



March 1998 EMG Case-of-the-Month

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HISTORY

The patient is a 5-day-old infant girl referred for evaluation of severe hypotonia, multiple joint contractures, and poor feeding. She is also referred to determine if she has a right brachial plexus palsy. At birth she weighed 3,414 grams and was 52 centimeters in length, the product of a 27-year-old woman, gravida 3, ectopic 1, abortion 1. During the pregnancy, fetal activity was diminished. The pregnancy was complicated by spotting at three months' gestation, streptococcal pharyngitis at 4 months and, increased contractions at 6-7 months for which ibuprofen was prescribed.

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

COMMENTARY I

This infant can be categorized into the so-called "floppy baby syndrome." The almost arthrogryptic features suggest intrauterine immobility. The most common reason for a floppy infant is usually cerebral in origin, which may or may not be associated with dysgenetic syndromes. The next most common category includes the peripheral neuromuscular disorders. Of these, spinal muscular atrophy (Werdnig Hoffman) is the most common. The second most common cause is myotonic dystrophy. Other etiologies to be considered include the congenital dystrophies, metabolic myopathies (the glycogen-storage disorders, lipid, and mitochondrial myopathies), as well as the very rare neuropathies which may present as a floppy infant.

Family history may be useful in elucidating the diagnostic possibilities.

HISTORY, continued

The mother has a long thin facies, with a transverse smile. She has slight weakness of her neck flexors, but her limb strength is normal. She has no clinical myotonia and no myotonic discharges on an essentially normal electromyographic examination. Her subsequent muscle biopsy was normal. The father has no known neurological abnormalities or dysmorphic features. There are no other obviously affected individuals in the immediate families by history, examination, and photographs.

- **If necessary, revise your differential diagnosis based on the additional clinical history.**



- **On what details of the physical examination do you think you should focus at this point?**

COMMENTARY II

The physical examination of the infant is of utmost importance since she remains the only clue to her diagnosis at this time. She must be examined for 1) her state of mental alertness, 2) ocular motility, 3) facial configuration, 4) bony abnormalities to include dislocating hips and scoliosis, 5) texture of the muscles on palpation, and 6) presence or absence of muscle stretch reflexes.

PHYSICAL EXAMINATION

The infant appears visually bright with good ocular movement. Severe hypotonia is observable with some asymmetry in that she does not move the right upper limb as much as the left. She has a long facies, a tented lip, and a high-arched, narrow palate. Her limbs are thin, and on palpation, muscle bulk is small with normal texture. She has a weak gag reflex and poor ability to swallow. Her muscle stretch reflexes are weakly present and difficult to elicit, without asymmetry in the upper limbs.

When supine, she assumes a long C-curve of the spine with shoulders in adduction and elbows fixed in extension. The pelvis is maintained in an oblique position, the left hip is obviously dislocated, and there is bilateral genu recurvatum. Long, tapering digits are observed, and there is a coarse, hairy patch over the lumbosacral spine. When the infant is suspended in the prone position, she is unable to extend her head or lift her lower limbs.

- **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

The infant's facies are diminished though present stretch reflexes probably mitigate the diagnosis of spinal muscular atrophy. The other peripheral neuromuscular disorders, with the exception of those which affect ocular motility, such as myotubular myopathy, the encephalomyopathies, and some of the congenital dystrophies, remain in the differential.

Because other organs can be affected in association with peripheral neuromuscular disease, it would be desirable to have information from an examination of the chest and abdomen.

PHYSICAL EXAMINATION, continued

No cardiac abnormalities are detected. There is no hepatosplenomegaly. Because the infant sucks and swallows poorly, tube-feedings have been initiated. However, she does not require assisted ventilation.

- **If necessary, revise your differential diagnosis based on the additional physical examination results.**
- **Are there laboratory or other tests that could help you in your differential diagnosis?**



COMMENTARY IV

The physical examination did not fit a storage disease, e.g., glycogen storage (Pompe disease), which would affect the heart, result in a rubbery texture of the muscles, and possible hepatomegaly. The other diagnostic possibilities remain, some more likely than others.

MRI of the brain and spine are requested to rule out cerebral abnormalities and possible spinal dysraphia. These are normal.

To approach the electrodiagnostic examination in logical sequence, it would be important to determine the integrity of the peripheral nerves and in this case to include most particularly the right brachial plexus supply. A tibial H-latency may be included because it provides a longer nerve segment for study and is easily elicited in infants.

Since the infant is diffusely affected, an upper and lower limb should be evaluated proximally and distally. In this case, the right upper and left lower limbs were evaluated.

ELECTROPHYSIOLOGIC DATA

ELECTROMYOGRAPHY										
n = normal incr = increased decr = decreased 0 = absent 1+ = minimal 4+ = maximal crd = complex repetitive discharge fasc = fasciculation potential myk = myokymic discharge myt = myotonic discharge nmt = neuromyotonic discharge p wave = positive sharp waves fib = fibrillation potentials recr = recruitment amp = amplitude dur = duration poly = polyphasic potential										
R/L	MUSCLE	INSERTION		SPONTAN		VOLUNTARY				
		activ	p wave	fib	other	recr	amp	dur	poly	effort
R	deltoid	incr	1+	1+	0	n	decr	decr	incr	-
R	biceps	n	0	1+	0	n	decr	decr	incr	-
R	brachioradialis	n	0	1+	0	n	decr	decr	incr	-
L	tib anterior	incr	1+	1+	0	incr	decr	decr	incr	-
L	gastrocnemius	incr	1+	1+	0	incr	decr	decr	incr	-
L	gluteous maximus	incr	1+	1+	0	incr	decr	decr	-	-

MOTOR NERVE CONDUCTION										
nr = no response										
NERVE	LATENCY (ms)			AMPLITUDE (mv)			CONDUCT VEL (m/s)			
	R	L	Norm	R	L	Norm	R	L	Norm	
	axillary	-	-	-	-	-	-	-	-	-
erb's pt. to mid deltoid	2.5	-	1.9-2.9	-	-	-	-	-	-	



musculocutaneous	-	-	-	-	-
erb's pt. to mid biceps	2.8	-	2.3-3.6	-	-
ulnar	-	-	-	-	-
elbow to hypothenar	6.8	-	-	3	-
wrist to hypothenar	1.8	-	-	-	-
peroneal	-	-	-	-	-
fibula head to EDB	7.5	-	-	-	-
ankle to EDB	3.7	-	2.8-3.1	3	-

H-REFLEX								
NERVE	STIMULATE	RECORD	LATENCY (ms)			AMPLITUDE (mV)		
			R	L	Norm (<)	R	L	Norm (>)
tibial	-	-	-	-	-	-	-	-
popliteal to mid gastrocnemius	-	-	16.2	-	15.95 + 1.45 Bryant & Eng	-	-	-

All motor nerve conduction values are within normal limits for age. The tibial H-reflex study is also within normal limits. The electromyographic examination of the right upper and left lower limbs reveals rapid recruitment throughout with minimal effort from the infant. Occasional positive sharp waves and fibrillation potentials are found in a widespread distribution. The motor unit potentials are of low amplitude and short duration.

- **On the basis of both the clinical and electrodiagnostic evaluations, formulate your final impression. List the most likely diagnosis followed by other possibilities that are not excluded by the data. Eliminate those diagnoses not supported by the data.**
- **What other diagnostic procedure are needed?**

DIAGNOSTIC IMPRESSION

1. This infant has a myopathic process. Possibilities include congenital myopathy, congenital dystrophy, and metabolic or inflammatory disorder.
2. There is no evidence of brachial plexopathy.

COMMENTARY

A "myopathic" EMG is not disease specific and may be found in a variety of congenital myopathies, i.e. , central core or multicore disease, nemaline myopathy, centronuclear myopathy, congenital fiber-type disproportion, and many incompletely classified muscle disorders. It is found in congenital dystrophies, the dystrophinopathies, and the



inflammatory and metabolic disorders. The electrodiagnostic data must be correlated with the physical examination and verified by pathologic and/or genetic data.

It is important when dealing with infants and young children who need an electrodiagnostic study to proceed with a carefully designed study. The examiner must neither inflict prolonged discomfort nor be so hasty as to miss important data.

In this infant, studies were performed in the right upper limb so that the generalized hypotonia and the possibility of brachial plexopathy could be evaluated without duplicating the same procedures. To gather information from a wider distribution, both lower limbs were studied, the right by conduction studies, the left by needle electromyography.

FOLLOW UP

A biopsy of the left quadriceps was performed at 3 1/2 weeks. On modified trichrome stains, a number of fibers showed "palisading clusters of dark staining rod-like bodies mostly in the periphery of the fibers." Electronmicroscopy revealed that "many of the fibers on longitudinal section contained nemaline rods . . . seen in areas of disorganization of the myofibrillar architecture."

Nemaline rod myopathy is one of the congenital myopathies which may present as a floppy infant with weak facies and feeding problems. In some cases, the disorder may not be obvious until the child is ambulatory and is afflicted with respiratory problems to the point that the child may require a tracheostomy. Because the lower limbs may remain relatively strong, the child may be dubbed "a walking tracheostomy."

Most cases of nemaline myopathy follow an autosomal-dominant pattern of inheritance, but autosomal recessive and sporadic cases have been described. The autosomal dominant form has been localized to 1p13-q25. A gene for the autosomal recessive type has been assigned to chromosome 2q.

The EMG in nemaline myopathy has been reported to show low-amplitude, short-duration motor unit potentials in a majority of cases. Fibrillation may be observed, particularly in neonates. The reason for this is speculative at present. Long-duration polyphasic potentials with reduced recruitment may be seen in older children.

SUGGESTED READING

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