility for ensuring the implementation of appropriate goal-directed therapeutic interventions that optimize the child’s functional capabilities. They also serve as information sources for available community-based resources. Additionally, specific care needs (eg,
Second Visit
A second patient visit with the specialist is an integral part of the diagnostic assessment of the young child with a developmental delay. The timing of this visit should be 4 to 6 months after the original visit. Developmental progress should be ascertained to rule out a progressive encephalopathy (ie, neurodegenerative process). Results of laboratory investigations should be reviewed, as well as the evaluations of the other professionals involved. The specialist should combine and integrate these reports and then counsel the parents regarding the precise nosology (ie, developmental delay subtype) of their child’s disability and the determination, or lack thereof, of an underlying etiology. The current provision of rehabilitation support services should be determined, and if lacking relevance to the needs of the child, the sources and means of obtaining such services should be discussed. Developmental progress in the interim should be highlighted and implications regarding future prognosis discussed. For the child nearing school age, educational (ie, schooling) options will need to be enumerated and their appropriateness for the particular child addressed.

Suggested Readings

Practitioner and Patient Resources
National Association for Child Development
The Weber Center
2380 Washington Blvd, 2nd Floor
Ogden, UT 84401
Mailing address:
PO Box 380
Huntsville, UT 84317
Phone: (801) 621-8606
E-mail: info@nacd.org
http://www.nacd.org
The National Association for Child Development is an international organization of parents and professionals dedicated to helping children and adults reach their full potential. Founded in 1979 by internationally recognized educator and lecturer Robert J. Doman Jr, NACD designs very specific home neurodevelopmental programs for infants, children, and adults.

Family Resource Services, Inc.
PO Box 1146
Magnolia, AR 71754
E-mail: dorothy@frs-inc.com
http://www.frs-inc.com
An autism and developmental disabilities resource catalog.