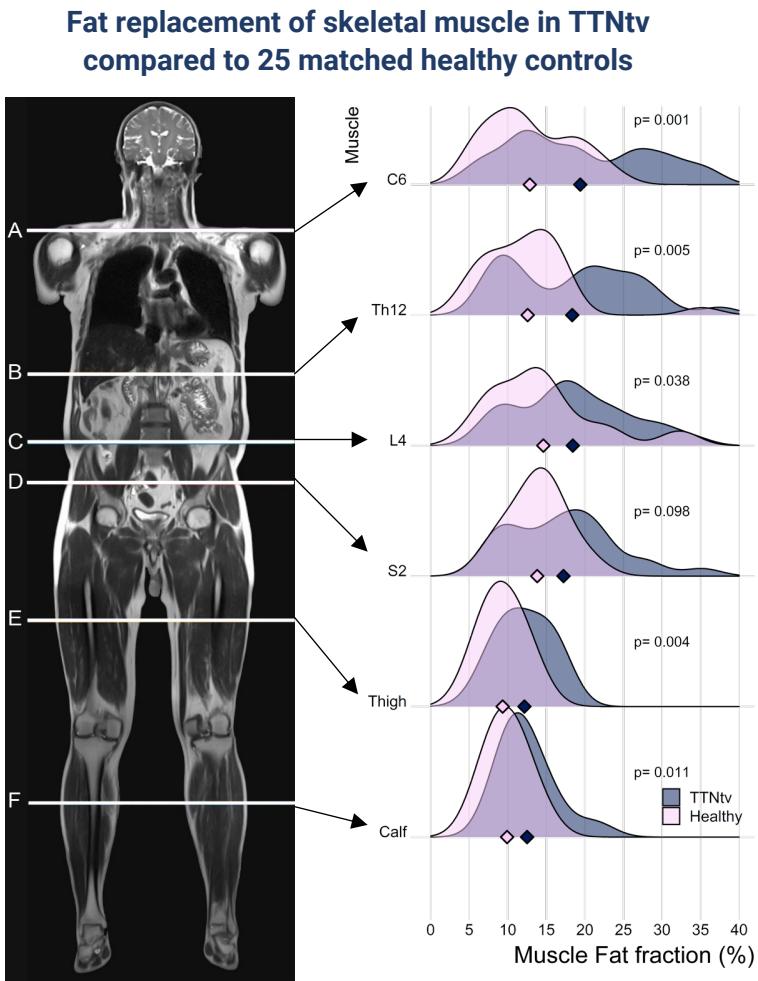
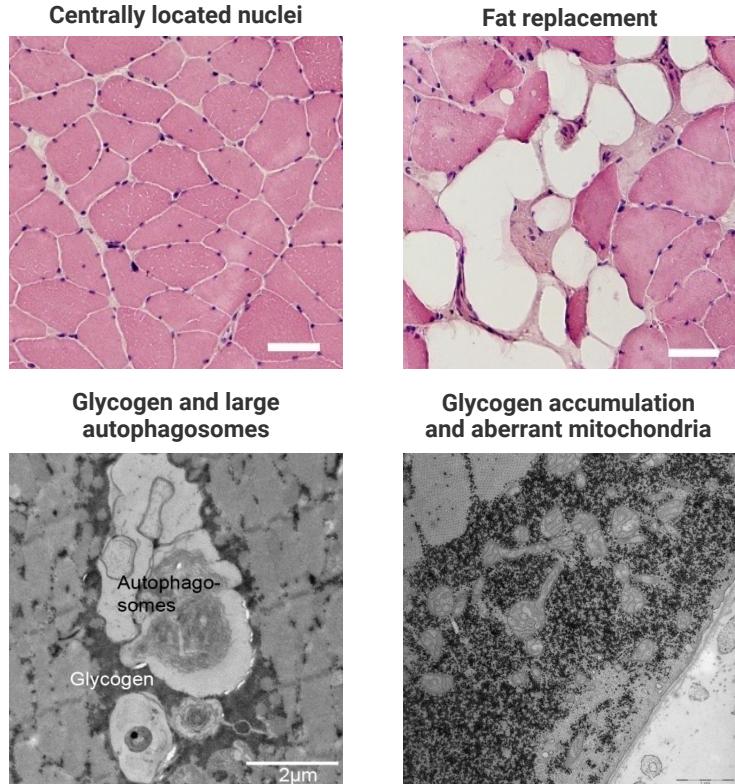


# CENTRAL ILLUSTRATION: Skeletal muscle phenotype in patients with truncating titin variants and familial dilated cardiomyopathy

- 25 individuals with TTNTv
- Fat fraction on muscle MRI
- Histology and ultrastructure on muscle biopsies



## Myopathic features and ultrastructural abnormalities from muscle biopsies



## Clinical message

Heterozygous TTNTv leads to affection of skeletal muscle. Specialized diagnostic work-up should be considered in patients with symptoms or objective findings of skeletal muscle involvement

# 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)

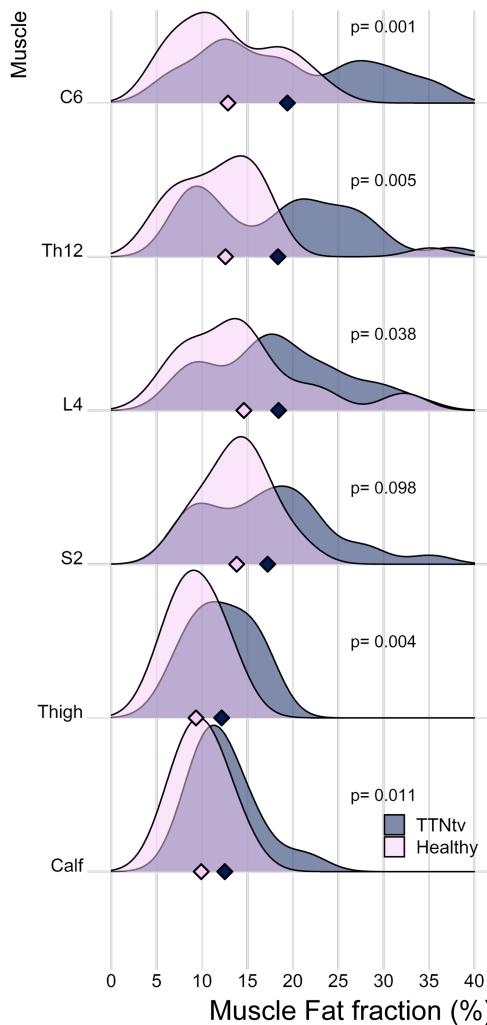


Fat fraction on  
muscle MRI  
(compared to 25 healthy  
controls and 7 controls with  
non-TTNtv genetic DCM)

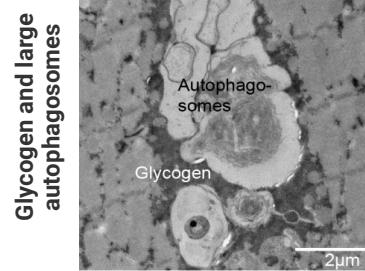
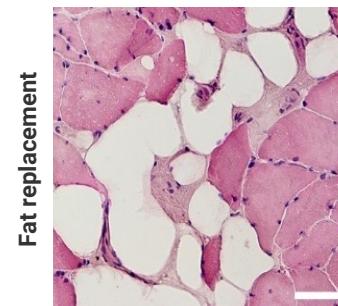
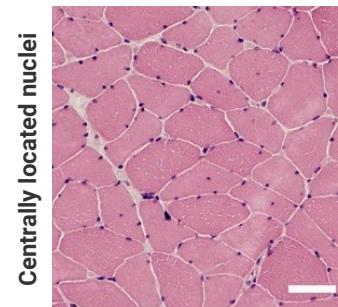


Histology and  
ultrastructure on  
muscle biopsies

## Fat replacement of skeletal muscle



## Muscle biopsy findings



## Clinical message

TTNtv

↓  
Skeletal muscle  
affection

↓  
Skeletal  
muscle fat  
infiltration

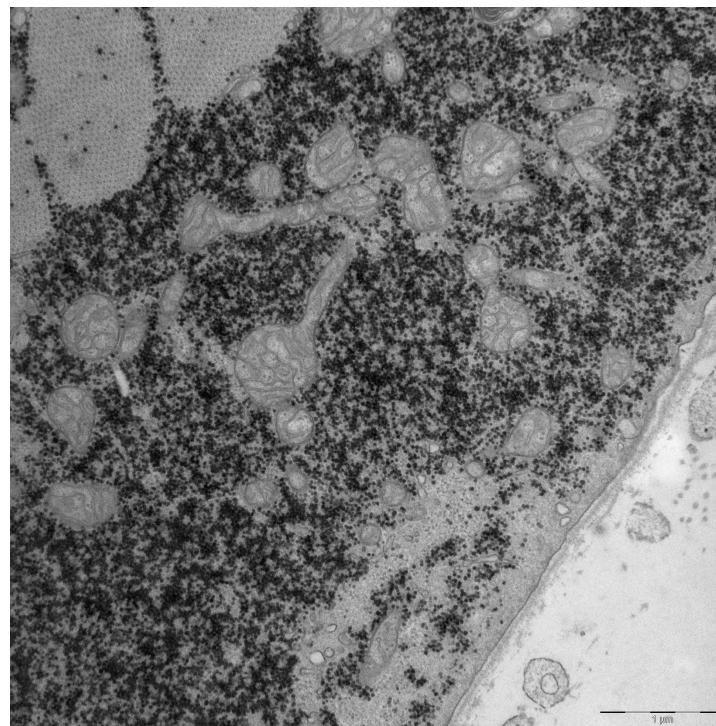
↓  
Myopathic  
findings

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

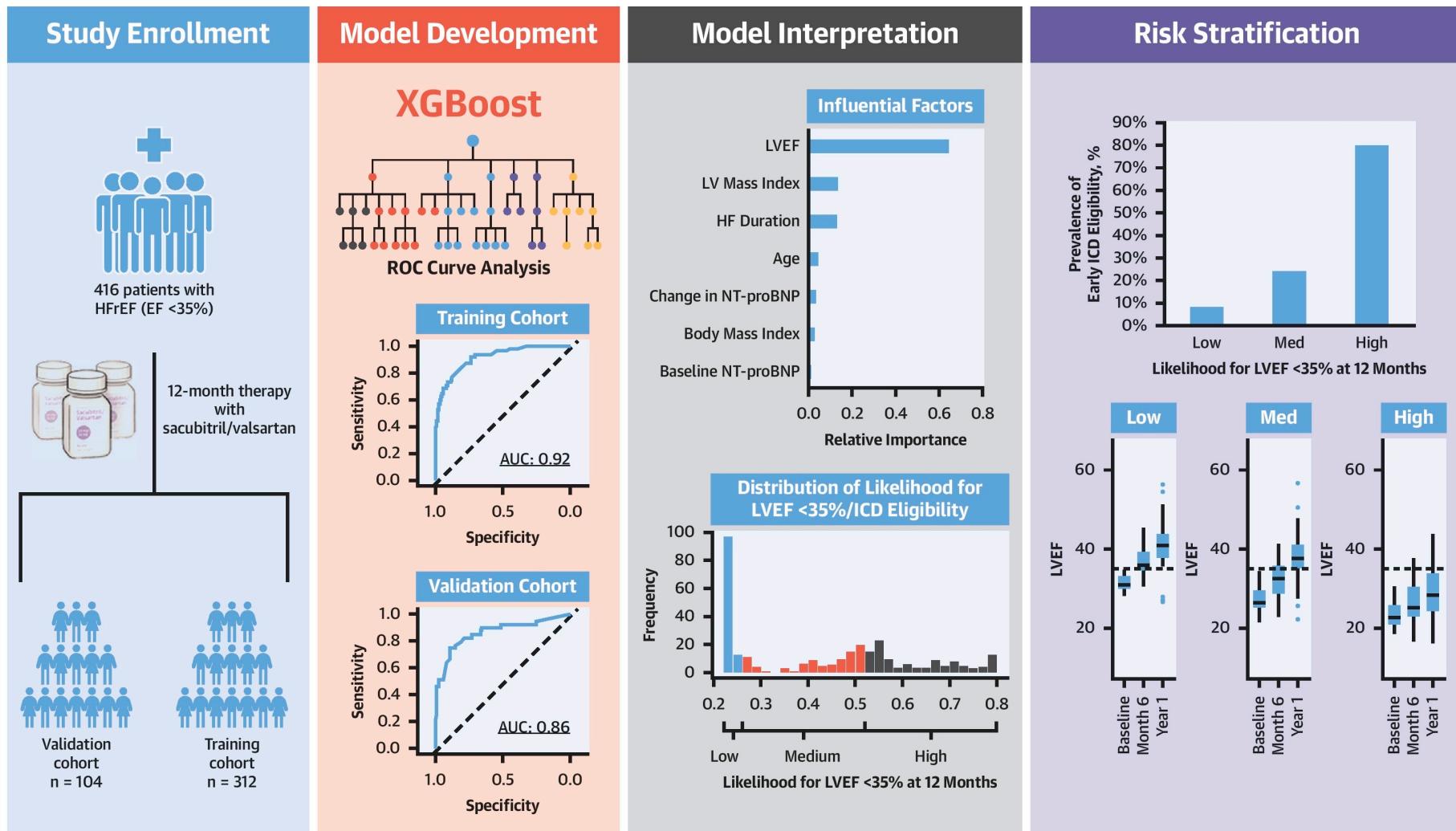
# TEM



LVEF  $45\pm10\%$   
GLS  $-14\pm4\%$



# CENTRAL ILLUSTRATION: Summary of Study Design, Model Development, Interpretation, and Performance



Mohebi R, et al. J Am Coll Cardiol HF. 2023;11(1):44-54.

