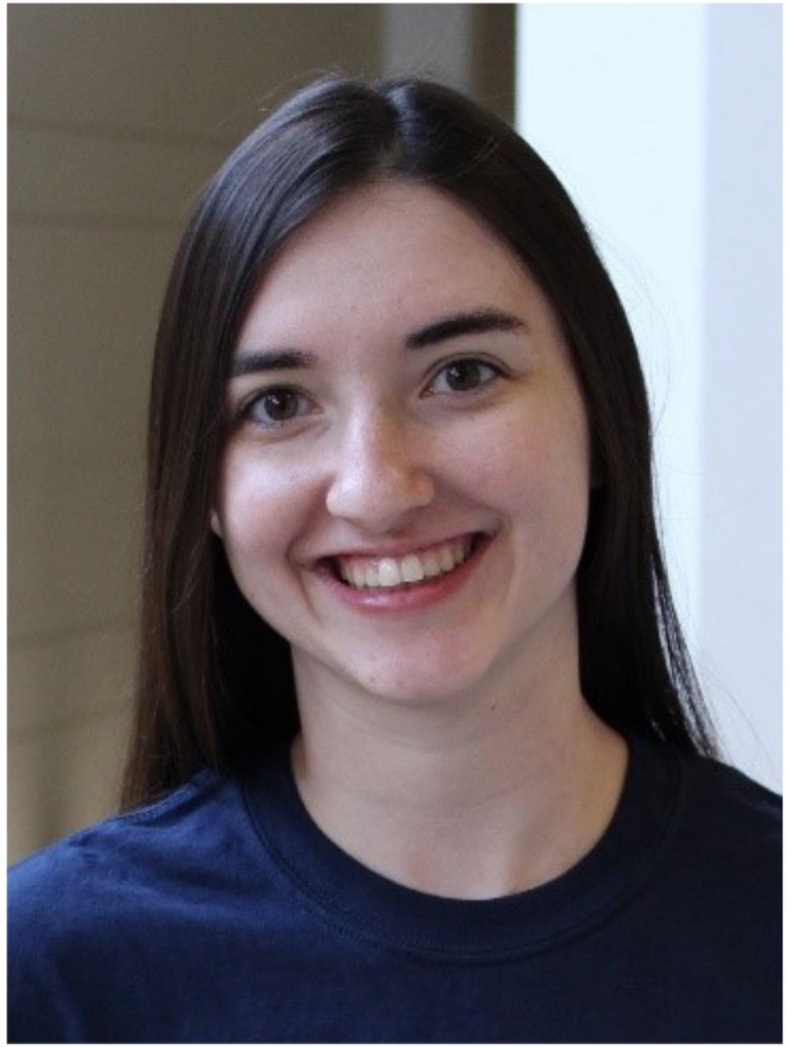

Katherine Koczwara
Lek Lab, Yale University

GNEM SYMPOSIUM SPEAKER SERIES

presented by The Neuromuscular Disease Foundation

Developing An AAV Gene Therapy For GNEM: Part II

Sunday, October 30th 2022 • 9am PT

Speaker Series #103022

Katherine Koczwara, Lek Lab, Yale University

Introduction & Summary Slide for:
"Developing An AAV Gene Therapy For GNEM: Part II"

1. Aim: develop an AAV gene replacement therapy for GNE myopathy

2. Method: optimize AAV transgene cassette *in vitro* before moving to *in vivo* AAV mouse biodistribution studies

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FINAL HIGHLIGHTS:

"Developing An AAV Gene Therapy For GNEM: Part II"

Key Takeaways

1. Optimization of AAV vectors in vitro indicate the CK8e-SV40-GNE1-V5 construct results in the high GNE expression in myoblasts
2. Preliminary results in wildtype mice show dose-dependent, muscle-specific

GNE expression using AAV9-CK8e-SV40-GNE1-V5 and AAV9-MCK-hGNE1

3. Future work and ongoing work will aim to determine GNE protein expression and gene therapy efficacy