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NDF collaborates with PerkinElmer for rare disease research

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In collaboration with PerkinElmer, NDF aims to use sequencing and metabolomics to study 100 patient samples.

US based Neuromuscular Disease Foundation (NDF) is launching an alliance with PerkinElmer to use whole-genome sequencing to investigate the molecular pathways involved in GNE myopathy, a progressive muscle-wasting disease.

It is a rare autosomal recessive condition in which a genetic mutation prevents the generation of sialic acid in cells, leading to muscle dysfunction and degeneration.

In collaboration with PerkinElmer, NDF aims to use sequencing and metabolomics to study 100 patient samples. The partners will also collect their findings into a database to help with patient recruitment for clinical trials.

PerkinElmer has been working to expand its clinical genomic testing services in recent months, offering whole-exome and whole-genome sequencing for genetic disease diagnostics in newborns and partnering with advocacy groups and foundations focused on specific diseases.

This is one of the first such known efforts made to understand the etiology of a rare disease at the molecular and biochemical level.