

## 60th ANNUAL DIAGNOSTIC SLIDE SESSION 2019

### CASE 2019-8

**Submitted by:**

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**Clinical History:**

This patient is a 16 year old male who presented with sudden onset of swelling in the hands and eyes. His creatine phosphokinase (CPK) was 870 IU/L in the Emergency Department and fluctuated between 600 and 900 over a period of several months. His ED labs were remarkable for a WBC of 20.0 and platelets of 138. MRI showed enlarged inguinal, external iliac, and popliteal lymph nodes, but no evidence of changes in the musculature. He was referred to neurology for his muscle weakness and hematology/oncology for his leukocytosis and thrombocytopenia. A muscle biopsy of the right vastus lateralis was performed.

**Material submitted:**

Right vastus lateralis, muscle biopsy: (1) H&E (frozen) (2) Gomori trichrome (frozen)

**Points for discussion:**

1. Workup and differential diagnosis based on the H&E and trichrome findings
2. Potential genetic findings in this patient and their correlation with the clinical and histopathologic findings