CASE No 10

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This 66-year-old Chinese man had a background of elevated cholesterol (on statin), hepatitis B carrier status and impaired vision from vitreous hemorrhages. He presented neurologically at age 61 years with dragging of right leg, foot drop, numbness, and tingling in the dorsum of the right foot. The diagnosis of peroneal neuropathy was supported by NC and EMG studies. He improved without treatment. At age 64 years he reported progressive leg weakness with difficulties climbing stairs and getting up out of a chair (the patient had given up golf 7 years previously because of difficulty with his swing!). Family history was significant for leg weakness in his late mother, heart disease and leg weakness in his late elder brother. The patient's three other siblings and his two adult sons were healthy. Neurological examination revealed proximal leg weakness, gait waddle, bilateral foot drop, and preservation of reflexes. Creatinine kinase was elevated at 387 U/L. Needle EMG disclosed no evidence for myopathy. Statin was discontinued but CPK did not normalize.

Muscle biopsy of quadriceps was done (November 2008). DNA was extracted from peripheral blood and the frozen muscle sample. Sequencing of the DES, CRYAB, MYOT, and ZASP genes unveiled no mutations.

Material submitted: cryosection stained with either H&E or HPS.

Points for discussion: diagnosis?