CENTRAL ILLUSTRATION: Skeletal muscle phenotype in patients with truncating titin variants and familial dilated cardiomyopathy

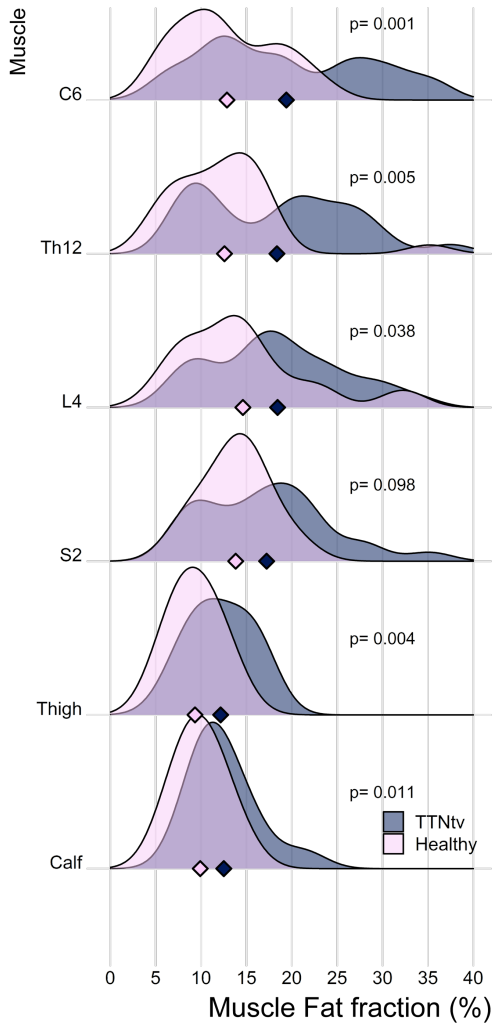
Design

25 individuals with TTNtv
(most common cause of DCM)

Fat fraction on muscle MRI
(compared to 25 healthy controls and 7 controls with non-TTNtv genetic DCM)

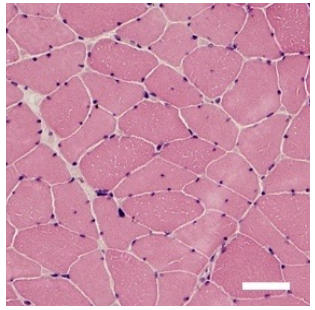
Histology and ultrastructure on muscle biopsies

Fat replacement of skeletal muscle

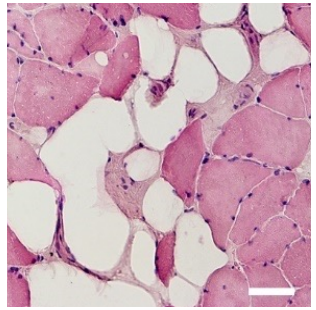


Muscle biopsy findings

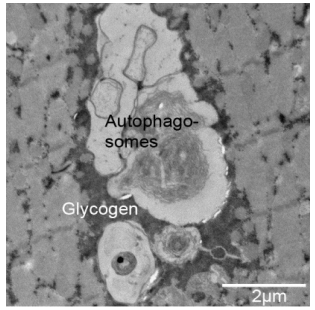
Centrally located nuclei



Fat replacement



Glycogen and large autophagosomes



Clinical message

TTNtv

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Skeletal muscle affection

- Skeletal muscle fat infiltration
- Myopathic findings

Consider specialized diagnostic work-up in patients with symptoms or objective findings of muscle involvement