Amyloid myopathy: not your usual suspects

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A 31-year-old man presented with a 6-year history of slowly progressive calf atrophy and weakness. EMG showed distal myopathy with fibrillation potentials. Creatine kinase (CK) was 2848 U/L (normal < 310). Muscle biopsy showed myopathy and interstitial amyloid deposits (figure). The search for extramuscular amyloidosis was unrevealing. Next generation sequencing identified known pathogenic c.6124C>T and novel c.145-1G>A variants in the dysferlin gene (DYSF). Intramuscular interstitial amyloid deposits can occur in systemic amyloidosis (AL or ATTR) or less commonly in muscular dystrophies (DYSF and ANO5). Longstanding symptoms, young age of onset, calf atrophy, markedly elevated CK, and lack of systemic involvement are suggestive of muscular dystrophies.

**Abbreviations**

AL – immunoglobulin light chain amyloidosis
ANO5 – anoctamin 5 gene
ATTR – Transthyretin amyloidosis
CK – creatine kinase
DYSF – dysferlin gene
EMG - electromyography
## Appendix 1: Authors

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<tr>
<th>Name</th>
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References

Figure: Interstitial amyloid deposits involving skeletal muscle, necrotic muscle fibers and markedly reduced dysferlin immunoreactivity. Frozen sections of left vastus medialis muscle show (A and B) congophilic deposits in perimysial blood vessels (arrows) and in adjacent endomysium encasing muscle fibers (arrow heads) on a Congo Red stained section viewed under (A) light microscopy and (B) rhodamine optics. (C) H&E stained section shows necrotic fibers replaced by macrophages (arrows). (D) Sarcolemmal dysferlin immunoreactivity is markedly reduced on patient’s muscle fibers compared to the control section.
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