Muscle cramps and a raised creatine kinase

A 40-year-old man presented with a 25-year history of cramps affecting the abdomen, neck, and limbs. Examination revealed fasciculation in the forearms, abdomen, and chin (video on the Neurology® Web site at Neurology.org). There was shoulder girdle wasting with bilateral mastectomy scars (figure, B and C). Creatine kinase (CK) was 1,650 U/L (normal < 310 U/L). Electrodiagnostic studies revealed sensory neuropathy with neurogenic changes on EMG. Genetic testing demonstrated excess CAG repeats in the androgen receptor gene, confirming Kennedy disease.¹ This X-linked disorder is the most common adult-onset spinal muscular atrophy. CK can be markedly raised.² Gynecomastia results from androgen insufficiency and can precede the development of neurologic symptoms.

AUTHOR CONTRIBUTIONS
Dr. Whittaker: design/conceptualization of the study, analysis/interpretation of neurophysiology data, drafting/revising the manuscript. Prof. Chinnery: drafting/revising the manuscript. Dr. Miller: drafting/revising the manuscript.

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From the Institute of Neuroscience (R.G.W.), The Medical School, Newcastle University; and the Wellcome Trust Centre for Mitochondrial Research (P.F.C., J.A.L.M.), Institute of Genetic Medicine, Newcastle upon Tyne, UK.

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