

**Abstract Title:** Exploring the possible mechanisms of pathogenesis in GNE Myopathy

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**Abstract:** GNE myopathy (GNEM) is one of many rare genetic disorders with neuro-muscular defects. Though the overall number is low, GNE myopathy patients are found all over the world. Due to its rare occurrence and the nature of clinical presentation that is similar to many diseases, a large fraction of patients has remained undiagnosed or misdiagnosed for many years. Due to the advent and improvement of DNA sequencing technology it has become comparatively easier to diagnose this and other genetic disorders. As a result, there has been a spurt in the number of newly diagnosed patients. International efforts to understand the molecular basis of GNEM and develop treatment options for patients have been limited to a few labs. Within India, this effort has been miniscule. The GNE encodes a bifunctional enzyme that catalyses a rate-limiting step in the sialic acid biosynthesis. Though, the possible mechanisms of skeletal muscle damage is not yet known, two possible hypotheses, hypo sialylation and loss of protein interactions have been proposed. Moreover, the molecular mechanism of late onset, disease progression and tissue specificity (skeletal muscle) are still not clear.

In this talk, the summary of our current understanding of the patho-mechanisms of the diseases, the current estimates of the number of patients, and suggest possible therapeutic possibilities based on the studies carried out around the world and by groups at the Jawaharlal Nehru University.

**Area of expertise:** Molecular Biology, Cell biology, Bioinformatics, Disease biology, Genomics