CHALLENGING CASE: DEVELOPMENTAL DELAYS AND REGRESSIONS

“By the Way, . . . Her Foot Is Turned Out.”*

CASE

At each health supervision visit in the first year of life, Jenny came across as a delightful, interactive, and playful infant. She achieved all developmental milestones at the expected times. Her social and language skills were consistently early.

At the 12-month office visit, Jenny’s mother, a school teacher who enjoyed every moment with her daughter, commented to her pediatrician at the end of the visit, “I know she’s healthy, but one thing concerns me. She seems to turn out her left foot all the time. Now that she is walking, it has become more noticeable.” Although a physical examination had been performed without any abnormal findings, the pediatrician reexamined Jenny. With weight bearing, both feet were everted, and the left turned out more than the right. She walked with a steady, symmetrical gait, although ambulation exaggerated the externally rotated, everted left foot. When she was examined in the supine position, the feet could be maneuvered passively to a neutral position. Lower extremity tone was normal, deep-tendon reflexes were 2+ and symmetrical, and ankle clonus was absent. Jenny’s mother was reassured that her observation did not indicate a problem. Jenny’s overall normal developmental achievements were reemphasized. The pediatrician documented the mother’s concern in the medical record to assure follow-up at the next visit.

Jenny was not seen again until age 15 months, her next health supervision visit. When the pediatrician asked her mother if she was still concerned about the positioning of Jenny’s feet, she responded that it seemed unchanged and continued to cause concern each time she observed Jenny walking.

The pediatrician examined Jenny with a precise focus on her lower extremities. When she walked, the everted, external rotation of her feet was unchanged (Figure 1). The atypical position was more prominent on the left foot. When supine, her outstretched legs revealed that the left leg was 1.0-cm shorter than the right leg. Measurement of midthigh and midcalf circumferences demonstrated detectable differences. The left thigh circumference was 1.5 cm smaller than the right thigh; the left calf was 0.75 cm smaller than the right side. A surprised pediatrician (the legs had looked similar in size!) repeated the measurement with the same results. Deep-tendon reflexes remained 2+ and symmetrical; ankle clonus was absent. Held upright, there was no evidence of scissoring of the lower extremities. Upper extremity tone, strength, and reflexes were normal. A bilateral precise pincer grasp was demonstrated. Parachute reflex was present and symmetrical. Head circumference was at the 75th percentile, similar to length and weight.

Index terms: leg-length discrepancy, medical communication, syringomyelia.

Dr. Martin T. Stein

Specific concerns of a parent during a health supervision visit may be apparent after the standard question, “Do you have any concerns about ___?” However, the response is not always predictable. That approximately one third of parents have an unspoken psychosocial concern when they bring their children to a pediatrician for an acute illness is an important guide.¹ For pediatric office visits that emphasize health prevention and anticipatory guidance, there are several methods available to discuss the parents’ main agenda. The use of previsit questionnaires with focused and open-ended questions,²


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conducting the clinical interview in a manner that encourages a parent to tell his or her story, and “active listening” with a practiced ability to repeat important phrases for emphasis and clarification are some of the methods that pediatricians use to discover a parent’s agenda. During a health-supervision visit, a focus on the most significant developmental or behavioral theme at a particular age can achieve this goal.

In this challenging case, the clinician’s initial assessment was a healthy 1-year-old child with normal developmental milestones. As the visit is about to end, the child’s mother asks an “out-the-door” question. With her hand on the doorknob of the examining room, the pediatrician becomes aware of a new problem. Her response reflects active listening and the wisdom to recognize a potential orthopedic or neurological problem.

The commentaries are from the perspective of a pediatric orthopedic surgeon and a pediatric neurologist. Dr. Peter Newton, a pediatric orthopedic surgeon, is an assistant clinical professor of Surgery at the University of California, San Diego. Dr. Majeed Al-Mateen is a pediatric neurologist and clinical assistant professor of Neurology at the University of Washington in Tacoma, Washington. Dr. Robert Pantell is professor of Pediatrics and director of Ambulatory Pediatrics at the University of California, San Francisco.

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A 15-month-old child is observed by her pediatrician to have asymmetry in length and circumference of her lower extremities, as well as eversion of the feet. Motor development has been normal, and there was an absence of abnormality in the neurological examination.

The eversion of the feet seems moderate by report. It is supple; as such, it should not generate substantial concern in the presence of a normal neurodevelopmental status. Flexible flat feet generally improve to some degree until age 10 years. A family history of similar deformity and/or generalized ligamentous laxity are often noted in these patients.

The differential diagnosis of a leg-length discrepancy broadly encompasses some of these conditions:

- Abnormalities of the neurological system at the level of either the brain or the spinal cord.
- Skeletal underdevelopment with a hypoplastic limb.
- An overgrowth syndrome with enlargement of a limb.

The neurological causes of leg-length discrepancy or underdevelopment of a limb may be related to a hemiparetic form of cerebral palsy. In addition, unilateral spinal cord abnormalities, either congenital or as a result of a tumor, may result in underdevelopment of a limb. With either of these conditions, it is likely that some neurological examination abnormality would be observed; in the case of a spastic hemiplegic patient, asymmetry in gait may be apparent. Even in the most subtle cases, posturing of the involved arm in a slightly abducted position during running may be seen. In addition, there may be an asymmetry of muscle tone and deep-tendon reflexes. A suspected anomaly of the spinal cord should prompt physical examination of the low back (skin dimple, hairy patch, or other cutaneous lesions overlying the spine), as well as consideration of radiographs of the lumbar spine. These steps may be undertaken to rule out any congenital anomalies of the vertebral bodies that might be associated with an underlying abnormality of the spinal cord.

Limb hypoplasia may come in several varieties. An idiopathic form exists in minor degrees in many patients in whom the cause is unclear. Other more substantial forms of limb underdevelopment consist of femoral hypoplasia, which in its most severe form is known as proximal femoral focal deficiency and results in marked shortening of the limb. Lower-segment hypoplasia may exist in the form of tibial or fibular hemimelia. Fibular underdevelopment may be associated with lateral ray deficiencies of the feet; in the mildest form, ankle valgus or an everted foot position may exist. Underdevelopment of the tibia results in the opposite deformity, with the foot in a clubfoot position.

Hemihypertrophy or increased limb dimension is also possible. It is difficult at times to know whether the larger of the limbs is the normal or abnormal side. Limb overgrowth may be associated with neurofibromatosis, arterial venous malformations, hemangiomas, lymphangiomas, and other overgrowth syndromes. Idiopathic hemihypertrophy can be associated with the development of Wilms tumor.

With this child, I am interested in the details of the examination of the spine. In addition, examination of the feet and tibia will be important to determine any asymmetry of the tibial torsion and/or foot deformity. A single anterior-posterior radiograph of the legs (hips to ankles) is a useful screening examination to assess the bony dimensions and to evaluate potential osseous malformations.

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It has now been more than 30 years since Barbara Korsch and colleagues first documented the importance of good communication between parents and physicians.1 Addressing a major concern of a parent is essential, whether it occurs as the chief complaint or on the way out the door, as occurred in this case and has in so many others. There is a myriad of reasons to do so. First of all, parents are remarkably accurate in suspecting underlying biological conditions. For decades, the best indicator of hearing loss in an infant has been parent report. In addition, parents have been shown to be remarkably sensitive in suspecting serious underlying bacterial illnesses, as documented in work by Paul McCarthy, although not always as specific as experienced clinicians.2 Sec-

Dr. Majeed Al-Mateen

“Where is the lesion?” A cerebral lesion is unlikely in this girl, who has achieved all developmental milestones, including acquisition of language and social skills at the expected times. The absence of upper motor neuron signs such as spasticity, hyper-reflexia, ankle clonus, and extensor plantar response (Babinski sign) also makes a cerebral lesion unlikely. Brain lesions that cause hemiparesis may begin as monoparesis. A right hemispheric lesion such as a brain malformation or congenital stroke is not likely given her symmetrical parachute reflex. Also, there was no scissoring, which is a feature of the spastic diplegic type of cerebral palsy. A thoracic spinal cord lesion causing paraplegia may also present as monoparesis. This is not likely in this case, given the absence of upper motor neuron signs, sensory disturbance, or sphincter dysfunction.

Because there are no upper motor neuron signs to implicate a central nervous system (brain or spinal cord) lesion, a disorder involving the motor unit (motor neuron, peripheral nerve, neuromuscular junction, and muscle) should be considered. However, there is no significant weakness, hypotonia, or decreased deep-tendon reflexes to implicate a motor unit disorder. The only evidence of a primary lower motor neuron lesion is the possibility that the diminished size of the left lower extremity is attributable to atrophy rather than to hypoplasia. Poliomyelitis was at one time the most frequent cause of atrophy of a single limb; as the name implies, it was the result of inflammation of the gray matter (motor neuron) of the spinal cord. Other enteroviruses may yield a similar clinical picture.1 Spinal muscular atrophy, presenting as weakness and atrophy of one limb, has also been reported but is unusual.2 Focal atrophy may also follow trauma and immobilization in children.3 Electromyography would demonstrate signs of denervation in all of these diseases of the motor neuron. Chronic disorders of the peripheral nerves, neuromuscular junction, or muscle would not be expected to cause such an asymmetry in limb size.

There are no unequivocal features of a central nervous system or motor unit disorder. Could a disorder affecting both levels of the nervous system explain the clinical picture? A lesion that involves a combination of the spinal cord and proximal nerve roots may cause monoparesis. Such insults are usually acute, however, and include inflammation of the lumbosacral plexus, traumatic injury, or mass lesions. Lower limb monoparesis caused by herpes zoster involving the lumbosacral dermatomes has been reported in adults.4,5 I have evaluated a 2-year-old boy with nonprogressive atrophy of the lower limb after an ipsilateral herpetic dermatomal cutaneous eruption in infancy; his mother had chickenpox in the second trimester. This is an example of reactivation of a latent varicella-zoster virus infection after intrauterine exposure to the virus.6 This reactivation most likely resulted in lumbosacral segmental radiculitis, which led to atrophy of the lower limb. In addition, a congenital malformation of the caudal portion of the spinal cord and nerve roots should also be considered when a leg size discrepancy seems nonprogressive.

In tethered spinal cord syndrome, as the child grows, the tether causes the cord to stretch and the lumbosacral segments to become ischemic; neurological dysfunction follows.7 The progression of symptoms and signs is insidious; at first, most children are believed to have a static problem. Children may have abnormalities of the feet (e.g., pes cavus and pes equinovarus) and stunted growth of an entire leg. The other leg may seem normal or have a milder deformity without growth disturbance. A magnetic resonance imaging study is the appropriate diagnostic test. The essential feature of a tethered spinal cord is a low lying conus medullaris. Surgical relief of tethering prevents further deterioration of neurological function. In the absence of a characteristic foot deformity and a normal neurological examination, it is recommended that this girl with flexible pes planus and leg-length (and circumference) discrepancy receive serial neurological examinations.

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second, parents come to our offices to learn about whether their child is growing and developing normally. It is almost impossible to reach the age of parenthood without having a sense of developmental variability. It is just as improbable that a parent will not, at some time, have concern about their own progeny. In fact, a number of studies have documented that developmental and behavioral concerns are raised in 25% to 50% of child health-supervision visits. Unfortunately, some studies indicate that pediatricians don’t always respond as thoughtfully as in this case study. Concerns are too often brushed aside in the hectic pace of premillennial pediatrics. The typical response is to watch and wait. Even when parents raise serious behavioral issues, some studies indicate that active interventions often do not occur. It is important to acknowledge that many problems need more time the next day, others require referral to counselors better able to receive reimbursement for their time, and some will do well with watchful waiting.

Although many problems resolve during periods of watchful waiting, many do not. However, the communication strategy during periods of watchful waiting is to let parents know you are available should there be any change in function.

In addition to being the principal tool for detecting deviations in development and behavior, effective communication has also been shown to be more than just an amenity. Although the relationship with patient satisfaction has been documented numerous times, the outcomes of effective communication include improved knowledge about managing childhood illness and, most importantly, improved functional status in children and adults. The best reason for spending that extra moment with parents before they leave the office does not lie in the increased likelihood of detecting a significant physical condition (although it is important to do so!). Eudora Welty, a Southern writer and photographer, reminds us that “learning is not a continuum, it is a pulse.” When parents ask a question, it is on a topic that is meaningful and salient to them. These are the moments in which you are most likely to have an impact and the moments we must continue to find in a busy day.

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Jenny’s visit to her pediatrician, and the subsequent observation of distinct abnormalities upon examination of her left leg, provide an opportunity to discuss three additional areas of interest:

- Active listening as a method to uncover a parent’s main concern.
- Reevaluation of an initial diagnosis.
- A neurodevelopmental anomaly that mimics a normal musculoskeletal variation.

The pediatrician’s initial physical examination at 12 months of age was considered normal. Even after Jenny’s mother expressed her concern about an exaggerated turning-out of the left foot, the pediatrician concluded, after a second examination, that she had a normal variation, an exaggerated everted foot. Although this conclusion seemed reasonable, the pediatrician wanted to acknowledge the mother’s concern by making a notation in the medical record as a reminder to discuss the concern at the next health-supervision visit. Dr. Pantell reminded us that a parent’s concern about a biological condition is a sensitive observation. A new screening test for developmental and behavioral concerns of parents supports the same concept.1

At the subsequent examination, the foot eversion was more pronounced, prompting the pediatrician to repeat a careful neurodevelopmental assessment. It was at that time that mild but reproducible leg-length and circumference discrepancies were detected. Normal upper extremity tone, strength, and deep-tendon reflexes in association with an intact parachute reflex suggested a monoparesis of the left lower extremity. Dr. Al-Mateen guided us through the differential diagnosis of a monoparesis.

To her credit and clinical acumen, Jenny’s pediatrician kept an open mind about the diagnosis. She was not constrained by her initial diagnostic hypothesis that the foot eversion reflected a normal developmental variation. A reassessment of physical findings led to an alternative diagnosis.

The pediatrician was concerned that Jenny might have a central lesion presenting slowly with a hemiparesis, initially involving the lower extremity. Alternatively, a spinal cord lesion might cause a monoparesis. She reexamined the patient and significantly altered the diagnostic hypothesis on the basis of new findings.

A pediatric neurologist was consulted, the physical findings were confirmed, and the possibility of a tethered cord was raised. A tethered cord refers to an abnormally low position of the conus medullaris (distal portion of the cord) associated with a short and thickened filum terminalis. Because a tethered cord might be associated with a spinal cord lesion, a magnetic resonance imaging study was performed.
A syrinx defect of the spinal cord, extending from T10 to the tip of the conus medullaris, was discovered (Fig. 2). The conus was in its normal position, but the filum terminalis seemed to be stretched. There was no evidence for tumor. The cervical and upper thoracic cord was normal, and there were no abnormalities of the brainstem or cerebellum.

A syrinx is a dilation of the central canal lined with altered glial cells. It may be localized, as in this patient, or may involve many segments of the spinal cord. The presence of a syrinx, a condition termed “syringomyelia,” may be associated with intramedullary tumors, spinal cord trauma, arterial insufficiency, and developmental anomalies of the cord.2 The latter condition seemed to be the explanation for Jenny’s syrinx. A pediatric neurosurgeon was consulted, and Jenny underwent a T11–T12–L1 laminoplasty with release of the tethered cord and placement of a catheter from the distal portion of the syrinx to the subarachnoid space.

Ten years after the diagnosis and neurosurgical procedure, Jenny is doing very well in the fifth grade, playing softball, riding a bike, and is free of any gait, bladder, or bowel symptoms.

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Pediatrics 2001;107;930

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