



EMG Case No. 52, June 2001

Presenting Symptoms: Joint Hypermobility; Knee and Ankle Subluxation

This case is no longer available for CME credit.

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Appropriate Audience: Residents and practicing physicians

Learning Objectives: After completing this educational activity, the participant will be able to (1) develop the differential diagnosis of a constellation of presenting symptoms in a critical manner based on history, physical examination, and EMG findings, (2) explain the etiology and pathology of this diagnostic category, and how to characterize and differentiate subgroups therein, (3) critically analyze the EMG findings and how they relate to hereditary vs. acquired forms of the disease, (4) explain the clinical course and natural history of the diagnostic category in question, and (5) define the possible neurological and musculoskeletal complications of this disease group and how to implement rehabilitation interventions.

History

A 16-year-old, right hand dominant girl presents to the outpatient physiatry clinic with a referral for management of joint subluxation and hypermobility. Her symptoms started 5 years ago with ankle and knee laxity, manifesting as frequent ankle sprains and patellar subluxation resulting in falls. Historically she met all normal childhood and adolescent milestones, and was previously active in sports although not adept at any. For 3 years she has refrained from sports due to the joint instability. Two years ago she had an unsuccessful left knee operation in an attempt to stabilize the patellar ligaments. Over the last year, she began to have bilateral wrist complaints including "cracking" and laxity. She denies any significant past medical history, trauma, constitutional symptoms, or bowel or bladder complaints.

- *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

The differential diagnosis for joint laxity is long and includes such conditions as Marfan's Syndrome, Ehlers-Danlos, other connective tissue disorders, bone diseases such as rickets, osteogenesis imperfecta, mucopolysaccharidoses, homocystinuria, and Noonan's syndrome. Most of these conditions would be diagnosed based on correlation with associated symptoms that this patient does not have. Additionally, symptoms starting this late in adolescence would essentially rule out some of the listed metabolic disorders. She was referred to the clinic from the pediatric rheumatologists with a presumptive diagnosis of Marfan's.

History, continued

Detailed family history reveals no specific similar problems in first or second-degree relatives, except for the mother who has mildly high arched feet. History of rheumatic



disease or arthritis is negative. She denies toxic exposure of any type, recent illness, or unusual travel history. The review of systems is otherwise negative, in particular for the neuromuscular system, except for a comment that, "Well, sometimes in the shower I can't feel the difference between hot and cold on my hands." She denies other constitutional symptoms, muscle fatigue, or muscle stiffness. She is not currently on any medications and the developmental history and Tanner stages are normal.

- *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

There are two clues that point towards a much broader differential diagnosis here. One is that the mother has high arches. This may be a sign of a neuropathic process including peripheral polyneuropathy or other distal neuropathy affecting the foot intrinsic muscles. In addition, the second (initially elusive) clue of sensory changes in the hands expands the differential. Now one must also consider central processes including stroke, multiple sclerosis, vascular or spinal cord etiologies, peripheral nerve problems such as compressive neuropathies, or problems caused by other systemic illnesses including thyroid disease, diabetes, or neoplasm. There is no history of toxins, recent infections or systemic illness, which would help exclude many types of neuropathies, including systemic or infectious peripheral neuropathies.

Physical Examination

The patient appears her stated age, communicates appropriately, and is in no apparent distress. Her vital signs are normal and she is of thin build, relatively tall, with an arm span to height ratio of 1.03. The ratio of upper body segment to lower segment was 0.85. Detailed musculoskeletal examination reveals mild atrophy of the hand intrinsics bilaterally, without fasciculations or myotonia of the thenar eminence. Joint range of motion is very lax in the fingers and wrists with ability to easily touch the tip of her thumb dorsally to the forearm. There is no obvious synovitis, redness, or warmth in the joints. Proximal upper and lower extremity joints are normal. In the lower extremities her arches are mildly high and her ankles have marked laxity with large unstable translation of the ankle in both inversion and eversion. The left knee has a surgical scar longitudinally along the medial aspect, and significant instability and translation of the patella laterally, left greater than right. The skin examination reveals no abnormalities or keloid formation.

- *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 physical examination reveals marked joint hypermobility symmetrically in the distal joints and knees. There are a number of described criteria for the diagnosis of Marfan's syndrome including arm span to height ratio greater than 1.05 (just missing criterion here) and upper to lower body segment of less than 0.86 (meets criterion for Caucasians). There is however, some atrophy in the distal hand muscles, which will warrant further neurologic evaluation. The absence of fasciculations and myotonia make motor neuron disease and myotonic dystrophy less likely.



Physical Examination, continued

The neurologic examination reveals marked decreased sensation to pinprick and vibration in a stocking-glove distribution to just above the wrists and ankles bilaterally. Proprioception is markedly impaired in the hands and feet as well. Manual muscle testing reveals hand intrinsic strength of 3+/5 (dorsal interossei) and grip of 4/5. Lower extremity strength was also decreased with inability to heel and toe walk and mild bilateral foot drop. Muscle stretch reflexes are 1+ at the biceps and triceps, trace at the knee, and absent elsewhere bilaterally. The cranial nerve examination was normal. Cerebellar exam and Romberg were normal without gait ataxia. The patient's muscle tone is normal and there is no palpable thickening of the nerves.

- *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

With the additional physical examination data, the differential diagnosis now can be focused more accurately. The presence of a distal, symmetrical process affecting both motor and sensory nerves is deduced. The hypoactive muscle stretch reflexes point away from a CNS process or motor neuron disease, which typically yield upper motor neuron signs. These findings should alert one to the possibility of a type of peripheral neuropathy, either acquired or hereditary.

Rheumatologic and screening laboratory testing was normal. Genetic testing for Marfan's was arranged, but we thought it important to obtain an EMG to characterize the neuropathy.

Electrophysiologic Data

SENSORY NERVE CONDUCTION STUDIES							
nr = no response							
NERVE	SIDE	STIM SITE	RECORD	cm	AMPL	LAT	CV
Sural sensory	L	Calf	Lat. Malleolus	14	nr	-	-
Ulnar sensory	L	Wrist	5 th digit	14	nr	-	-
Median sensory	L	Wrist	2 nd digit	14	nr	-	-
Radial sensory	L	Forearm	Snuff box	10	nr	-	-
Sural sensory	R	Calf	Lat. Malleolus	14	nr	-	-



MOTOR NERVE CONDUCTION STUDIES							
nr = no response							
NERVE	SID E	STIM SITE	RECORD	cm	AMPL	LAT	CV
Peroneal motor	L	Anterior ankle	E.D.B.	9	nr	-	-
Peroneal motor	L	Fibular head	Anterior tibialis	7	6.9	3.8	-
Peroneal motor	L	Popliteal fossa	Anterior tibialis	17	7.1	6.1	43.5
Tibial motor	L	Medial ankle	Abductor Hallicus	8	0.9	6.3	-
Ulnar motor	L	Wrist	Hypothen ar	7	9.0	3.0	-
Ulnar motor	L	Elbow	Hypothen ar	26.5	8.4	6.7	52.7
Median motor	L	Wrist	Thenar	7	10.4	3.6	-
Median motor	L	Elbow	Thenar	28	8.5	7.8	50.0
Ulnar F-response	L	Wrist	Hypothen ar	-	-	-	27.5

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+									
AMPplitude: 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
L	Biceps brachii	N	0	0	N	N	N	N	N
L	First dorsal interosseous, hand	Sust	3+	0	N	Dec 1+	Inc 3+	Inc 1+	N
L	Vastus medialis	N	0	0	N	N	N	N	N
L	Anterior tibialis	Sust	2+	0	N	N	N	N	N



L	First dorsal interosseous, foot	Sust	2+	0	N	Dec 1+	Inc 1+	Inc 1+	Inc 1+
L	Gluteus maximus	N	0	0	N	N	N	N	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.)*

Electrophysiologic Data, continued

Repetitive Nerve Stimulation

Stimulus delivered via surface electrodes under the same set up as ulnar motor, with immobilization of left wrist and hand.

No decrement after 2Hz stimulation and no facilitation immediately after 10 seconds of exercise.

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

Diagnostic Impression

The history, physical examination, and electrodiagnostic findings support the diagnosis of hereditary motor sensory neuropathy type II. The presence of a distal, symmetric neuropathy without evidence of prolonged distal latencies or conduction velocities helps place this into the type II category with hereditary axonal degeneration. The lack of temporal dispersion helps clarify the process as an hereditary disorder.

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

Commentary V

This patient presents in the second decade of life with an insidious onset, slowly progressive loss of motor and sensory function in a distal to proximal gradient, manifesting as hypermobility and joint subluxation. A thorough history in these patients is very important, because after this diagnosis was made, additional family history revealed that her father had also had difficulties with his feet. As discussed above, her sensory symptoms were not mentioned initially, and only on a thorough review of systems of the endocrine system did this come to light. Included in the differential for this patient are other causes of toxic, metabolic, or disease associated axonal neuropathies, Freidreich's ataxia, myotonic dystrophy, and motor neuron disease. She has no upper motor neuron signs suggestive of a more central process. She also has no nerve thickening (onion bulb formations), that is more characteristic of Charcot Marie Tooth Disease (HMSN type I).



Electrodiagnosis proves to be very useful in formulating a diagnosis for this hereditary neuropathy. The key point is that the distal latencies and conduction velocities, including proximal responses such as the F-response, were normal in this patient. This is in contrast to what would be observed in a demyelinating process such as CMT I, in which motor CVs are often 50% below the lower limit of normal. If ever in doubt on how to differentiate HMSN type I from II on nerve conduction studies, blink reflexes and facial motor latencies are normal in type II. On needle examination, both types can have muscle denervation, but in HMSN type II the upper extremities tend to be less involved, with a propensity for weakness of the ankle plantar flexors.

The natural history of both of these diseases is slow progression with a normal life span, and most patients remaining ambulatory with mild disability. Interestingly, the type I form tends to be detected in the 1st or 2nd decade, whereas the type II tends to present in the 3rd and 4th decades. One explanation for this patient's early presentation is a wide range of expression of the genetic defect in HMSN type II. Rehabilitation interventions are aimed at preventing contractures, foot deformity, and injury to insensate soft tissues with the use of positioning, splinting, orthosis, and custom shoe wear. Exercise and strength training can be beneficial to help compensate for weak distal muscles. Psychological counseling as well as environmental access and adaptive equipment are also frequently needed to assist patients with adjustment to disability and for optimizing function.

Bibliography

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