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Office Evaluation of the Child With Developmental Delay

Michael Shevell, MD, CM, FRCPC

Developmental delays are a common problem in child health and a frequent reason for referral to a pediatric subspecialist. The office evaluation of the young delayed child has a number of objectives including primarily precisely categorizing the delay subtype together with rationally selecting investigations for determining a possible underlying etiology. Counseling the affected family regarding the diagnosis and its prognosis, identifying possible coexisting conditions that merit interventions, and ensuring appropriate rehabilitation service provision are also important objectives of this office assessment. An outline of the key features of the relevant history and physical examination together with guidelines regarding investigations are provided to best meet all these objectives within existing time and practice constraints.
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Developmental delays are a common problem in child health.[1] Although standardized developmental screening has been recommended by pediatric professional societies,[2] typically the delayed infant or young child is initially identified through a process of developmental surveillance in which parental concerns are elicited by targeted questioning at the time of a routine well-child visit.[3] Frequently, the delayed child is then referred by the primary care provider to a pediatric subspecialist (ie, pediatric neurologist or developmental pediatrician) for a detailed medical evaluation.[4]

Such a specialty evaluation of the preschool-aged child has a number of interdigitating objectives[5,6] including (1) confirmation and categorization of the child's developmental delay; (2) the identification of a possible underlying etiology; (3) referral to and assurance of provision of appropriate rehabilitation services and resources; (4) counseling of the affected family regarding diagnosis, etiology, and prognosis; and (5) the identification of any possible coexisting conditions that may require medical intervention and ongoing management. The first objective of confirming and precisely categorizing the type of delay is predominant because this will then direct efforts focusing on the remaining nonhierarchical, and indeed equally important, objectives.

The specialty evaluation of the delayed child begins with a detailed and particular history and physical examination that is an essential precondition to realizing the objectives listed earlier.[5-7] With our increasing multicultural and multilinguistic practice environments, every effort should be made to obtain the history and conduct the examination in the language most familiar to the family unit.

History

A detailed background is necessary to precisely situate the child's particular story. This begins with a family history. A 3-generation pedigree of the referred child's family is required that at a minimum ascertains health and developmental status of individual family members as well as the occurrence of specific neurologic conditions. Examples of such disorders (eg, neuromuscular, mental retardation, and epilepsies) may need to be overtly suggested so that relevant conditions are not inadvertently omitted by the parents. Maternal pregnancy losses or early neonatal or infantile deaths may be suppressed because of their emotional pain of recall and also may need to be specifically asked for. The possibility of parental consanguinity, precise ethnic heritage, and geographic origin are relevant questions that, although uncomfortable to probe for, have to be asked.

The possibility of a prenatal or perinatal etiologic origin to the child's developmental delay requires that attention be

From the Departments of Neurology/Neurosurgery & Pediatrics, McGill University, Division of Pediatric Neurology, Montreal Children's Hospital-McGill University Health Centre, Montreal, Quebec H3H 1P3, Canada.

Address reprint requests to Michael Shevell, MD, CM, Room A-514, Montreal Children's Hospital, 2300 Tupper Street, Montreal, Quebec, H3H 1P3. E-mail: michael.shevell@muhc.mcgill.ca

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devoted to obtaining details regarding the mother's pregnancy and delivery of the affected child. The provision of antenatal care should be documented as well as the occurrence of any antenatal ultrasounds and amniocentesis and their results. Specific questions concerning possible prenatal adverse events such as per-vaginal bleeding, gestational diabetes, pregnancy-induced hypertension, intercurrent infections, intrauterine growth retardation, or maternal medical conditions should be directly asked. Maternal prescription medication, alcohol, tobacco, or illicit drug use should be documented. In the nonprimigravida mother, the relative quantity of antenatal fetal movements compared with other pregnancies experienced may be of interest. The timing of labor (preterm or term), its onset (spontaneous or induced), duration, presentation (vertex or breech), and mode of delivery (vaginal, assisted, or cesarean) should be determined. The reason for a cesarean is important to note as are suggestions of problems during the delivery process itself such as the prolonged rupture of membranes, maternal fever, meconium staining, and any abnormal fetal heart rate monitoring.

Birth weight, APGAR scores (especially beyond 5 minutes if low initially), need for, and specifics of, caseroom resuscitation and possible admission to a neonatal intensive care unit are important indicators of an infant's immediate postnatal status. Determining the duration of a child's post delivery hospital stay is a simple way of ascertaining if there were any clinically significant neonatal concerns. If beyond that expected locally, the reason(s) thereof should be documented. In particular, possible postnatal signs of suspected encephalopathy or feeding difficulties are good markers of a possibly compromised newborn nervous system. Suggestions of adverse prenatal, perinatal or neonatal events may require directly obtaining the maternal obstetrical or the child's neonatal records for detailed review.

The child's post neonatal medical history then needs to be documented. This includes any chronic medical conditions, hospital admissions or surgical procedures. Concurrent medication use, prior assessments or provision of specialty services pertaining to developmental concerns, especially if rehabilitative in character, require documentation. To understand the child's social and family context, parental origin (ie, foreign or domestic if foreign current immigration standing), socioeconomic status (ie, educational attainment and parental employment), marital situation, custody, and child-care arrangements should be determined.

Once this background is obtained, the evaluation can then move to a specific and detailed developmental history. This begins with determining the age and domain (ie, motor, language, and social) of initial parental concern. Developmental trajectory in each domain should be established, although parental recall, especially in multiplex families, may be difficult to pin down precisely (Table 1).[8] Key milestones usually recalled well are walking independently and first meaningful specific words. Comparing a child with other children (their own or peers) or recalling a child's developmental performance at a specific personal milestone (ie, first or second birthday) may provide a snapshot of delay. Essential in this elicitation is the careful probing for any possible loss of developmental skills or regression and establishing whether the child's delay is global, domain specific (ie, motor and language), or has autistic features. The latter is ascertained through specifically asking regarding eye contact, emotional awareness and appropriateness, desire for sameness, presence of repetitive behaviors or obsessive preoccupations, and the quality of social interactions especially that pertaining to play behaviors. Current developmental attainment in all domains should be documented subject to confirmation through the physical examination as well as functional attainment pertaining to activities of daily living such as feeding skills, toileting, dressing, and self-hygiene that may or may not be age appropriate.

With a developmental profile established by history, a functional inquiry can then be pursued that targets coexisting conditions that occur frequently in the setting of developmental delay.[7] These include paroxysmal disorders that may be epileptic in origin, sleep disturbances (ie, frequent noctur-

Table 1 Guide To Early Child Development and Functional Milestones

	Motor	Language	Social/play
2 months	Head up in prone		Smiles, fixes, and follows
3 months	Head/chest up in prone, grasp placed object	Coos	Laughs
4 months	Rolls, reaches		
6 months	Sits with support, transfers	Babbles, turns to sound	Mouthing objects
8 months	Sits without support, weight bears	Turns to name	
10 months	Pincer grasp, starting to cruise, crawling	"Bye-bye" wave	Drinks from cup
12 months	Walks but falls easily	First words	Finger feeds, objects in and out of containers
15 months	Walks steadily, scribbling	Pointing, multiple single words	Spoon use, assists in dressing
18 months	Up/down stairs with assistance, climbing, throws ball	Two-word phrases, pointing to body parts	Build towers, play with others
24 months	Up/down stairs, 1 step @ time, kicks ball	Three-word phrases, pronoun	

nal awakening, failure of sleep consolidation), behavioral concerns that are disruptive to the family unit, and attentional deficits that may be reflected in hyperactivity and limit the effectiveness of rehabilitations interventions attempted and feeding difficulties. In many families, it is these coexisting conditions that may be the greater familial challenge rather than the actual delay itself.

Physical Examination

The physical examination is an essential part of the comprehensive assessment of the delayed child. The physical examination comprises general physical, neurologic, and developmental elements. Results obtained may (1) confirm an etiologic suspicion suggested originally by history, (2) put forward a novel etiologic possibility previously unsuspected, or (3) document findings that may suggest a heightened probability of finding an etiology on screening tests. In addition, developmental elements of the examination will also allow precise categorization of the child's developmental delay. The examination should be conducted in a child friendly environment in which ideally age appropriate play things such as paper, crayons, blocks, picture books, puzzles, stuffed animals, dolls, and small pretend play situations (ie, doll houses and work settings with figurines) are made available to the child. During the initial extensive history taking, observation of the child's own spontaneous exploration of these play opportunities will provide insight into the child's neurodevelopmental profile, encompassing language, cognition, motricity, play, and sociability in a detailed nonthreatening manner. This also serves the purpose of putting the child at ease for the more formal aspects of the examination in which cooperation at times may be tenuous.

Physical proximity of the child with a caregiver is reassuring and should be maximized. Thus, in the infant or toddler, much of the examination may take place with the child on the caregiver's lap. Establishing a rapport with the child is essential. Even the preverbal child needs to be told what to expect next. Fluidity of examination is required to take advantage of opportunities as they present themselves. Direct manipulation or the placing of hands on the child needs to be deferred to the end so that cooperation is maintained for as long as possible. These caveats, however, should not preclude comprehensiveness of examination.

The general physical examination requires obtaining and plotting current somatic measurements for height and weight. Possible dysmorphic features need to be looked for within the context of ethnic and familial variation. Possible stigmata of a neurocutaneous disorder (ie, café-au-lait spots and hypomelanotic macules) or myelodysplasia mandates undressing the child fully and inspecting the skin and spine thoroughly. Hepatosplenomegaly and coarsening of the facies may be a tip off to an underlying storage disorder.

Obtaining and plotting an occipital-frontal (ie, head) circumference is essential to obtain a gender- and age-appropriate percentile. Documented microcephaly (less than the 2nd percentile) or macrocephaly (greater than the 98th percentile) requires obtaining and plotting prior measurements for the child and obtaining and plotting the head circumference for each biological parent and the child's siblings if available.

Formal neurologic assessment include cranial nerves to document any possible aberrant pupillary responses, visual field defects, retinal abnormalities, nystagmus, facial paresis, excessive drooling, head tilt, dysphagia, or dysarthria. Primary sensory impairments affecting vision or hearing occur commonly in the delayed child and should be screened for in the office setting; however, "normal" office results should not preclude a more detailed ophthalmologic and audiometric assessment by specialists. Motor examination targets by observation any evidence of lateralizing features, asymmetries, or dyskinesias (ie, dystonia, athetosis, chorea, and tremor). Formal testing of tone and stretch reflexes allow for postulation of possible upper or lower motor unit pathology. Arising from a supine or squatting position, going up and down stairs, walking or running down an extended hallway, jumping, and hopping in place all provide for observational assessment of the motor system's integrity. Copying figures and simple ball games provide an insight into motor planning, dexterity, and coordination skills. Cerebellar function can be assessed by the observation of gait and the smoothness and accuracy of reaching for objects.

A formal developmental assessment supplements and fills in the information obtained through initial observation of the child's integration and play in the examination setting. Fine motor skills are assessed through manipulation of blocks and pen and paper tasks. Gross motor skills are revealed through ball playing, gait, running and going up and down stairs. Spontaneous speech provides insight into vocabulary, grammatical and semantic capabilities. Story telling and following complex commands provide an illustration of the child's comprehension. Language can also be assessed through picture, body part, color, shape and item recognition and the use of analogies and oppositional concepts (ie, short/long, big/small, open/close, on/under) which also provide insight into cognitive capability.

Formal developmental measures have been developed for use in the office by physicians.[7] A number of such measures exists; however, the simple reality is physician time is too limited in the busy office setting to use these measures on a regular basis.[3] Essentially, allied health professionals in related disciplines (ie, occupational therapy, physiotherapy, and speech language pathology) have more experience, time, and skill in applying such standardized measures and their expertise, where available, on this aspect that should be deferred to.

Diagnosis

Once the history and physical examination are completed, sufficient information typically exists to provide a diagnosis of a specific neurodevelopmental disability. Such a diagnostic formulation is essential because it guides further testing and referral. If delay is apparent in more than 1 developmental domain (typically all domains are then affected) in a young child less than 5 years of age, a global developmental delay can be diagnosed.[7,9-11] If delay is restricted to a single domain

(ie, motor or speech/language), either a gross motor delay or developmental language impairment can be inferred to exist.[12] If in addition to motor delay, evidence for spasticity or dyskinesias are apparent, with or without coexisting cognitive and speech limitations, cerebral palsy may be diagnosed.[13] If sufficient qualitative impairments in either social and/or language skills are apparent, an autistic spectrum disorder can then be diagnosed.[14] Sometimes a threshold for such a diagnosis may not be readily apparent in the clinical situation because some children with global developmental delay may have some autistic features present of insufficient quantity to merit an autistic spectrum diagnostic label.

Laboratory Investigations

Recent studies have shown that etiologic yield and appropriate laboratory testing is highly dependent on the specific subtype of early childhood developmental delay diagnosed.[12,15-19] Searching for an etiology is important for reasons pertaining to recurrence risks, prognosis, ongoing medical management, treatment intervention (although rarely currently), and especially for family empowerment. Indeed, often finding a precise underlying diagnosis may serve to improve access to rehabilitation service delivery and provide closure to the first stages of a family's dealing with their child's developmental delay.

Laboratory testing in the setting of childhood developmental delay needs to be selective and rationally based because extensive nondirected testing is neither justified nor feasible on the basis of interventions, yield, invasiveness, associated risks, or costs. At present, 2 practice parameters have been formulated regarding the diagnostic approach to the child with either global developmental delay[11] or cerebral palsy,[20] two situations in which an etiologic yield can be expected more often than not.

For a child with a global developmental delay, an etiologic yield can be expected in between 50% to 60% of cases, especially if no autistic features are noted.[13,18,21] Three quarters of etiologic diagnoses can be accounted for by 4 diagnostic categories: (1) intrapartum asphyxia, (2) cerebral dysgenesis, (3) chromosomal abnormality (including Fragile X) or specific genetic syndrome, and (4) antenatal toxin exposure (ie, alcohol or drug). If subsequent to the history and physical examination a specific diagnosis is strongly suspected, laboratory investigations should selectively target this possibility (ie, neuroimaging for intrapartum asphyxia, fluorescent in situ hybridization for Prader Willi syndrome or Angelman syndrome, and FMR1 triplet repeat analysis for Fragile X).[11] In the absence of any suspected diagnosis, at present high-resolution banding karyotyping, FMR1 triplet repeat testing, and neuroimaging are suggested on a screening basis with a positive yield in approximately one sixth of all cases.[11] The yield of neuroimaging improves 3-fold if any microcephaly or lateralizing findings are present, whereas the yield of karyotyping is consistent whether dysmorphology is documented or not.[18] With respect to neuroimaging, magnetic resonance imaging is preferable to computed tomography where readily available.[11]

Routine metabolic testing (capillary blood gas, lactate, ammonia, liver function studies, serum amino and urine organic acids, and very long chain fatty acids) at present cannot be justified except in certain clinical situations that include prior family history of a similarly affected child, parental consanguinity, documented developmental regression, episodic decompensation, suggested dysmorphology, involvement of nonectodermal derived organ systems, the suggestion of white matter involvement, or nonscreening as a newborn.[11] Electroencephalography should be pursued only if there is a possibility of a possible paroxysmal event (ie, seizures) based on the history obtained.

In the setting of isolated language delay, careful evaluation rarely yields a specific etiology.[22] Routine laboratory testing in this clinical setting should be restricted to detailed audiometric assessment and perhaps an electroencephalogram if suspicion regarding a possible acquired epileptic aphasia (ie, Landau-Kleffner syndrome) exists, and, if suspected, this should include a sleep study. For isolated motor delay with a physical examination that suggests a possible peripheral (ie, lower motor unit) etiology, studies targeting the integrity of the muscle and nerve should be pursued.[12] This would include creatinine kinase, capillary blood gas, lactate, and electromyography/nerve conduction studies. Molecular genetic studies are often used to precisely specify a suspected etiology (ie, dystrophin analysis for muscular dystrophy, CTG triplet repeats in the myotonin gene for myotonic dystrophy, and PMP-22 in Charcot-Marie-Tooth syndrome) with biopsy of the muscle and/or nerve now largely restricted to the elaboration of structural or metabolic myopathies and axonal neuropathies.[23] For children with documented cerebral palsy, neuroimaging is usually sufficient.[20] If a cerebral vascular accident is documented, detailed coagulopathy studies may then be pursued.[20] Some of the cerebral dysgeneses may call for specific molecular studies (ie, XLIS and LISI for lissencephaly).[20]

For the child with an autistic spectrum disorder, recommended testing consists of a Fragile X molecular genotype and karyotyping, especially if there is a family history, coexisting mental retardation, or suggestive dysmorphic features.[24] A backdrop of regression of social or communication skills may suggest the possibility of a Landau-Kleffner syndrome giving rise to the need for an electroencephalogram, which should ideally include a sleep study, although such an investigation may be challenging to undertake given behavioral limitations. At present, routine neuroimaging is not recommended for children with an autistic spectrum disorder.

These recommendations reflect our present knowledge and technology limitations. Advances in genetics such as complimentary genomic hybridization, subtelomeric probes, genomic microarray, and proteonomics, together with advances in neuroimaging that provide complementary means of assessing brain structure or function such as volumetric magnetic resonance imaging, diffusion tensor imaging, functional magnetic resonance imaging, and magnetic resonance spectroscopy may radically alter the diagnostic approach to the delayed child in the near to intermediate future.[7]

The comprehensive and complete evaluation of the preschool-delayed child does not take place in isolation.[6] Ongoing management also requires that representatives of other disciplines beyond the medical become involved and actively engaged in providing needed assessments and services. Complementary expertise is required to fully evaluate developmental concerns often through the application of standardized assessments that objectively documents a child's developmental profile and apparent deficits in a more rigorous way than that which can be accomplished in the office evaluation described previously. These health professionals include occupational therapy, physical therapy, speech-language pathology, and psychology. In addition to assessments, these professionals will typically assume responsibility for the provision of goal-directed therapeutic interventions and the obtainment of appropriate community resources and will be useful adjuncts as information resources and counselors to the family as they adapt to their child's developmental concerns and limitations.

The high frequency of primary sensory impairments affecting either the vision or hearing apparatus and their relative correctability, together with limitations in routine office screening for such impairments, mandates careful consideration of both formal audiometric and ophthalmologic evaluations.[25,26] The high frequency of genetic etiologies and concerns, together with the increasing complexity of available molecular laboratory testing, often calls for the involvement of a genetic consultant. Specific care needs related to issues concerning behavior, feeding, respite care, or financial concerns or available programs may mandate the involvement of psychology, nursing, and social service expertise in this population.

Not to be overlooked is the value of a second encounter with the child and family 3 to 9 months after the initial visit and assessment.[6] Recent longitudinal studies have noted that developmental trajectories are not necessarily smooth or predictable, thus highlighting the dynamic nature of development across all domains.[27] A second visit serves to validate or correct initial impressions. It also serves to either support or refute the possibility of a progressive encephalopathy or a neurodegenerative process that would feature a loss of previously acquired skills and new findings on examination. The existence of such a possibility will call into play an especially vigorous etiologic search often involving quite rare disorders and esoteric investigations. A second visit allows for the review and integration of evaluations from allied health disciplines that provides precise specification of the child's developmental delay subtype. Results of laboratory investigations can be reviewed and issues of etiologic diagnosis addressed that may require further testing to be arranged. The provision, or lack thereof, of rehabilitation services can be confirmed together with a plan for long-term community-based resources. It also provides a forum for families to bring forth specific care needs related to issues of feeding, behavior, or sleep that may be more challenging to the family unit than the child's actual delay. Most importantly, it provides a forum for the family to bring forth questions concerning their child's present status, future prognosis, and realistic expectations.

Summary

The office evaluation of the young child with a developmental delay challenges the pediatric subspecialist on a number of levels that addresses both the science and art of medicine as a diagnostician, service provider, and counselor. A "quick fix" is not possible and precious time is required to meet all the necessary objectives and goals attached to this evaluation. Personal satisfaction can be found in meeting these challenges and realizing one's integral role in facilitating a family's recognition and adaptation to their child's chronic disorder and thus optimizing the child's eventual functional attainment.

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