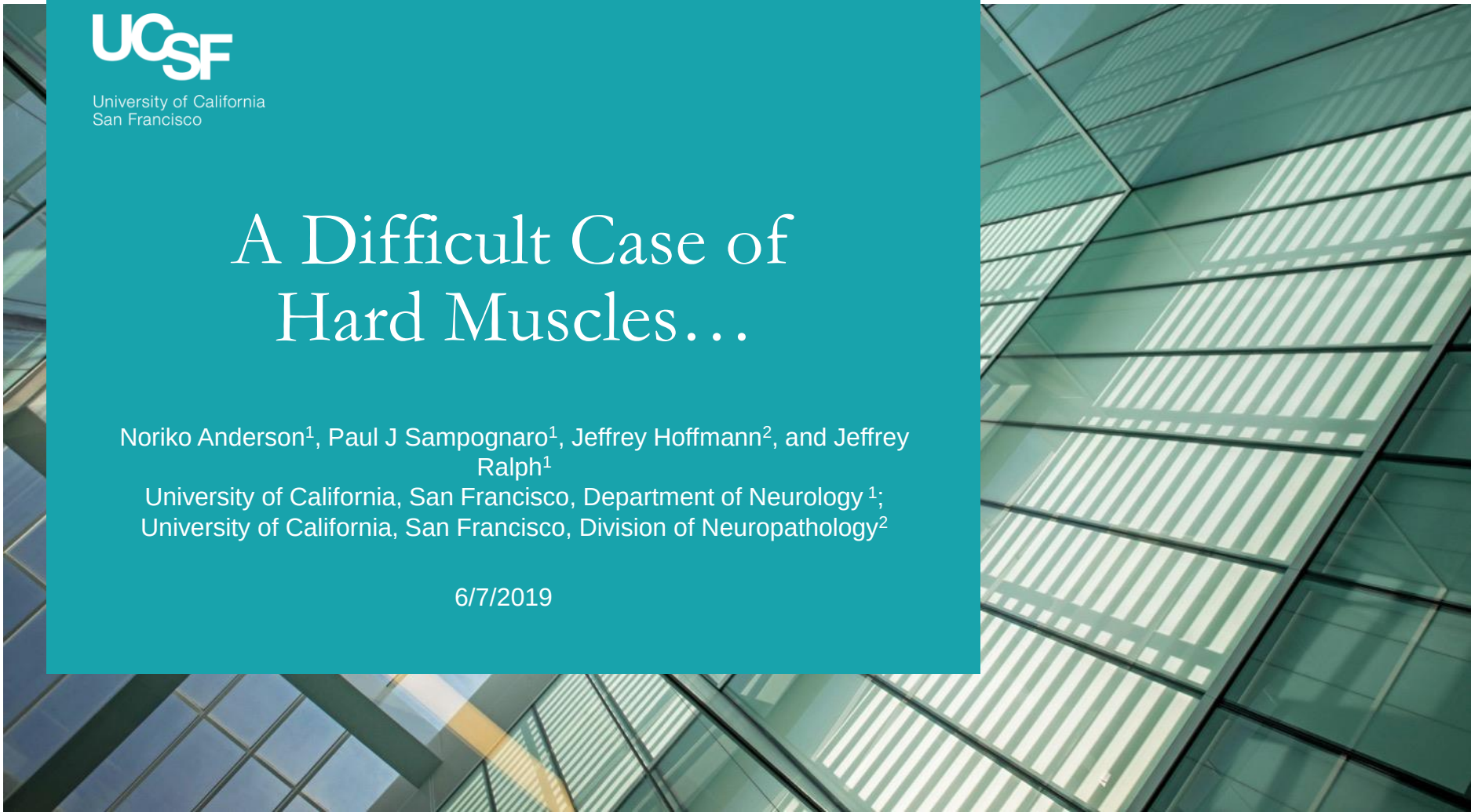

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A Difficult Case of Hard Muscles…

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6/7/2019

The Case

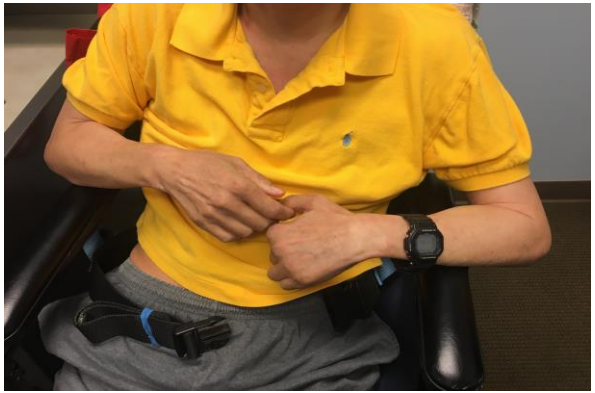
- 56-year-old, right-handed man with a history of two prior ischemic medullary strokes and residual left-sided weakness presents who presented for progressive muscle stiffness
- August 2016 about a year after his stroke, he noticed the gradual onset of tightness in the right > left side of his body
- There is no pain, muscle twitching, prominent startle response, excessive sweating, difficulty opening his eyes or letting go of objects from his hands, paralytic episodes in early life or from cold temperatures
- He has never been able to whistle

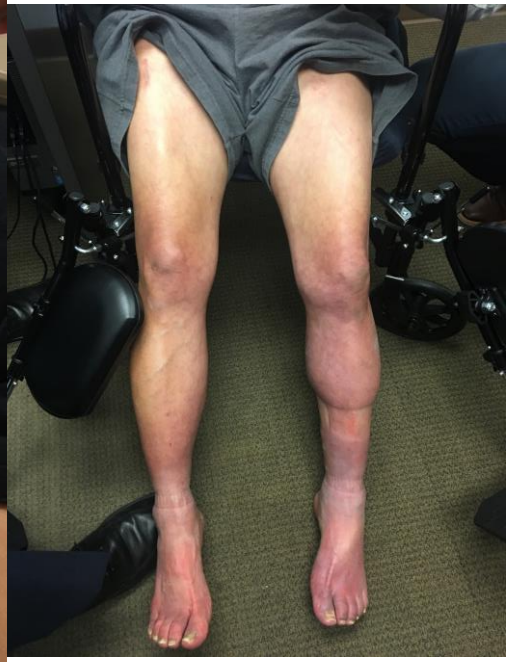
Past Medical History

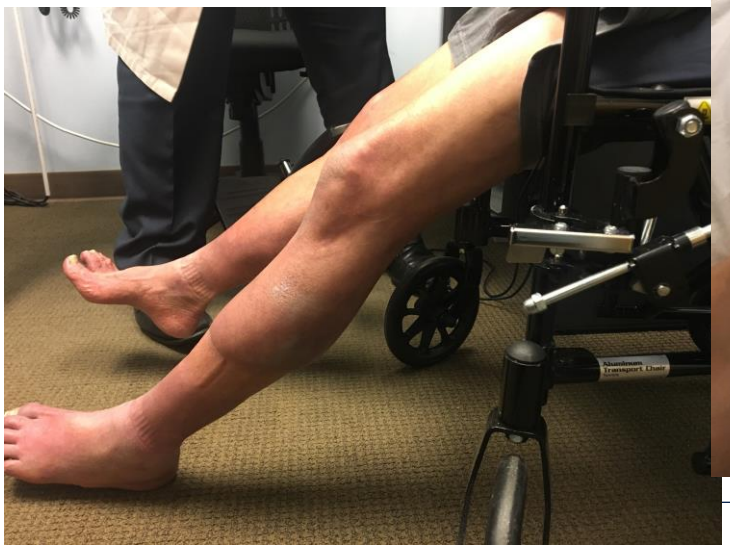
- PMH: paroxysmal atrial fibrillation, diabetes, hypertension, mild transaminitis, CAD, left knee arthroscopy in 1989
- Medications: atorvastatin 10 mg, tizanidine 6 mg TID, carbamezapine 200 mg BID, metformin 1000 mg BID, lisinopril 20 mg daily, metoprolol 300 mg daily, dabigatran 150 mg BID, B12 1000 mcg daily, vitamin D
- Allergies: procaine
- FH: diabetes and hypertension in his brother and parents, no muscular dystrophy, his 90-year-old father has cataracts and CAD, a younger brother with frontal balding, mother with PD, cousin with Down Syndrome, nephew with autism and 2nd cousin with seizures, no early cataracts, cardiac arrhythmias
- SH: married, never smoked, no alcohol use, works in tech sales

Exam

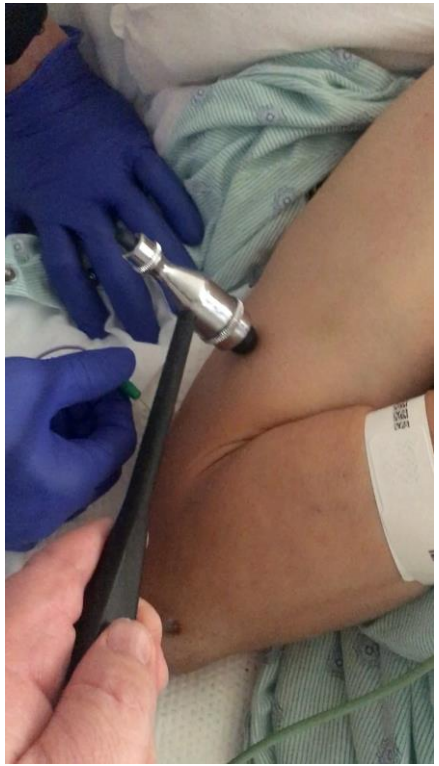
- General exam notable for + LE edema, contracture at right elbow, hyperextended right knee, left ankle in AFO, some temporal wasting and frontal balding
- NEUROLOGIC EXAM:
  - CRANIAL NERVE: +Dysarthria
- MOTOR: Muscle tightness on right greater than left arm and leg, proximal > distal muscle tightness. Hemiparesis on left with an UMN pattern (FE 1, WE 1, HF 2). On the right: HF 2, unable to flex knee (locked, rigid in a hyperextended fashion with spasm of both quads and hams), some resistance on right elbow flexion limited due to continuous contraction of right triceps, brachioradialis, and biceps (B 4, T 4), able to extend fingers. No grip or blink myotonia. No clear hypertrophy of calves or thigh.



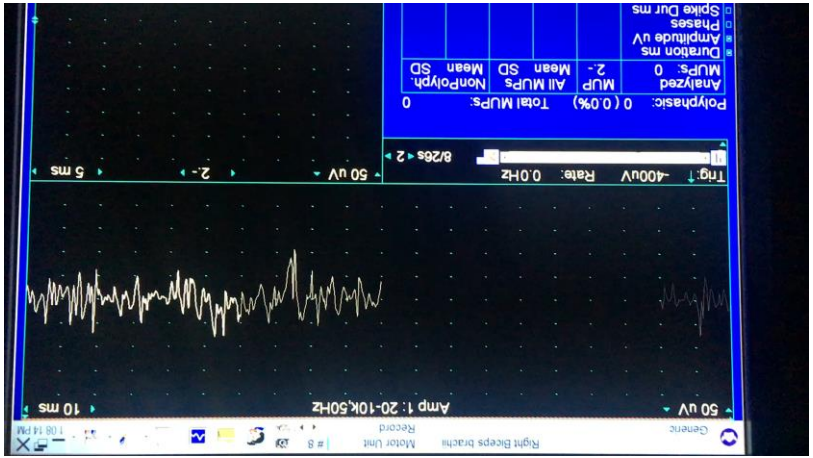




Diagnostic Studies

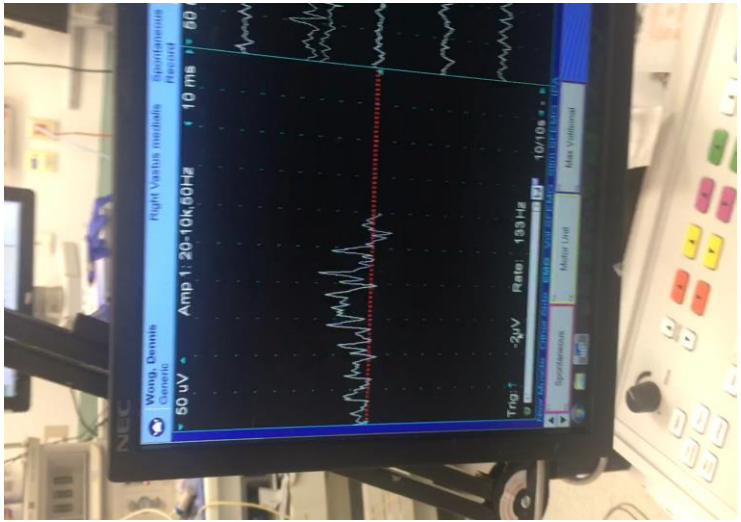


- CK 3597 -> 2738 (CK 116 in 9/15)
- MRI of the brain which was unremarkable for age, MRI of the c-spine with mild degenerative disc disease
- CT scans of the chest, abdomen and pelvis - negative for cancer
- Negative serum GM1 antibodies & VGKC complex antibodies



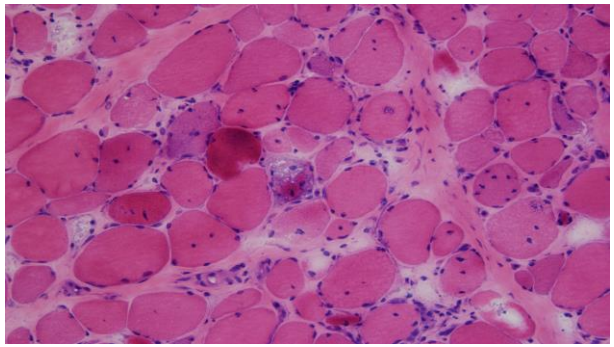
Diagnostic Studies

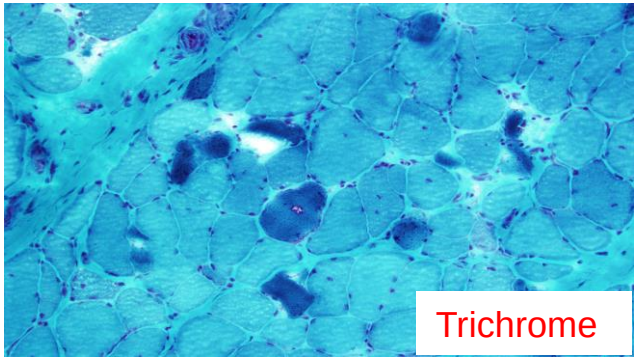
- Neuromuscular blockade with rocuronium was attempted prior to biopsy
- Rocuronium 100 mg IV given. After 1 min., TOF intact at left hand. Additional stimulators placed on face and right hand. TOF intact at those sites
- Another 50 mg of rocuronium given. After 5 min., TOF abolished at face and right hand but some responses remained at left hand
- Anesthesiologist did not agree to give more rocuronium
- His right biceps and quadriceps were still hard to the touch

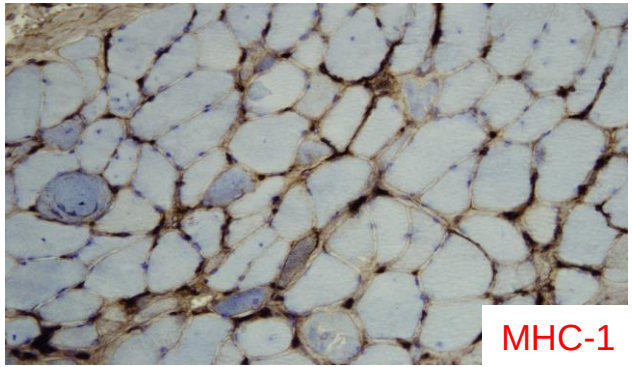


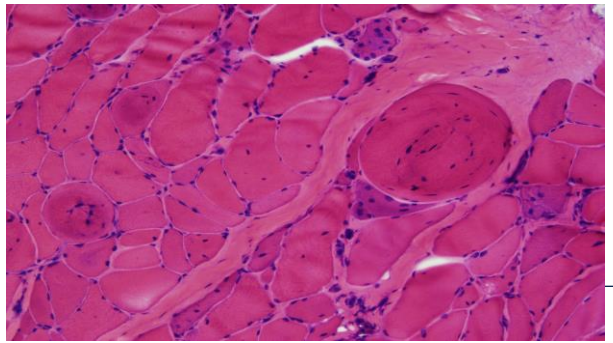
Muscle Biopsy

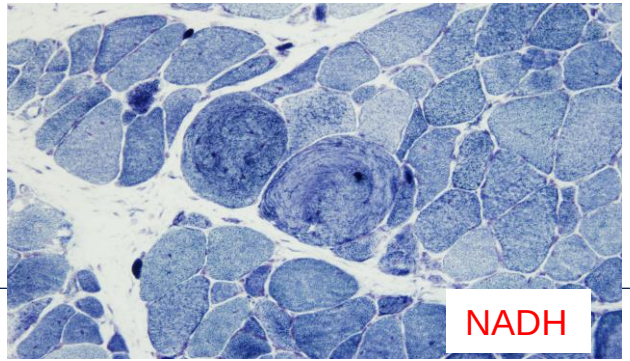
- Right vastus lateralis muscle biopsy showed…

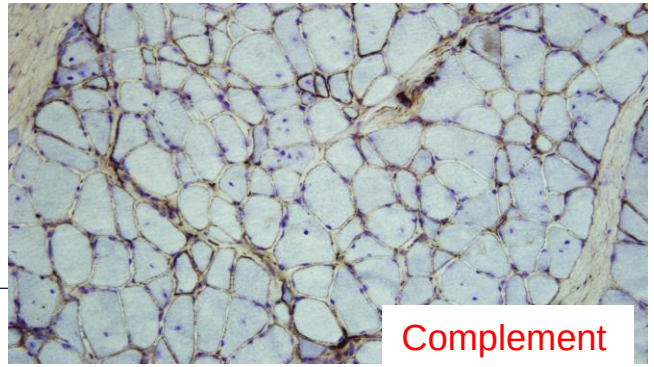












Further Diagnostic Studies

- Myotonic dystrophy testing (Athena comprehensive myotonia evaluation) - negative
- 19 item myositis antibody panel (ARUP) - negative
- LGMD panel pending

- **HMGCR antibodies > 200 (ARUP)**

Discussion

- Continuous myotonia is a rare clinical finding and can be seen in genetic disorders
- Myotonic discharges have previously been reported in autoimmune necrotizing myopathy
- Our case demonstrates an extremely rare presentation of acquired continuous myotonia (myotonia acquisita) secondary to an HMGCR-associated necrotizing myopathy

Patient Update

- Atorvastatin was discontinued
- He was started on prednisone, currently at 20 mg BID and recently started on methotrexate
- His CK has normalized
- He has improved strength in his arms and legs
- His swallowing has improved
- His left leg tightness has decreased

References

- GOUR, KN. Myotonia acquisita. Ind Med Gaz. 1951 Mar;86(3):99-100.
- MAAS, O. Observations on myotonia acquisita. Monatsschr Psychiatr Neurol. 1951 May;121(5):292-6.
- Eskild Colding-Jørgensen, Morten Duno, John Vissing. Autosomal dominant monosymptomatic myotonia permanens. Neurology/ Jul 2006, 67 (1) 153-155; DOI: 10.1212/01.wnl.0000223838.88872.da
- Hahn C, Salajegheh MK. Myotonic disorders: A review article. Iran J Neurol. 2016;15(1):46-53.
- Matthews E, Fialho D, Tan SV, et al. The non-dystrophic myotonias: molecular pathogenesis, diagnosis and treatment. Brain. 2009;133(Pt 1):9-22.