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A PATIENT WITH CHOREA-ACANTHOCYTOSIS AND DILATED CARDIOMYOPATHY

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We discuss the case of a 43-year-old male with a 20-year history of a slowly progressive neurodegenerative syndrome. He first presented with epilepsy at the age of 26 but over the subsequent 10 years personality and memory changes were noted and the epilepsy became refractory to therapy. Aged 35 he became dysarthric and by the age 40 had developed extrapyramidal features, axial limb rigidity and postural instability. He also reported exercise intolerance. An ECG showed left bundle branch block, cardiac monitoring detected episodic non-sustained VT and an echocardiography and cardiac MRI demonstrated severe left ventricular systolic impairment. A biventricular defibrillator was implanted. Serological and cerebrospinal fluid investigations were unremarkable throughout his illness except for a persistently elevated creatinine kinase (2794 IU/l). Electromyography demonstrated proximal myopathic changes and a muscle biopsy revealed mild non-specific myopathic changes with possible fibre-type grouping. MRI of

the brain showed bilateral atrophy of the head of caudate nuclei and globus pallidus. Serial blood film examination eventually demonstrated a significant proportion of acanthocytes and a Chorein Western Blot confirmed the diagnosis of chorea-acanthocytosis. The association of this rare neurodegenerative syndrome with a dilated cardiomyopathy is novel finding. Another important learning point is to persist with blood film examination if the clinical suspicion of neuroacanthocytosis arises.



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