**LEGS WEAKNESS, QUADRICEP ATROPHY, AND ELEVATED CPK**

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**Abstract**
The patient is a 46 year old male with at least 20 years history of legs weakness and elevated CPK. He has most difficulty with climbing stairs and is no longer able to run. He was admitted to the hospital with rhabdomyolysis at age 25 with CPK above 10000 U/L. He had a muscle biopsy in 1988 and was given the diagnosis of myoadenylate deaminase deficiency. Over the years his CPK level remains between 1800-2600 U/L. He has noticed left thigh atrophy for the past few years. Examination revealed left quadriceps atrophy and weakness, and mild weakness of iliopsoas bilaterally. Left patella reflex was absent. EMG/NCS of both legs and left arm showed myopathic units in the proximal muscles; fibrillation and positive sharp waves and chronic repetitive discharges were seen in the left vastus lateralis. MRI with and without contrast of the left thigh showed marked atrophy of vastus lateralis, vastus intermedius, and vastus medialis as well as moderate atrophy of gluteus maximus and adductor magnus. Minimal edema was seen in the left anterior thigh muscles. Left vastus lateralis biopsy and genetic testing were performed.

**Clinical History**
- 46 year old RHD presented with longstanding history of legs weakness and elevated CPK.
- Never strong or fast as a child, but active physically.
- Age 25 - hospitalized with rhabdomyolysis. CPK reportedly >13000 U/L. Given the diagnosis of myoadenylate deaminase deficiency.
- Throughout the years continued to have subjective legs weakness:
  - Difficulty climbing stairs - legs tired out with only 1-2 flights
  - Can no longer run but walking is preserved
  - No distal weakness, no arms symptoms
- Past couple of years noticed left quadriceps getting smaller
- No episodes of tea/coke color urine
- CPK consistently ranges between 1800-2600 U/L
- Denies dysarthria, dysphagia, shortness of breath, facial weakness, ocular symptoms, bowel or bladder problems.

**History:**
Unremarkable medical, surgical, social and family history.

**Examination**
- Normal general examination
- Cranial nerves normal.
- Mild atrophy of left quadriceps
- Iliopsoas 4+-5 bilaterally
- Left quadriceps 4+-5
- Remaining muscles were strong.
- Absent left patellar reflex, remaining reflexes were normal. Planar response flexor bilaterally.
- Remaining neurologic exam normal.

**Investigation:**
- CPK 1623, 2207, 1621 U/L
- Aldolase 16.1 U/L
- EMG/NCS
  - NCS normal
  - EMG: moderate amount of myopathic units in proximal muscles.
  - 1 fibrillation potential, 2 positive sharp wave in the left vastus lateralis only.
- MRI LEFT THIGH:
  - Marked atrophy with minimal edema involving the vastus lateralis, intermedius, and medialis muscles.
  - Moderate atrophy of gluteus maximus and adductor magnus.
  - Minimal edema and contrast enhancement likely secondary to denervation atrophy.
- LEFT VASTUS LATERALIS BIOPSY:
  - NADH
  - Dystrophin 1
  - Dystrophin 2
  - Dystrophin 3
  - alpha sarcoglycan
  - merosin

**Dystrophinopathy presenting as isolated quadriceps myopathy:**

**Genetic testing:**
- Dystrophin duplication/deletion - negative
- Limb girdle muscular dystrophy panel - negative
- Considering dystrophin sequence analysis

**Discussion:**
- Differential diagnosis of selective quadriceps involvement:
  - Hereditary:
    - Dystrophinopathy presenting as isolated quadriceps myopathy
    - Becker’s muscular dystrophy
    - LGMD: 1B, 2B, 2H
    - Emery-Dreifuss: lamin A/C
    - HIBM3
    - Spinal muscular atrophy type III and IV
  - Acquired:
    - Focal myositis
    - Inclusion body myositis
    - Diabetic or non-diabetic lumbosacral plexopathy
    - L3-4 radiculopathy

**Literature cited:**