

CASE 1990-11

Brian W. Little, MD, PhD
Lehigh Valley Hospital Center
Allentown, Pennsylvania

The patient is a 22 year old male. He was the product of a normal, full-term pregnancy with a "complicated delivery". Within the first year of life he developed a myoclonic seizure disorder requiring phenobarbital and steroid therapy. The diagnosis of "spastic cerebral palsy" was made. Motor milestones were delayed; he sat at 19 months and walked at 48 months. A diagnosis of mild mental impairment was made. His seizures were difficult to control, but he stabilized on a regimen of Tegretol, Depakote and Diamox.

At age 21 his caregivers complained that he would collapse after walking a short distance. Immediately before falling, he would complain of pain in his hips and thighs. He was evaluated by an orthopedic surgeon who found no evidence of disease in the pelvis or lower limbs to explain the fatigue and pain. A myelogram, CT scan and MRI were performed which showed a C-5 to C-7 stenosis. A cervical decompression was performed seven months later with some relief of symptoms for two to three months. Five months later his fatigue had progressed to the point where he could not walk 200 feet or climb one flight of stairs. His upper extremities were less affected. He saw a cardiologist who undertook exercise testing and echocardiography. The summary of the exercise testing was: "Marked impaired exercise capacity. It seems to be a neuromuscular rather than cardiovascular basis. No evidence of [cardiac] ischemia. No hemodynamic instability." The echocardiogram was normal.

Examination at age 22 showed his spastic quadriparesis to be unchanged. The weakness was documented as above. Sensory examination and EMG/NCV were not undertaken due to poor cooperation from the patient. Optic disk pallor and nystagmus were unchanged from prior examinations. Routine laboratory tests were normal, except for a slightly elevated cholesterol [217 mg/dl] and triglyceride [194 mg/dl]. A creatine kinase was 560 U [Ref range 35-232U]. A muscle biopsy was performed.

SLIDES PROVIDED

1. One H&E stained, paraffin embedded slide.
2. One 2x2, 35 mm slide.

FOR DISCUSSION:

1. What is your morphologic diagnosis?
2. What further diagnostic or therapeutic steps would you undertake or recommend?