

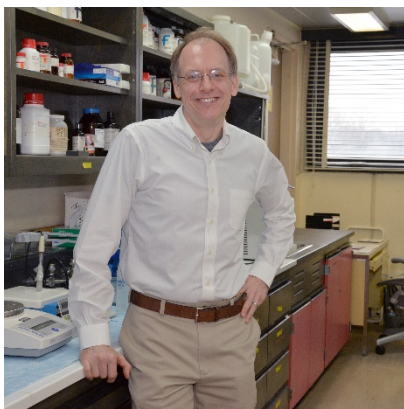


Yale SCHOOL OF MEDICINE

GENETICS DEPARTMENT SEMINAR SERIES

Rare Human Diseases: Ogden syndrome and the amino-terminal acetylation of proteins

The Lyon laboratory focuses on the discovery of new and/or underexplored rare human diseases, such as Ogden, NAA15, TAF1 and KBG syndromes. The laboratory discovered and characterized the first genetic disease involving amino-terminal acetylation of proteins, with a missense mutation in the X-linked gene NAA10. We named this rare disease Ogden syndrome (OS) in honor of the hometown (Ogden, Utah), where the first family we identified with OS lived. The affected boys have a distinct combination of craniofacial anomalies, hypotonia, global developmental delays, cryptorchidism, cardiac anomalies, and cardiomegaly. We and others then found more than 50 families with overlapping phenotypes with additional mutations in NAA10 in this pathway; we also reported that de novo truncating or missense mutations in NAA15, encoding a binding partner for NAA10, are involved in congenital heart defects and/or neurodevelopment. Over the past few years, we have established various mouse models for OS, along with the characterization of cardiomyocytes derived from human induced pluripotent stem cell (iPSCs), as part of our long-term goal to understand the mechanistic basis of OS.



Dr. Gholson Lyon, MD, PhD

Principal Investigator

New York State Institute for Basic Research
in Developmental Disabilities

Host: Syndi Barish

Graduate Student, Brueckner Lab
YSM Department of Genetics

Tuesday, May 11, 2021

11:30am - 12:30pm

Zoom Link

pw: 7852649

The Genetics Calendar of Events can be viewed on-line at
<https://medicine.yale.edu/genetics/events/seminars.aspx>