Penn Medicine

Neuromuscular Variant Resolution Clinic

Penn Medicine is proud to offer a first of its kind clinic dedicated to interpretation of genetic variants of uncertain significance (VUS) for adult patients with suspected neuromuscular diseases.

Variant Review

Our highly trained team will review all variants following ACMG/AMP guidelines utilizing the most up-to-date medical literature and genomic databases.

Deep Phenotyping

Detailed clinical and family history information will be gathered for each patient to improve variant interpretation. This may include a thorough physical exam, RNA studies, muscle biopsy, electrodiagnostic studies, biochemical studies, and/or imaging.

Genetic Counseling

All patients will be provided with thorough genetic counseling to ensure understanding of their genetic testing results. The genetic counselor may also assist with family member testing, locating support services, and identification of clinical trials.

SCHEDULE AN APPOINTMENT

For more information or to refer a patient, please email kelsey.johnson1@pennmedicine.upenn.edu or call (215) 839-6002.

Program Features

- Open to any patient with neuromuscular symptoms and a variant of uncertain significance (VUS) on genetic testing. Priority given to genes/conditions with current or upcoming clinical trials.
- Consult service with results returned to referring provider.
- In-person or telemedicine visits (PA and NJ).

MEET OUR PROVIDERS

Kelsey Johnson, MS, LCGC
Kelsey is a licensed, certified genetic counselor (LCGC), with experience in the neuromuscular field and variant interpretation.

Paul McIntosh, MD
Dr. McIntosh is an Assistant Professor of Clinical Neurology.

The Neuromuscular Variant Resolution Clinic is supported by the generous donation of Sarepta Therapeutics.

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