CASE 1991-4

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CLINICAL ABSTRACT:

This 26 year old single hispanic female noticed lack of sensation and clumsiness in all digits of the left hand. The initial symptoms evolved over a matter of days to several weeks. The numbness became more dense, but was not associated with paraesthesia or pain. Subsequently she noted numbness in the 4th and 5th digits of the right hand and then below the left knee; more recently progressive clumsiness, difficulty with walking, and altered balance supervened and have compromised her athletic ability. On awakening one morning she noted both pupils to be dilated. Since, she notes that bright sunlight causes unusual glare and she has difficulty reading. Past medical history and family history were noncontributory and there was no known toxic exposure.

Neurologic examination was remarkable for pupils dilated to 8 to 9 mm and minimally reactive to light. No accommodation was seen during convergence, although extraocular movements and other cranial nerve functions were normal. Motor examination showed minimal weakness of the deltoids, right triceps, intrinsic musculature of the right hand, left biceps and right iliopsoas. The deep tendon reflexes were brisk at the biceps with bilateral inverted tricep jerks and absent ankle and knee reflexes. Sensory examination showed a remarkable reduction of vibratory sense at the ankles with a mild reduction of vibration at the fingers. Appreciation of light touch and pin prick was decreased over the entire left hand and the lateral 3rd, 4th, and 5th digits of the right hand. The patient's gait was unsteady, especially on tandem, but she could walk unassisted. Romberg was decidedly abnormal. The remaining features of her clinical exam were normal. Needle examination of multiple muscle groups showed large numbers of normal motor units, normal insertional activity, and no active denervation potentials. Sural nerve sensory responses were unobtainable, with normal peroneal and posterior tibial nerve motor nerve conduction velocities and F-wave latencies. Left and right median, left and right ulnar, and left radial sensory responses were unobtainable. The left ulnar motor response amplitude showed a 20% decrement of distal motor latency across the elbow. These electrical findings were consistent with a generalized and somewhat asymmetric, primarily sensory polyneuropathy.

Pertinent laboratory studies included a normal complete blood count, ESR, blood chemical profile, and serum protein electrophoresis and immunoelectrophoresis. Serology for ANA, anti-Ro, anti-La, anti-single and double stranded DNA was negative. C26/C22 and C24/C22 long chain fatty acid ratios and serum phytanic acid levels were normal. Lipid panels and several serum lipoprotein electrophoreses were normal. Separation of lipoproteins followed by electron microscopy showed no abnormalities and denaturing gradient gel electrophoresis of lipoprotein particle distribution was unremarkable (Dr. Trudy M. Forte, University of California, Berkeley). Mass spectroscopy of extracted frozen peripheral nerve showed excess amounts of cholesterol and cholestanol (Dr. Steven Tint, East Orange).

MATERIAL SUBMITTED: One toluidine blue stained slide, one kodachrome of oil red 0 stained section and one 35-mm EM slide of sural nerve biopsy.

POINTS FOR DISCUSSION:
1) Is this a variant or an early stage of Tangier disease?
2) Is this a new disease?