CHALLENGING CASE: DEVELOPMENTAL DELAYS AND REGRESSIONS

Early Delay in Motor Development*

CASE 1

Three-month-old Juanita was being evaluated by her pediatrician during a health supervision visit when bilateral lower extremity hypertonicity is noted for the first time. Tone appears to be mildly to moderately and symmetrically increased in her hip adductors and at the ankles. Three beat ankle clonus bilaterally, intact sensation to light touch, and normal spontaneous movements of the legs are recorded. Deep tendon reflexes at both knees and ankles are 2+. She bears weight on both legs when held upright. Upper extremity tone, strength, and reflexes are normal. Juanita’s full-term gestation, labor, and delivery (vertex) were uncomplicated. Apgar scores were 8 and 9 at 1 and 5 minutes, respectively. Neurological examinations at birth and at 2 weeks of age were normal.

CASE 2

Ten-month-old Cedric is seen by a pediatrician for the first time for an upper respiratory infection. The physician makes the observation that Cedric cannot sit steadily without support; he maintains a sitting position with the forward support of both hands in a tripod position. Recognizing that this may represent a delay in motor development, the pediatrician performs a complete neurodevelopmental examination.

There is mild bilateral hypotonia with intact, symmetrical, deep tendon reflexes, 3+, and appropriate spontaneous movements of the four extremities. Placed prone, he lifts his head and chest 60°. Held in his mother’s lap, he reaches out for objects with both hands used equally; his grasp is crude and clawed. He will not transfer an object from one hand to the other. His social responses are appropriate; he interacts well with his mother and the examiner, babbles in a dialogue fashion using consonant sounds, and visually tracks people and toys with interest and accuracy. His head circumference is at the 75th percentile, consistent with his length and weight.

Cedric was born after a full-term, uncomplicated pregnancy without any prenatal problems. Family history is negative for developmental delays in young children or neurological disease. Marital stress, financial insecurity, and a recent move highlight the social environment in which Cedric lives.

CASE 3

A pediatric nurse practitioner, working in an urgent care clinic, makes the observation that an 18-month-old male is not walking. A screening birth history reveals a 36-week normal gestation without any perinatal complications. Neurological examination is normal, and a Denver II developmental screening test reveals that other gross motor, fine motor, social, and language milestones are normal. The nurse decides to consult with a developmental pediatrician to determine any further evaluation studies.

Dr. Martin T. Stein

The detection of delay in motor development among infants occurs frequently in primary care practice. This event often raises several important questions:

• With the current knowledge about the wide spectrum of normal motor development, could these children be diagnosed as having a pathological developmental lag?
• Is this an isolated motor delay or are other domains affected?
• Does the delay reflect a neurological disorder? If so, are early diagnosis and referral critical?
• When uncertainty exists, what is the next step and how is uncertainty conveyed to the parents?

The boundary between normal developmental variation and delay may be murky. With more pediatric clinical experience, clinicians develop a greater appreciation for the breadth of the normal developmental spectrum in both directions. This experience often results in a higher sensitivity to the detection of a developmental delay at an early stage, as well as a finely tuned sensitivity to normal variations of delayed development at the other end of the spectrum. Without these acquired skills, an excessive number of inappropriate referrals may be made, or there may be a hesitancy to refer a patient when a variation in development is inappropriately assessed as normal. Neither practice serves the best interest of children or their families. Knowledge about early child development and skill in the assessment and interpretation of developmental evaluation is critical.

Primary care clinicians who care for young children confront uncertainty about the appropriateness for a developmental referral frequently. Each of the three case scenarios represents potential uncertainty over the interpretation of a neurodevelopmental assessment. Two pediatricians who see young children with developmental delays at a different stage in the evaluation process were invited to comment on each case.

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icine. He is an active clinician with responsibilities for evaluation and treatment recommendations for a wide variety of children with developmental disabilities. Dr. Bennett is active in clinical research with an emphasis on neurodevelopmental maturation and delay. He is currently the President of the Society for Developmental and Behavioral Pediatrics.

Dr. Myles Abbott is a Clinical Professor of Pediatrics at the University of California, San Francisco. He is a general pediatric practitioner who has taught pediatrics to students and residents for many years. In addition, he has organized numerous continuing education conferences for general pediatricians in collaboration with the American Academy of Pediatrics.

Dr. Forrest C. Bennett

Individual differences in motor development are common during the first 2 years of life. This is the area of development most readily scrutinized throughout infancy by parents and professionals (motor milestones are usually easier to list than language or social milestones), so the primary health care provider is frequently called on to distinguish the probably normal from the possibly abnormal. The decision whether to provide reassurance and “watchful waiting” or to initiate diagnostic and therapeutic procedures is often difficult.

There are several key concepts to keep in mind as one evaluates the adequacy of an infant’s early motor development: (1) Normal variations are much broader and more typical in gross motor than in fine motor development.1 The single milestone of “walking” may be normally attained from 9 to 18 months whereas such fine motor parameters as “pincer grasp” should be mastered in a much narrower time frame; (2) comparisons to other areas of development (e.g., language, social, cognitive) are critical. Global developmental delay suggests different explanations than specific delay or deviation in motor development alone; (3) prematurity must be taken into account. Evaluation in infancy should include both a full adjustment for gestational age and also special consideration of unique aspects of preterm motor development such as the common phenomenon of “transient dystonia”2; (4) gross motor development is the domain of development least affected by environmental events or psychosocial deprivation. Language and social development are much more vulnerable to deleterious parenting aberrations; and (5) as with all areas of human development, the pace and quality of motor development tends to be familial. Infants whose parents and siblings experienced extremes in motor development (fast or slow) can be anticipated to demonstrate a similar pattern. Exceptions to the family norm should be noted.

The first case, “Juanita,” describes an ostensibly low risk infant (i.e., term gestation, no perinatal complications, normal initial examinations) with a worrisome neurological examination at 3 months. Even though we are not told, I will presume that there are no associated metabolic or general medical symptoms, including seizures or bladder/bowel dysfunction. The most relevant clinical issue is whether this infant is developing the signs of a static encephalopathy, e.g., cerebral palsy. Findings of concern include increased lower extremity muscle tone, ankle clonus, and weight bearing on both legs. However, two of these observations (ankle clonus and weight bearing) can be normally elicited at 3 months. The positive support primitive reflex, i.e., weight bearing, is typically present during the first 2 to 3 months and should be diminishing in prominence at this time. It would be essential to know the status of other motor (head control, reaching, rolling) and nonmotor (visual tracking, smiling, cooing) developmental milestones.

Cerebral palsy is a nonprogressive disorder with a developmental presentation over the first 6 to 18 months of life.3 Its primary mode of clinical presentation, particularly in mild-moderate cases, is a gradual increase in muscle tone (legs more than arms) coincident with an increasing delay in motor development. Deep tendon reflexes may be relatively normal during early infancy, only to accentuate over time. The fact that Juanita is a term infant is more concerning because increases in lower extremity tone are frequently encountered transiently in preterm infants.4 Nevertheless, it is too early in this case to make a definitive diagnosis. Juanita’s subsequent motor development should be monitored closely at approximately 2-month intervals. If the early concerns are confirmed by increasing abnormalities (typically by 8 to 10 months of age), additional diagnostic studies such as neuroimaging may be performed and physical therapy intervention instituted. There is no reliable evidence that beginning physical therapy treatment in early infancy before a likely diagnosis alters ultimate motor outcomes.5

The second case, “Cedric,” also describes a biomedically low risk infant but one who is at increased environmental risk. Cedric presents the common primary care dilemma of how vigorously to pursue relatively mild motor delays. Independent sitting is delayed as is transferring and grasping. However, language and social development appear adequate, making generalized developmental retardation unlikely. Cedric’s mild hypotonia offers a most plausible explanation for his gross motor delays and possibly for his fine motor delays as well, although these are somewhat more troubling. The recorded deep tendon reflexes 3 argue against a lower motor unit disorder but at this young age do not completely exclude Duchenne muscular dystrophy. Although most boys with this condition do not develop characteristic signs and symptoms for several years, many demonstrate mildly delayed gross motor development during infancy.6

The most likely etiology of Cedric’s specific motor delays is the somewhat vague entity usually described as “benign congenital hypotonia.” Infants with this condition are noteworthy for: (1) diffuse hypotonia, (2) delayed motor milestones, (3) appro-
prietie nonmotor milestones, (4) gradual improvement into the average range over time, and (5) the absence of any other definitive diagnosis. However, the relatively accentuated 3+ nature of Cedric's deep tendon reflexes requires the continued consideration of a hypertonic form of cerebral palsy. Again, careful monitoring for the next 12 to 24 months will help to clarify this differential. Diagnostic testing might include formal developmental assessment (e.g., Bayley Scales of Infant Development II) to confirm the adequacy of nonmotor development and a laboratory test (serum creatinine phosphokinase) to rule out Duchenne muscular dystrophy. Treatment may include referral to a physical therapist to assist the parents in encouraging normal motor activities. The family stressors are unlikely to explain Cedric's motor delays.

The third case can certainly be managed by a "watchful waiting" approach. The history relates that this 1-month preterm boy of 18 months is appropriate in all neurodevelopmental aspects but not yet walking independently. The mild prematurity is probably a minor issue because, even with gestational age adjustment, we still have a child who is not walking at 17 months. Although this may be quite acceptable and normal for this boy and family, I would at least consider a laboratory test (serum creatinine phosphokinase) to rule out Duchenne muscular dystrophy. The child should, of course, be monitored over the next 3 to 6 months to ascertain the onset of walking. No interventions are required at this time.

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REFERENCES

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Juanita's lower extremity hypertonicity (Case 1) suggests that she may have an early form of spastic cerebral palsy. If this diagnosis is correct (and only time will tell), then no immediate treatment or intervention is necessary. However, the pediatrician must consider other diagnostic possibilities, which in her case are limited. She may be normal, or she may have an early manifestation of a tethered cord. Early recognition and corrective surgery can halt or prevent progression in a tethered cord, so the clinician must focus first on that insidious but treatable condition.1

The most common neurological presentation of a tethered cord is a motor disturbance. This is what should arouse the pediatrician’s suspicion in Juanita's case. The ankle clonus that she exhibits may be normal or abnormal at 3 months of age; its clinical significance is hard to determine.2 Furthermore, Juanita does not manifest other neurological signs of a tethered cord, such as pain, bowel/bladder dysfunction, or sensory loss. Nonetheless, her hypertonicity is enough to cause concern.

On physical examination, half of children with tethered cords have external signs of lower spinal abnormalities such as a tuft of hair, subcutaneous lipoma, or dermal sinus.3 These signs are often very subtle; the pediatrician should look for them carefully. A rectal exam, including a test for sensation in the skin of the anal area, should be performed to evaluate anal sphincter function of the rectum. If the examination still shows no signs of a tethered cord, some pediatricians might elect to not perform any diagnostic imaging studies at this time but simply monitor Juanita clinically. However, because a tethered cord is a treatable disorder, I would recommend an imaging study of her spine. Although magnetic resonance imaging is the most definitive diagnostic procedure, it is expensive and most likely would require that Juanita be sedated; therefore, I would not recommend it. The incomplete ossification of the spine allows ultrasonography in a 3 month old, so this would be my imaging study of choice if a pediatric radiologist experienced in performing this procedure is available. Ultrasonography is highly accurate for this condition, is relatively inexpensive, and does not require sedation.4,5 I would order this test within a few days of the office visit.

In concert with these efforts to rule out a spinal cord abnormality, the pediatrician must explain to the parents that Juanita's increased tone may be an early sign of cerebral palsy, possibly spastic diplegia. Although she has no risk factors commonly associated with cerebral palsy (e.g., low birth weight, prematurity, low Apgar scores), neither do 50% of the children who have that condition.6 It is too soon, however, to make a definitive diagnosis or prognosis.

Assuming a spinal defect is not found, Juanita should be monitored closely. Physical therapy should be considered. If the family desires, or if her condition remains unchanged or worsens over time, Juanita should be enrolled in an early intervention program and referred to a pediatric developmentalist.

In evaluating the hypotonic child (Case 2), the pediatrician must first determine the anatomical site of the motor problem. The anatomical site can be categorized clinically by the absence or presence of muscle weakness.7 Hypotonicity without weakness indicates a central (i.e., brain) disorder, and hypotonicity with weakness suggests a peripheral neuromuscular condition (i.e., spinal cord, anterior horn cell, peripheral nerve, myoneural junction, or mus-
Cedric’s problem appears to be centrally located because there is no apparent weakness on examination. He spontaneously moves all four extremities, lifts his head and chest 60° in the prone position, and reaches with both hands equally.

The next step is to determine whether Cedric’s hypotonicity is static or progressive. If it is static, he probably has cerebral palsy. If it is progressive, there could be many causes, including a neurodegenerative disorder, infectious process, or metabolic condition. Clues that suggest a progressive disorder include regression of previous milestones, normal development followed by slowdown, or parental consanguinity. Although the case study suggests that Cedric’s condition is static because there is no evidence of motor deterioration or developmental slowdown, the clinician does not have enough history to be sure. The pediatrician should obtain more developmental information from the mother, especially whether Cedric had appreciable weakness in the first 6 months of life and whether he could sit unassisted at 6 months. The pediatrician should also request Cedric’s prior medical records and plan to speak with his previous health care provider(s).

Cedric appears to have a static encephalopathy, probably a hypotonic form of cerebral palsy, with normal intelligence. With this working diagnosis, there is no emergency that requires immediate diagnostic procedures or consultative services. If the pediatrician gets a more detailed history that suggests the condition might be progressive, then laboratory tests would be appropriate and could include a thyroid panel, electrolytes, calcium, phosphorus, and urine for amino aciduria and mucopolysaccharidosis.

The timing for consultative referral to a pediatric neurologist or developmentalist depends on the parents’ and the pediatrician’s anxiety levels. It would be completely appropriate to follow this patient over the next few months without a referral. Cedric has presented with cold, and there is probably not enough time to discuss the implications of your neurodevelopmental findings in detail at this visit. It is not appropriate to label this child as having cerebral palsy or to initiate any form of therapy until the family understands the implications of the child’s hypotonia. The pediatrician should: address the cold and its management; tell the parents that you are concerned about his muscle tone and the family’s problems (marital stress, financial insecurity, recent move); initiate steps to obtain prior medical history; and schedule a longer appointment for a more detailed discussion of Cedric’s neuromuscular problem with both parents in the next 2 to 4 weeks.

At that time, the pediatrician should explain Cedric’s condition, prognosis, and treatment options. The prognosis should be given cautiously because the spectrum of possibilities is so broad: from significant motor and gait disturbances to a 30% chance of normal motor skills by 7 years of age.6,8 The family should be encouraged to enroll in an early intervention program that offers support, education, and treatment.

At first glance it might seem that an 18 month old who does not walk (Case 3) may have a significant neurological and/or developmental problem. However, this child is most assuredly normal. The best way to manage this child is to understand and allay any parental concerns, educate the parents about the normal variations of the onset of walking, and to follow the child with standard care.

Walking is a complex neurodevelopmental activity that occurs at an average age of 12 to 15 months with a 2-SD range of 9-17 months.2,9 Given these data, it is understandable that a practitioner may become concerned when an 18 month old is not yet walking. In fact, delayed walking is a sign of a serious condition when it is accompanied by neurological and developmental deficits. But based on clinical experience and research, it is almost certain that the child presented in this case study is normal.

Researchers at Oxford10 studied 257 children who were not walking by 18 months and who had no previously suspected cause for delayed walking. After these children were entered into the study, a pediatrician performed a complete neurological and developmental assessment. Of the 257 participants, nine children (3.5%) were found to have cerebral palsy, and six children (2.3%) had minor neurological abnormalities. Thus, the total number of children with neurological abnormalities was 15 out of 257, or 5.8%. The other 242 children, or 94.2%, were neurologically normal; half of them were walking at 19.5 months and 97% by 2 years of age.

In the case study, we already know that the child has a normal neurological examination and a normal Denver II developmental screening test. Therefore, parental reassurance for continued normal development, including walking, is justifiable. This family’s specific concerns regarding delayed walking in their child should be thoroughly explored and discussed. They should be informed that children begin walking at different ages because of many factors. For example, some children are timid by nature and fearful of walking. Familial patterns may play a role, as might ethnic and racial variations.

With the child in the case presented, I would schedule a health maintenance visit in 3 months. I also would encourage the parents to let the child try to walk, without undue pressure, in a safe environment. I would strongly discourage walking exercises, infant walkers, or designer shoes because they have no proven benefit and documented drawbacks.

Guided by a familiarity with the medical literature, along with this child’s normal history and neurological and developmental assessment, the prognosis is excellent. Primary care providers do not need to routinely refer such a child to a developmental pediatrician or a pediatric neurologist.

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The three case histories of motor delay in early childhood illustrate important variables in the approach to a diagnosis and management by a generalist and a specialist. Some differences are obvious; others are subtle.

A potential lag in motor milestone, without an apparent deficit in language or social development, was the major finding in each patient. These case histories were chosen to point out the process by which a primary care pediatrician can isolate a delay in one developmental domain with a cautious degree of certainty. As Dr. Bennett observed, motor skills are more assessable than social and language milestones to accurate recording in infancy. However, in each case, a standard pediatric neurological and developmental examination isolated the problem in the motor domain. By this process, a global developmental delay was unlikely in each patient.

General pediatricians are trained to assess early developmental milestones. A comprehensive and complete neurodevelopmental examination is sufficient as a screening instrument in primary care practice. A variety of check lists have been developed to facilitate this process; they are useful for students and residents as they learn to incorporate a developmental inventory into a physical examination. Among the standardized screening instruments, the Denver II developmental screening test is the most widely used. It is valuable in the cases under discussion when a generalist wants to make a more precise assessment of motor, language, and social skills from birth to 6 years of age. The Denver II has the advantage that it is easy to apply to young patients in a practice setting, and it has been adequately tested and fulfills criteria for a screening test of early development. The testing format is not only easy to use, it is effective in providing parents a visual format of their child’s initial assessment and progress. It is a medical record and teaching tool simultaneously.

The Denver II has limitations as a screening instrument. Alternative instruments for the assessment of infant development are available. An interesting question for clinical teachers in developmental-behavioral pediatrics is “Should we teach pediatric residents more precise methods of developmental assessment?” For example, the Gesell, the Amiel Tison, and Bayley scales of infant development could be taught during residency training. The ability to perform these scales in a primary care practice on selected patients would add precision to the assessment of the three patients under discussion. Is a greater degree of precision necessary to make a decision about a presumptive diagnosis and initial management? I do not think so. At the least, and in response to the expanded behavioral-developmental curriculum required under the new Residency Review Curriculum guidelines, all pediatric residents should be exposed to more sophisticated developmental evaluation instruments during training.

The first two cases illustrate the difficulty pediatricians often face when young children are found to have a possible developmental delay. In the first case, a 3 month old is discovered to have three signs on physical examination that may or may not be consistent with cerebral palsy. As pointed out by Dr. Bennett, increased tone, ankle clonus, and weight bearing with the lower extremities may each represent a transient phenomenon of early motor development. In addition, Dr. Abbott raised the possibility that hypertonia limited to the lower extremities may not only reflect a spastic diplegic form of cerebral palsy but also may be the result of a tethered spinal cord secondary to a lesion in the spine. In the second case, the 10 month old with moderate hypotonia associated with a delay in independent sitting, grasping, and transferring, is consistent with benign congenital hypotonia but may also be consistent with an early diagnosis of hypotonic cerebral palsy. Furthermore, Dr. Bennett points out that the characteristics of both the hypotonic infant and the 18 month old who is not walking may be consistent with Duchenne muscular dystrophy at an early stage.

These dilemmas are reflected in the clinicians’ uncertainty about the correct diagnosis and need for referral to a specialist at this moment. Dr. Abbott’s candor reflects the uncertainty principle in these situations. In the first case, he takes the position that a pediatrician must explain to the parents that the physical findings may reflect early signs of cerebral palsy, and then he concludes, “It is too soon...to make the definitive diagnosis or prognosis.” An alternative approach may be to withhold the equivocal physical findings from the parents because they may be transient and insignificant and to plan periodic re-evaluations. This approach is consistent with the knowledge that hypertonicity, ankle clonus, and weight bearing may be normal at 3 months and that a diagnosis of cerebral palsy in early infancy will not alter the ultimate motor outcome through early intervention with physical therapy.

Equivocal signs of development can be listed in the problem list of the medical record to insure that they will be assessed at the next visit. In a practice in which other clinicians may see the patient for an intermittent illness before the next scheduled examination, I have found it useful to write the letters “NM” (not mentioned) next to the problem in the medical record. This alerts a colleague that the findings have not been discussed with the parents pending further evaluation.

These cases raise the thorny issue about the bene-
fits and drawbacks that may result from informing parents about a “possible” delay in motor development. For some parents, it is difficult to disassociate the term “motor delay” from “cerebral palsy.” When the neurodevelopmental examination suggests mild, but equivocal, motor delay or cerebral palsy, most parents will be alarmed. Some will respond with equanimity to a careful discussion of the findings and their meaning; most will respond with anxiety, disappointment, or anger, all appropriate responses to hearing the “bad news.”

Many clinicians take the point of view that it is consistent with medical ethical standards to “tell the truth” even when the truth is uncertain. With appropriate planned follow-up visits, they point out that these cases of possible motor delay will sort out between a pathological diagnosis or a normal variant. Other clinicians find that learning to live with a medical uncertainty is an important developmental stage for physicians. It is a process in the professional development of physicians that can be learned through clinical experience and knowledge of diagnostic strategies and clinical decision making.4

In the context of the three children with variants of isolated motor delays, uncertainty that leads to a “watchful waiting” approach is a reasonable pathway. This decision can be made with a recognition that, although a tethered cord discussed by Dr. Abbott is possible in the 3 month old with hypertonic lower extremities, it is a very low probability in the absence of abnormal reflexes, weakness, or a cutaneous lesion of the lower spine. The decision to withhold a diagnostic study or referral can be based on the low probability of a tethered cord and the uncertainty of cerebral palsy. In contrast, Dr. Bennett noted that, although the 10 month old with hypotonia and the 18 month old who was not walking have a low probability of Duchenne muscular dystrophy, an early diagnosis may have a significant impact through genetic counseling about management options for a subsequent pregnancy.

In my experience as a general pediatrician, diagnostic uncertainties about a developmental problem occur frequently. Paradoxically, as sensitivity increases for the subtleties and normal variations, the opportunity for uncertainty does not go away. The British pediatrician, John Apley, wrote that, “in diagnosis the doctor should be ambitious.”5 He also observed the value of viewing a diagnosis as a working hypothesis that can be modified in response to progress or treatment. The three infants with motor delay are examples of coupling scientific knowledge about the motor component of neurodevelopment with cautious “ambition in diagnosis” and respect for developmental variation. Incidentally, all three children had normal developmental and neurological examinations at 2 years of age.

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