



COLLEGE OF PHARMACY  
**PHARMACEUTICAL SCIENCES**  
UNIVERSITY OF MICHIGAN

## **Pharmaceutical Sciences Seminar**

Wednesday, December 7, 2022

4:00pm

2548 NUB or [Zoom](#)

### **“Respiratory Directed Gene Therapy for Pompe Disease”**

Presented by:



#### **Mai ElMallah, MD MS**

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Associate Director, Duke Cystic Fibrosis Center  
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**Abstract:** Pompe disease is an autosomal recessive neuromuscular disease caused by a deficiency of acid alpha-glucosidase (GAA), an enzyme that hydrolyzes lysosomal glycogen. Glycogen accumulation in the muscles of respiration, the motor neurons and the airways results in significant respiratory insufficiency. Recent work from our laboratory also shows that there is significant distal airway pathology in Pompe disease with a disruption in the alveolar type II cells. The only FDA approved therapy for Pompe disease is enzyme replacement therapy (ERT) which does not completely correct the respiratory pathology. My research is focused on using adeno-associated viral gene therapy to target and treat respiratory insufficiency in Pompe disease.

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