Speaker Series #52220

Dr. Marjan Huizing, NHGRI/NIH

Introduction & Summary Slide for:

"NIH Research Studies for GNE Myopathy"

An Overview of what to expect in this talk:

1. NHGRI receives funding from NDF to perform selected research studies for GNE myopathy.

2. Dr. Huizing will be talking about (pre-)clinical research studies for GNE myopathy ongoing at NHGRI.

3. These studies include genetic studies, biomarker studies and ManNAc -therapy related studies.

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Key Takeaways:

1. Diagnostic Challenges of GNE Myopathy

Accomplished:

- Increased awareness of GNE-M
- Unified name and mutation nomenclature
- GNE gene inclusion in neurologic disease screening panels

Future:

- Document all reported GNE mutations and GNE-M cases

2. Biomarker Discovery for GNE Myopathy

Accomplished:

- Muscle Lectin Staining
- White Blood Cell SA and CMP-SA levels

Future:

- Blood biomarker based on protein sialylation status

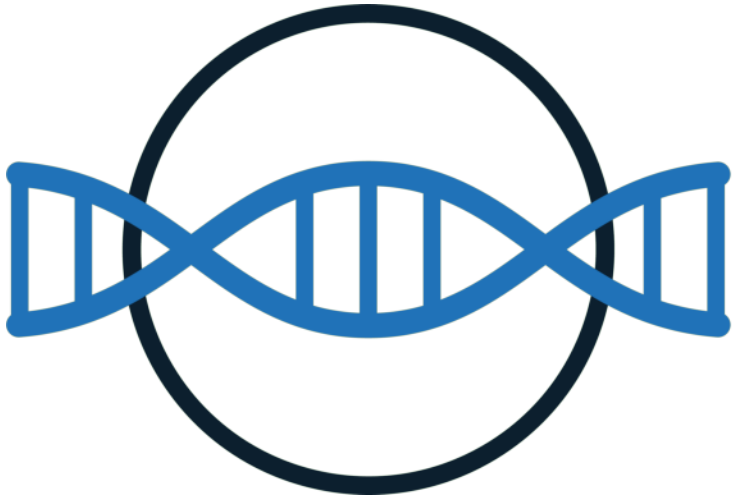
3. NIH Trials for GNE Myopathy

Accomplished:

- Natural History study
- ManNAc Phase 1 and Phase 2 studies
- Established outcome measures for future GNE-M clinical trials

Future:

- Pivotal study for ManNAc in GNE-M
- Validate outcome measures for GNE-M clinical trials
- Establish and maintain disease models to support future therapy development, study disease mechanism and study disease modifiers



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