

## Discovery and follow-up studies of an X-linked infantile lethal disorder caused by decreased amino-terminal acetylation of proteins

**Gholson J. Lyon**

Cold Spring Harbor Laboratory, USA

We previously identified the genetic basis of a previously unrecognized X-linked, infantile lethal Mendelian disorder. This is the first human genetic disorder identified that results from a hypomorphic allele in the N-terminal acetylation pathway, with Nt-acetylation being one of the most common protein modifications in humans. The functional implications of Nt-acetylation for the thousands of proteins that are modified by it remain quite elusive, although recent data suggest a role as a destabilization signal for proteins and involvement in the secretory pathway. To begin our long-term study of this disease, we are utilizing several different cellular model systems and model organisms. We are also performing X-inactivation studies in the female carriers, along with further characterizing the pathophysiology and phenotype of this disease. I will present some results in regards to this understudied protein modification, now for the first time implicated in a human disease.

### Biography

Gholson Lyon, M.D. Ph.D., is an assistant professor in human genetics at Cold Spring Harbor Laboratory and a research scientist at the Utah Foundation for Biomedical Research. He is also a board-certified child, adolescent and adult psychiatrist. In addition to his research on the genetics of neuropsychiatric illnesses. Dr. Lyon is focusing on the genetic basis of rare Mendelian diseases.

[gholsonjlyon@gmail.com](mailto:gholsonjlyon@gmail.com)