



EMG Case No. 56, October 2002

Presenting Symptoms: 2 year old with walking difficulty and lower extremity weakness

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Appropriate Audience: Residents and practicing physicians

Learning Objectives: After completing this educational activity, participant will be able to: 1) identify symptoms and signs pertaining gait difficulties and lower extremity weakness in a child, 2) utilize the patient's history and physical to devise an adequate differential diagnosis of lower extremity weakness in a child, and 3) perform appropriate nerve conduction studies and needle examination to evaluate for lower extremity weakness in a child.

History

A 2 year, 7 month old girl presented with a history of walking difficulty. She was the product of a normal pregnancy, and began walking at 16 months. After a few months, she was noted to have right foot inversion and she was falling frequently. She was also walking on her toes. The only treatment she had received had been stretching exercises performed by the family. The family's impression was that the right foot inversion had become worse over the past few months, but they were not absolutely sure.

- *Prior to continuing, please develop a differential diagnosis and list each possible diagnosis in order of likelihood.*
- *Is there any additional information regarding the clinical history that might be helpful in clarifying your differential list or changing its order of priority?*

Commentary I

At this point the differential diagnosis includes:

- **Brain injury/lesion**
 - Cerebral palsy
 - Hydrocephalus
 - Brain tumor
 - Stroke
- **Spinal cord process**
 - Tethering of the cord
 - Diastematomyelia
 - Spina bifida/Myelomeningocele
 - Tumor (Neuroblastoma)
 - Parasitic infection of the cord



- **Motor neuron disease**
 - Type II SMA

- **Plexus injury**
 - Idiopathic plexopathy
 - Tumor (Neuroblastoma)
 - Retroperitoneal bleed

- **Peripheral nerve**
 - Toxic neuropathy
 - Chronic inflammatory neuropathy
 - Sciatic neuropathy (bilateral)
 - Multiple mononeuropathies (bilateral peroneal, tibial)

- **Muscle**
 - Dystrophy

The history suggests an asymmetric process to some degree, but the toe walking suggests that symmetric processes should be considered, at this point. While many cerebral processes lead to more global deficits, this is certainly not always the case. Many cases of cerebral palsy are diagnosed after one year of age, particularly if they are relatively mild. The hallmark of cerebral palsy, however, is that it does not get worse with time, except due to increasing muscle tightness resulting from growth spurts. On the other hand, sometimes symptoms are simply not noticed until the child becomes more active and mobile. Abscess of the brain or spinal cord is not included due to lack of fever or other expected symptoms. Strokes are rare in children, and generally would be accompanied by a more acute change in function. Spina bifida is often associated with tethering of the cord and other neurologic abnormalities. It frequently presents with neurocutaneous stigmata in the low lumbar or sacral area of the back, such as a tuft of hair, a deep dimple, or a lipomatous swelling. There are other syndromes of cord tethering such as diastematomyelia that can present as the child grows. These syndromes will cause progressive weakness, generally accompanied by spasticity and bowel and bladder signs. World travelers may be at risk for schistosomiasis of the spinal cord. Spinal muscular atrophy Type II presents at 3-24 months, and is autosomal recessive. A tumor causing compression in the spinal cord or the plexus most likely represents a neuroblastoma, which originates in any site which contains sympathetic nervous tissue (e.g., the adrenal glands). The onset of symptoms does not follow the usual course of a congenital neuropathy, but toxic exposure or a demyelinating neuropathy (with a fairly chronic course) could be considered. Multiple mononeuropathies, such as bilateral sciatic neuropathies or bilateral peroneal/tibial neuropathies could suggest mononeuritis multiplex, but this would be an extremely unusual presentation. The symptoms are not classic for any particular myopathy, but this must be considered as well.

History, continued

Her review of systems was significant for constipation. Other developmental milestones, such as fine motor function and language, were achieved appropriately.

Her medical history was notable for two episodes of UTI. There is no history of trauma, surgery, or dermal abnormalities on her back.



Family history was negative. Social history revealed that she lived with her parents, 1 year old sister, and dog. Both parents smoke in the home. There was no history of travel or exposure to other toxins.

- *If necessary, revise your differential diagnosis based on the additional clinical history.*
- *On which details of the physical examination should you focus at this point?*

Commentary II

- **Brain injury/lesion**
 - Cerebral palsy
 - Hydrocephalus
 - Brain Tumor
 - Stroke
- **Spinal cord process**
 - Tethering of the cord
 - Diastematomyelia
 - Spina bifida/Myelomeningocele
 - Tumor (neuroblastoma)
- **Motor Neuron Disease**
 - Type II SMA
- **Plexus Injury**
 - Idiopathic plexopathy
 - Tumor (Neuroblastoma)
 - Retroperitoneal bleed
- **Peripheral Nerve**
 - Chronic inflammatory neuropathy
 - Sciatic neuropathy (bilateral)
 - Multiple mononeuropathies (bilateral peroneal, tibial)
- **Muscle**
 - Dystrophy

The lack of travel and the absence of any history of toxic exposure make the parasitic infection and toxic neuropathy unlikely (although not impossible! You never know with a history). The history of UTIs and constipation suggests a process that would affect the bladder and the bowel, pointing toward the spinal cord or possibly the plexus. With the bladder/bowel history and a history of otherwise normal development, a process in the brain or a myopathy seems much less likely. A congenital process such as spina bifida may still be possible, even with the other normal milestones, since a mildly involved child could have good language function and upper extremity function with involvement of the lower extremities, bowel, and bladder. Again, the pattern of onset (symptoms present from birth



versus later onset) was not clear. However, the suggestion that it had become somewhat worse points more toward a progressive process, such as a tethered cord or a tumor.

Physical Examination

On examination, she has bilateral equinovarus positioning, right more than left. Range of motion was full, but extra force was required to bring the right foot into eversion compared to the left. There was no evidence of spasticity. She demonstrated active movement in all muscle groups except eversion and dorsiflexion on the right. Right foot inversion also appeared weak. She appeared to have decreased sensation of the right foot. All lower extremity reflexes were decreased.

- *At this point, review your differential diagnosis and revise as appropriate.*
- *Are there additional observations on physical examination that might be helpful in narrowing your differential list?*

Commentary III

At this point, the most likely differential includes:

- **Spinal cord process-roots and cauda equina**
 - Tethering of the cord
 - Diastematomyelia
 - Spina bifida/Myelomeningocele
 - Tumor (Neuroblastoma)
- **Plexus injury**
 - Idiopathic plexopathy
 - Tumor (Neuroblastoma)
 - Retroperitoneal bleed
- **Peripheral Nerve**
 - Sciatic neuropathy (bilateral)
 - Multiple mononeuropathies (bilateral peroneal, tibial)

The examination clearly demonstrated the asymmetry, so symmetric processes such as neuropathy, SMA, and myopathy seem much less likely. The absence of spasticity and the decreased reflexes make a central nervous system process less likely as well. Physical findings such as the presence of atrophy or deformity could help differentiate between a chronic or more acute problem. In addition, the back should be checked for neurocutaneous stigmata, which would help determine if a congenital spinal problem was present.

Physical Examination, continued

Atrophy of the distal right lower extremity was noted. Her gait demonstrated supination of both feet, but much more so on the right. She bore weight on the lateral surface of the foot, with calluses on the lateral right foot. In fact, she was also developing a callus on the dorsum of her right foot due to severe supination. She had foot drop and steppage gait with hyperextension of her knee. Examination of the back did not reveal neurocutaneous stigmata.



- *If necessary, revise your differential diagnosis based on the additional physical findings.*
- *Design your approach to the electrophysiologic examination based on the existing data.*

Commentary IV

- **Spinal cord process-roots and cauda equina**
 - Tethering of the cord
 - Diastematomyelia
 - Spina bifida/Myelomeningocele
 - Tumor (Neuroblastoma)
- **Plexus Injury**
 - Idiopathic plexopathy
 - Tumor (Neuroblastoma)
 - Retroperitoneal bleed
- **Peripheral nerve**
 - Sciatic neuropathy (bilateral)
 - Multiple mononeuropathies (bilateral peroneal, tibial)

The presence of atrophy and of calluses suggests a chronic process. The steppage gait also suggests that she has had some time to compensate for her weakness. The lack of neurocutaneous stigmata does not completely rule out a chronic spinal process.

Please note as we approach the electrophysiological study that such considerations as SMA and myopathy should not be totally eliminated from differential due to the asymmetry. They are just much less likely.

Electrophysiologic Data

MOTOR NERVE CONDUCTION STUDIES							
NERVE	SIDE	STIM SITE	RECORD	Cm	AMPL	LAT	CV
Peroneal Motor	R	Ankle	Foot	7.0	1.0	3.5	-
Peroneal Motor	R	Below Knee	Foot	110	0.1	6.7	34.4
Tibial Motor	R	Ankle	Foot	40	3.1	2.6	-



Peroneal Motor	L	Ankle	Foot	5.8	1.5	3.9	-
Peroneal Motor	L	Below Knee	Foot	110	1.3	7.0	35.5

SENSORY NERVE CONDUCTION STUDIES

NERVE	SIDE	STIM SITE	RECORD	Cm	AMPL	LAT	CV
Sural Sensory	R	Calf	Ankle	6.5	14.4	1.8	-
Temperture	R	Calf	31.2 degrees	-	-	-	-

NEEDLE ELECTROMYOGRAPHY

INSERtional activity: N, sust, unsust
 FIB: 0, 1+, 2+, 3+, 4+
 OTHer: 0 or fascic, myotonia, myokymia
 EFFort: N, decr
 RECruitment: N, inc or dec 1+, 2+, 3+, 4+
 AMPlitude: N, inc or dec 1+, 2+, 3+, 4+
 DURation: N, inc or dec 1+, 2+, 3+, 4+
 POLyphasia: N, inc or dec 1+, 2+, 3+, 4+

R/L	MUSCLE	INSER	FIB	OTH	EFF	REC	AMP	DUR	POL
R	Anterior Tibialis	N	0	0	N	Dec 2+	Inc 3+	Inc 2+	Inc 1+
R	Medial Gastrocnemius	N	0	0	N	Dec 2+	Inc 3+	Inc 2+	Inc 1+
R	Vastus Medialis	N	0	0	N	N	N	N	N
R	Internal Hamstring	N	0	0	N	Dec 2+	Inc 2+	Inc 1+	N
R	Gluteus	N	0	0	N	N	N	N	N



	Medius								
R	Lumbosacral Paraspinals	N	0	0	N	-	-	-	-
L	Anterior Tibialis	N	0	0	N	Dec 2+	Inc 2+	Inc 1+	N
L	Medial Gastrocnemius	N	0	0	N	Dec 1+	Inc 1+	Inc 1+	N

- *On the basis of both the clinical and electrophysiologic evaluations, formulate your diagnostic impression. List the most likely diagnosis first and follow in order with the other possibilities that are not excluded by the data. Eliminate those diagnoses not supported by the data.*
- *Are there additional electrophysiologic data that you feel would further delineate the diagnosis? (Remember, collecting data that are not needed for the diagnosis is costly and uncomfortable for the patient.)*

First of all, it is clear from the electrophysiological study that the patient has an "old" (not more than 2.5 years, of course!) history of denervation followed by reinnervation to both lower extremities. There is no evidence of abnormal spontaneous or insertional activity, which would indicate active denervation. The peroneal nerve is more affected than the tibial nerve based on nerve conduction studies, but the needle examination indicates that the problem is more wide spread than just the peroneal nerve. There is proximal involvement, based on the findings in the internal hamstring. The abnormalities are in the L5-S1 distribution, except that the gluteus medius is not involved. Paraspinal musculature was normal.

Even though the abnormalities were confined to the distribution of both sciatic nerves, neuropathies in that distribution would be extremely unusual without a history of trauma. Plexopathy is a much more likely consideration in the absence of paraspinal involvement. The normal study of the gluteus medius is not consistent with a plexopathy, however, especially since the peroneal musculature was preferentially involved.

A cauda equina lesion is also a possibility, but paraspinal involvement would be expected. Mononeuropathy of the peroneal or tibial nerve is ruled out by involvement of the proximal musculature.

Some of our other initial differential diagnostic options are definitely ruled out by the electrophysiological examination. SMA would typically present with ongoing denervation. Myopathy would have demonstrated myopathic motor units. A cerebral process would not demonstrate lower motor neuron findings such as motor unit morphology changes.



- **Make the final revisions of your diagnostic impression(s).**

Diagnostic Impression

Bilateral lumbrosacral plexopathy, right more affected than left. There is evidence of old (at least several months) reinnervation, but no evidence of ongoing denervation. As noted above, etiologies include idiopathic plexopathy, tumor, or retroperitoneal bleed.

- *What other diagnostic procedures (laboratory tests, etc.), if any, are needed?*
- *What treatment would you recommend?*

An MRI of the spine and pelvis was ordered, partially due to the results of the electrophysiological study. It revealed a large right paraspinal mass, extending from T10 to L4-5, with some displacement of the right kidney. The mass extended into the T12-L1 neural foramina. There was no frank compression of the thecal sac. A biopsy and staging work up was performed, which led to a diagnosis of Neuroblastoma, Grade III with Intermediate risk. The patient was started on chemotherapy, and continues to receive treatment.

Commentary V

Neuroblastoma is diagnosed in about 500 children per year in the United States. Two thirds of these children are under five years old. The tumor starts in any place where there is sympathetic nerve tissue, including the neck, chest, abdomen, or pelvis. The most common site is the adrenal gland.

The tumor can present in many ways, including neurological symptoms as seen in our patient. Aside from compression of the plexus in the retroperitoneal space, there can be spinal cord compromise due to invasion of the cord via the neuroforamina. Abdominal neuroblastoma can present with abdominal distention and respiratory compromise. Retrobulbar metastases can present with proptosis or periorbital ecchymosis. Some children present with paraneoplastic findings, such as cerebellar ataxia or myoclonus.

Neuroblastoma is staged based on location (one sided versus crossing the midline) and dissemination. It is also graded as high, intermediate, or low risk based on age of the patient, stage, and histology. Common sites of metastases include lymph nodes, bone, marrow, liver, and skin.

A review of the anatomy suggests that the tumor was primarily compressing the lumbrosacral plexus, including the L4-5 lumbar trunk and the sacral branches. The compression is more prominent on the right, but the left is affected as well due to compression or possibly due to a traction injury due to some displacement from the tumor. It is difficult to explain the lack of involvement of the gluteus medius. This is particularly true considering the peroneal innervated musculature was more affected than the tibial innervated musculature. Both the peroneal nerve and the gluteal innervation stem from the posterior divisions of the roots as they form the plexus, suggesting that they would both be abnormal. This remains unexplained.

There are several other muscles that could have been tested that would have helped formulate a more definitive diagnostic impression. Other proximal muscles of the right lower extremity, and several more muscles on the left side would have been helpful. However, with a patient who is 2 years old, it is always important to remember that the examiner must carefully decide which muscles must be tested to gain the most information with the fewest needle insertions. On the other hand, it is important to try to continue the examination until a diagnostic impression can be obtained with some degree of clarity. This case is a



good example of this concept, as her neuromuscular problem was the presenting symptom of a much larger problem. It would have been crueler to this child to leave her situation unclear, and possibly delay the proper diagnostic evaluation, because of a fear of doing a few more needle sticks.

Bibliography

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