Genetic Testing in Neuromuscular Disease: Sponsored Testing Programs (as of April 2023)*

A Resource for Clinicians

Background

- Several genetic tests are available through commercially-sponsored programs.
- Those listed here are up-to-date as of April 2023 and will be updated on an ongoing basis. Click on the links to learn more.
- Information in this document was developed as part of an MDA-hosted mini-webinar with a genetic counselor. View the companion mini-webinar here.

Sponsored Programs (as of April 2023)*

Muscular Dystrophy/Myopathy

Invitae Detect Muscular Dystrophy Program[1]

Test options

- Dystrophinopathies test (DMD only)
- Limb-girdle muscular dystrophy panel
- Comprehensive muscular dystrophy panel
- Comprehensive neuromuscular disorders panel

Lantern Project[2]

Focused Neuromuscular Disease Panel

- Note: includes PABPN1 analysis for detection of OPMD

Decode Duchenne[3]

Diagnostic testing for suspected DMD/BMD

- Serum CK analysis
- Genetic testing
- RNA sequencing
  - For patients with no identifiable mutation but abnormal muscle biopsy
  - Requires muscle

Carrier testing

- Offered for all at-risk female family members or males with confirmed dystrophinopathy

BMD, Becker muscular dystrophy; CK, creatine kinase; DMD, Duchenne muscular dystrophy; OPMD, oculopharyngeal muscular dystrophy; PABPN1, polyadenylate-binding protein nuclear 1.

1. INVITAE. https://www.invitae.com/en/sponsored-testing/detect-mdys. 2. PerkinElmer Genomics. https://www.perkinelmergenomics.com/lanternproject/ 3. Parent Project Muscular Dystrophy. https://www.parentprojectmd.org/about-duchenne/decode-duchenne/carrier-testing/

***Each program has specific eligibility criteria.**
Be sure to check program criteria for patient eligibility before ordering

Developed with the expertise of Jordan Bontrager, MS, CGC, University of Rochester Medical Center
Last Reviewed: April 2023

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Neuropathy

Invitae Alnylam Act hATTR Amyloidosis[1]

Only available to patients aged ≥18 years

Test options

- Cardiomyopathy comprehensive panel
- Comprehensive neuropathies panel
- ATTR amyloidosis test

Ambry hATTR Compass Genetic Testing[2]

Test options

- ATTR amyloidosis test
- CardioNext panel
- NeuropathySelect panel

Spinal Muscular Atrophy

Invitae SMA Identified[3]

ALS

Invitae ALS Identified[4]

Includes C9orf72 and ALS panel

PreventionGenetics ALS Testing Program[5]

Includes analysis of ATXN2 as well as C9orf72 and ALS panel

ALS, amyotrophic lateral sclerosis; ATTR amyloidosis, transthyretin-mediated amyloidosis; hATTR, hereditary ATTR; SMA, spinal muscular atrophy.

1. INVITAE. https://www.invitae.com/en/alnylam-act-hattr-amyloidosis. 2. Ambry Genetics. https://www.ambrygen.com/partners/hattr-compass/healthcare-provider. 3. INVITAE.. https://www.invitae.com/en/sponsored-testing/sma-identified. 4. INVITAE.. https://www.invitae.com/en/sponsored-testing/als-identified. 5. Prevention Genetics.. https://www.preventiongenetics.com/sponsoredTesting/Ionis_ALS.

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Mitochondrial Disease

UMDF Genetic Testing for Suspected Mitochondrial Disease Program[1]

Nuclear mitochondrial panel + mtDNA analysis

Patient must have no insurance or insurance that will not cover costs for testing

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PreventionGenetics No-Cost Genetic Testing Program for TK2 Deficiency[2]

Lysosomal Storage Diseases

Lantern Project[3]

Test options

- Fabry disease
- Pompe disease
- Gaucher disease
- Niemann-Pick types A and B
- Mucopolysaccharidosis I (MPSI) and other MPS disorders

Periodic Paralysis

Invitae Uncovering Periodic Paralysis[4]

MPS, mucopolysaccharidosis; mtDNA, mitochondrial DNA; TK2, thymidine kinase 2; UMDF, United Mitochondrial Disease Foundation.

1. United Mitochondrial Disease Foundation (UMDF). https://www.umdf.org/genetic-testing-clinicians. 2. Prevention Genetics. https://www.preventiongenetics.com/sponsoredTesting/Zogenix. 3. PerkinElmer Genetics. https://www.perkinelmergenomics.com/lanternproject/. 4. INVITAE. https://www.invitae.com/en/uncoveringperiodicparalysis/.

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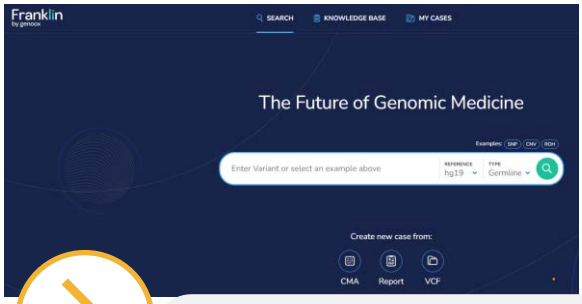
Additional Resources on Genetic Testing

Find a genetic counselor in your area

- National Society of Genetic Counselors



Access the Franklin variant assessment tool

- Open professional genomic community
- Search engine helps obtain insights from real-world data

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