MDA Muscular Dystrophy Association

Ask a Genetic Counselor: Leveraging Genetic Testing to Optimize NMD Care

Medical Education Team

October 28, 2024

Meet Our Panelists

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Who are we and why are we here?

Not everything happens the same way across institutions

- Each clinic is unique, with its own processes and framework for care
- Different staffing, resources, and provider roles

Potential missteps

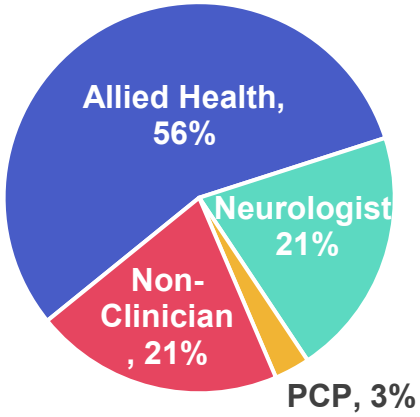
- *We could have helped/tested sooner*
- *Right question, wrong test*
- *Right test, wrong patient profile*

Forging a path: Logistics to consider

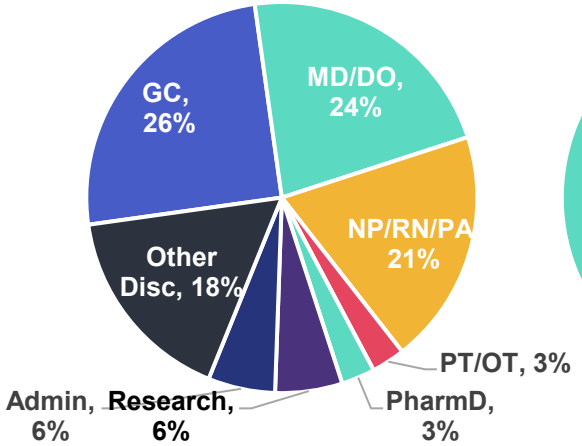
- *How do access a test for […]*
- *When do I involve a genetic counselor in [...]*

Our attendees: Pre-webinar survey results*

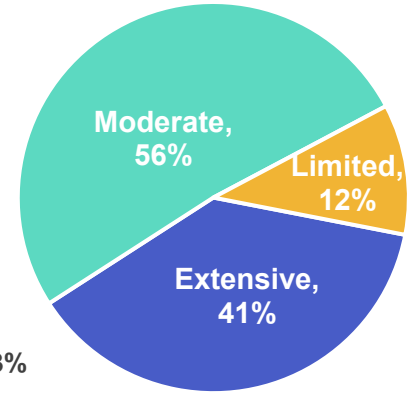
Specialty



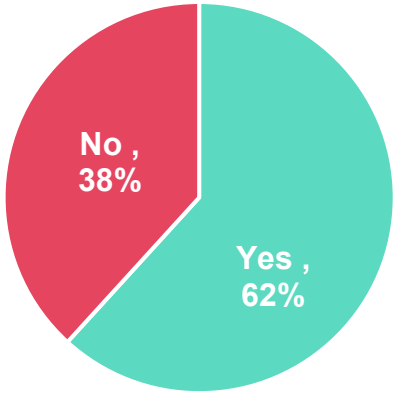
Role/Degree



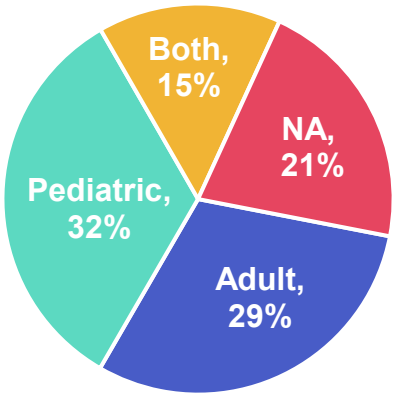
NMD Experience



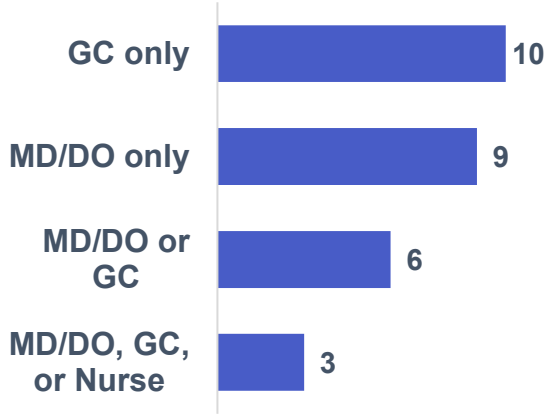
MDA-Affiliated



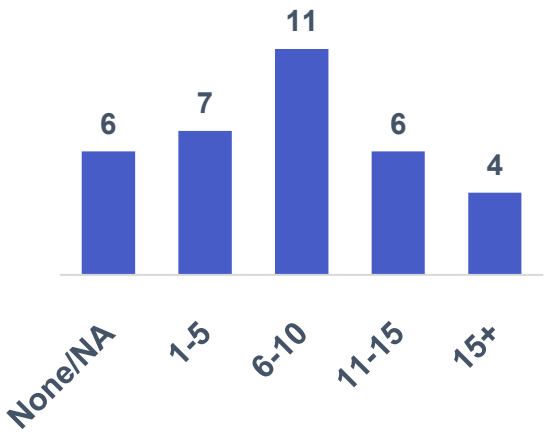
Patient Population



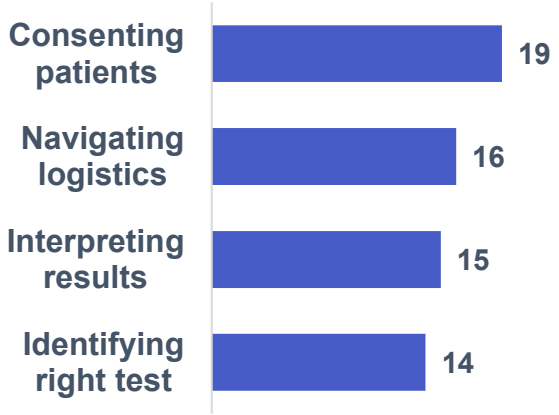
Who orders tests



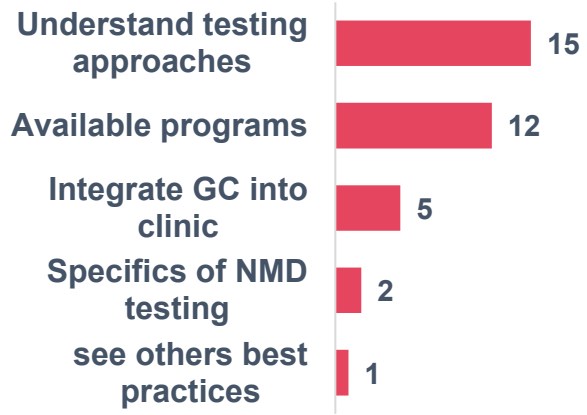
Tests per month



Most comfortable



Reason for joining



*Total registrants as of 10/25/24 (n=34)

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Agenda

Today

- Relevance & Role of Genetic Testing in NMD
- Perspectives: Integrating Genetic Testing & Counseling Into NMD Care
- Types of Genetic Tests & Relative Pros & Cons
- Factors to Consider When Ordering a Genetic Test

Coming soon…

Ask a Genetic Counselor Workshops

- *Choosing the Right Test*
- *Navigating Billing*
- *Interpreting Test Results*
- *Variants of Unknown Significance*

Learning Objectives

Following this program, attendees will improve their ability to:

- Identify patients with NMD who may benefit from genetic testing
- Adopt best practices for integrating genetic counseling and testing into care (eg, "right test, for the right patient, at the right time")
- Leverage available genetic testing programs and resources in NMD to increase patient access and support referral to relevant specialists

Relevance & Role of Genetic Testing in Neuromuscular Disease (NMD)

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Genetics of Neuromuscular Disease

Neuromuscular disorders (NMDs) consist of a genetically and phenotypically heterogenous group of diseases that can disrupt skeletal muscle, neuromuscular junction, or nerves[1]

NMDs AFFECT 1 IN 1000 INDIVIDUALS WORLDWIDE[2]

Inheritance patterns for NMDs vary by disease or gene[3-6]

Autosomal dominant

A single copy of the mutated gene can cause the disorder

Examples
Myotonic dystrophy, OPMD, CMT1A

Autosomal recessive

Two copies of the mutated gene are required to cause the disorder

Examples
SMA, Pompe disease, sarcoglycanopathies

X-linked inheritance

Mutated gene is located on the X chromosome; males often more severely affected than females

Examples
Dystrophinopathies, CMT1X

Mitochondrial Inheritance

Only maternal genetic material passed from parent to offspring

Example
MELAS

CMT1A, Charcot-Marie-Tooth disease type 1A; CMT1X, X-linked Charcot-Marie-Tooth disease; OPMD, oculopharyngeal muscular dystrophy; SMA, spinal muscular atrophy.

1. Ng KWP, et al. *Front Neurol*. 2022;13:997551. 2. Zatz M, et al. *Genet Mol Biol*. 2016;39(3):339-348. 3. National Human Genome Research Institute. https://www.genome.gov/genetics-glossary/Autosomal-Dominant-Disorder. 4. National Human Genome Research Institute. https://www.genome.gov/genetics-glossary/Autosomal-Recessive-Disorder. 5. National Human Genome Research Institute. https://www.genome.gov/genetics-glossary/X-Linked. 6.Genetic Alliance; District of Columbia Department of Health. Understanding Genetics: A District of Columbia Guide for Patients and Health Professionals. Washington (DC): Genetic Alliance; February 17, 2010. https://www.ncbi.nlm.nih.gov/books/NBK132149/

Identifying Hereditary Disease in NMD

1 Consider if patient's symptoms have a genetic origin: detailed history

Ask questions related to early milestones and birth/prenatal history. Features that may suggest a hereditary process:

- Longstanding or slowly progressive deficits
- Multisystem disease
- Normal workup for acquired disease causes

- Early onset of disease
- Similar symptoms in other family members

2 Localize disease process (e.g., muscle, motor neuron, etc.)

Localizing disease can help facilitate the workup for genetic testing

NMD, neuromuscular disease.

Barp A, et al. *Diagnostics (Basel)*. 2021;11(4):701.

Benefits of Genetic Testing[1,2]

Why does it matter?

- Genetic testing may help identify a genetic cause for the patient's symptoms, help determine the risk of developing a genetic disorder, and/or confirm a definitive diagnosis
- May allow patients to avoid unnecessary or invasive tests, such as muscle or nerve biopsies, and avoid unnecessary, potentially harmful treatments (e.g. immmunosuppresives)
- Diagnosis with genetic testing can allow for determining appropriate treatment, management, and identify other at-risk organ systems to guide proper management (e.g. cardiac or pulmonary surveillance)
- May provide improved access to clinical trials and emerging treatments
- Knowledge of disease can aid in family planning, testing at-risk family members, and assessing reproductive risk

National Library of Medicine. MedlinePlus. Genetic Testing. https://medlineplus.gov/download/genetics/understanding/testing.pdf. 2. Ng KWP, et al. *Front Neurol*. 2022,13:997551.

What Is a Genetic Counselor?

"Genetic counselors have advanced training in medical genetics and counseling to guide and support patients seeking more information about how inherited diseases and conditions might affect them or their families, and to interpret genetic test results based on personal and family history."

–National Society of Genetic Counselors (NSGC)

- Genetic Counselors have a 2-year Master of Science degree in Genetic Counseling and must pass a board examination to become certified
- In addition to certification, most states require licensure to practice

National Society of Genetic Counselors. https://www.nsgc.org/

NSGC Status Survey 2024

Table 23. All Areas of Practice	N	%
Cancer genetics - adult	1,029	41%
Prenatal	627	25%
Pediatrics	583	23%
Preconception/Reproductive screening	479	19%
General adult genetics	450	18%
Cardiology	349	14%
Neurogenetics	307	12%
Genetic counselor training/education	300	12%
Cancer genetics - pediatric	280	11%
Laboratory sciences (molecular/cytogenetics/biochemical testing/variant science)	270	11%
Genomic medicine	221	9%
Neuromuscular	166	7%
Metabolic disease	164	6%
Preimplantation genetic testing, ART/IVF, infertility	158	6%
Hematology	133	5%
Newborn Screening	131	5%
Utilization management	129	5%
Ophthalmology	121	5%
Audiology/Hearing loss	116	5%
Pharmacogenetics	94	4%
Nephrology	91	4%
Endocrinology	86	3%
Immunology	62	2%
Consumer genomics/Personal genomics	45	2%
Public health	37	1%
Psychiatric	25	1%
Other practice area	149	6%
No specific area of practice	83	3%
Total	**2,540**	**100%**

National Society of Genetic Counselors. (2024) *2024 Professional Status Survey*. *https://www.nsgc.org/Policy-Research-and-Publications/Professional-Status-Survey*

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Panel: Perspectives

Integrating genetic testing into the care framework

- What are different ways that genetic counselors work with clinicians at different timepoints?
- What challenges have you encountered among clinics trying to integrate counseling into care framework?

To find a genetic counselor in your area, consult the National Society of Genetic Counselors: https://findageneticcounselor.nsgc.org/

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Types of Genetic Tests & Associated Pros & Cons

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Types of Genetic Tests

Single-site (targeted) testing[1]

Investigate for presence or absence of known familial variant

Single-gene testing[2]

Analysis of single gene when disease with one known genetic cause highly suspected

Cytogenetic testing[3]

Focused on detecting chromosomal abnormalities

Next-Generation Sequencing[1,4]

- Panel testing:
  Looks for changes in a predefined set of genes in a single test
- Whole exome/genome testing:
  Analyzes a large bulk of an individual's DNA to locate genetic variations

1. National Library of Medicine. MedlinePlus. Genetic Testing. https://medlineplus.gov/download/genetics/understanding/testing.pdf. 2. Orengo JP, Murdock DR. Genetic testing in neuromuscular disorders. July/August 2019. https://practicalneurology.com/articles/2019-aug-july/genetic-testing-in-neuromuscular-disorders. 3. Berisha SZ, et al. *Birth Defects Res*. 2020;112:293-306.
4. Ng KWP, et al. *Front Neurol*. 2022,13:997551.

Types of Genetic Tests: Single-Site Testing

- Used when there is a family history of genetically confirmed disease
- Most sensitive testing option
- Least possibility of uncertain or unanticipated findings
- Prenatal testing/PGD

Need confirmation of familial mutation

- Preferred: family member's test report
- Acceptable: verbal or written confirmation of genetic alteration nomenclature
- Can be difficult to obtain, depending on family dynamics

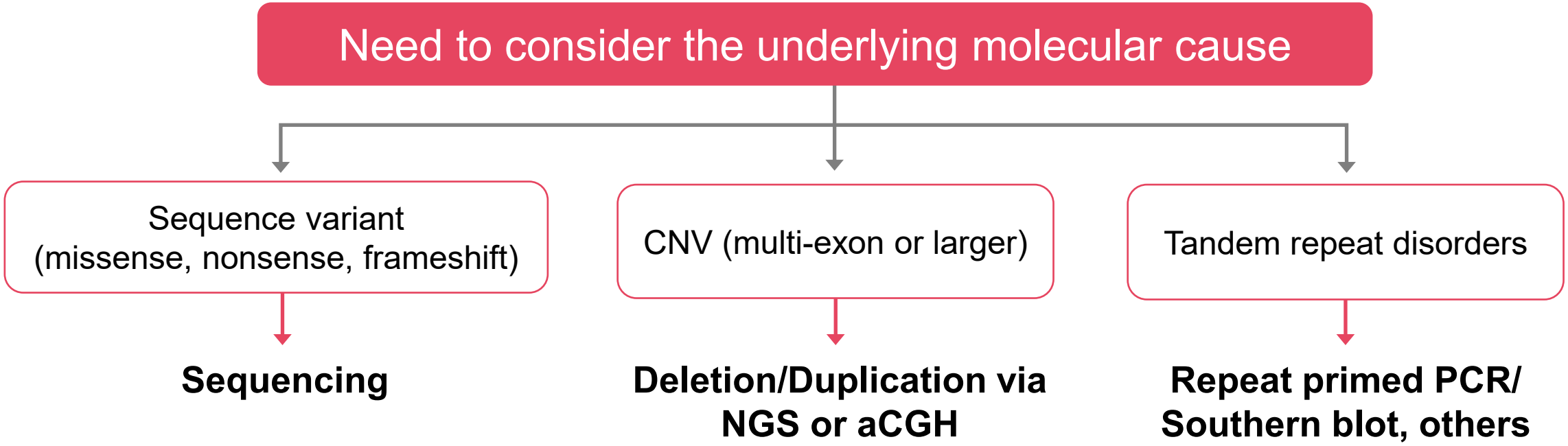
Best to test at lab where proband tested

PGD, preimplantation genetic diagnosis.

MedlinePlus. What are the different types of genetic tests? https://medlineplus.gov/genetics/understanding/testing/types/.

Types of Genetic Tests: Single-Gene Testing[1,2]

Used when diagnosis with one known genetic cause is highly suspected



Examples: DMD analysis, PMP22 deletion/duplication testing for CMT1A, CNBP repeat analysis for DM2, TTR sequencing + deletion/duplication for hATTR

CMT1A, Charcot-Marie-Tooth disease type 1A; CNBP, CCHC-type zinc finger nucleic acid-binding protein; CNV, copy number variant; DM2, myotonic dystrophy type 2; DMD, Duchenne muscular dystrophy; hATTR, hereditary transthyretin amyloidosis; HD; Huntington disease; HTT, Huntingtin; PCR, polymerase chain reaction; PMP22, peripheral myelin protein 22; TTR, transthyretin.

1. Lalonde E, et al. *Front Pediatr*. 2020;8:373. 2. Barp A, et al. *Diagnostics (Basel)*. 2021;11(4):701.

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Types of Genetic Tests: Cytogenetic Testing[1,2]

Cytogenetics is focused on the detection of chromosomal abnormalities

Karyotyping

Visual representation of chromosomes

Can detect extra or missing chromosomes and/or structural differences

Fluorescence in situ hybridization (FISH)

Detects chromosome abnormalities targeting specific genetic sequences using fluorescently labeled probes that attach to complementary DNA

Chromosomal microarray analysis (CMA)

Detects missing or extra portions of a chromosome, which may include one or multiple genes

Genome-wide detection of gross deletions, duplications, and rearrangements

More commonly used for pediatric and complex phenotypes:

CMA is recommended first-tier test for patients with developmental delay, intellectual disability, ASD, and/or multiple congenital anomalies

ASD, autism spectrum disorder.

1. Berisha SZ, et al. *Birth Defects Res*. 2020;112:293-306. 2. Ozkan E, Lacerda MP. Genetics, cytogenetic testing and conventional karyotype. In: StatPearls. Treasure Island, FL: StatPearls Publishing; August 8, 2022. https://www.ncbi.nlm.nih.gov/books/NBK563293.

Types of Genetic Tests: Panel Testing[1,2]

This type of lab test is often used to pinpoint a diagnosis when a person has symptoms that may fit a wide array of conditions, or when the suspected condition can be caused by variants in many genes

Multiple genes sequenced at the same time

Gene panels vary in size depending on the phenotype being tested (e.g., from 2 genes for SMA to 230 genes for a comprehensive neuromuscular panel)

Genes usually are grouped together based on producing a similar phenotype when mutated

Phenotype groupings may be quite targeted (e.g., non-dystrophic myotonia panel), or broad (e.g., large neuromuscular disease panel)

Run on next-generation sequencing platform, which typically includes **sequencing and deletion/duplication of exonic regions and a small portion of intron (usually +/– 10 base pairs)**

ASD, autism spectrum disorder; SMA, spinal muscular atrophy.

1. Ng KWP, et al. *Front Neurol*. 2022;13:997551. 2. Barp A, et al. *Diagnostics (Basel)*. 2021;11(4):701.

Types of Genetic Tests: Panel Testing

Panel Selection

Choose

Accredited lab

- CAP/CLIA certification
- Choose a laboratory with a history of testing in that disease area
  - More likely to be optimized for clinical sensitivity
  - Fewer VUS

Capture

Genes of interest

- Does the panel capture all high-priority genes?
- Consider limitations of NGS panel

Logistics

Logistical considerations

- Sample type
- Patient location
- TAT

Consider

Is bigger always better?

- Comfort level of the patient with uncertain results
- Often will need parental (or other familial) studies to clarify findings
- Larger panels = More returned variants

Cost

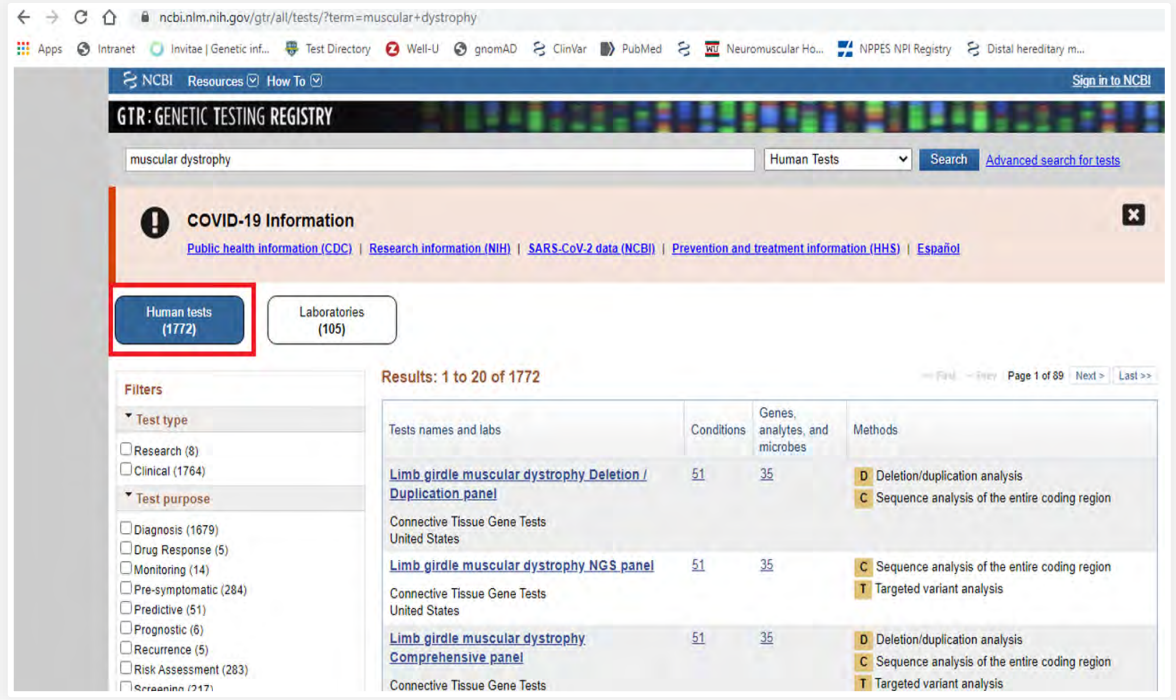
- Check patient's insurance coverage and test turnaround time
- May want to consider sponsored testing options, if available

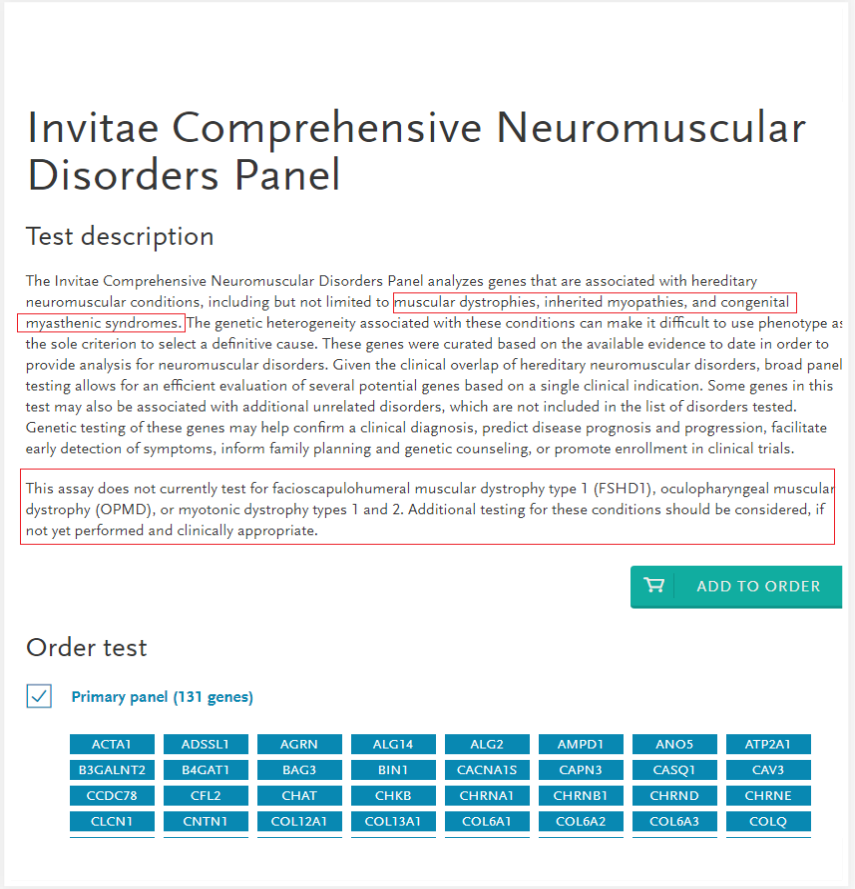
NGS, next-generation sequencing; VUS, variant of unknown significance.

Ng KWP, et al. *Front Neurol*. 2022;13:997551.

Types of Genetic Tests: Panel Testing

Panel Selection[1,2] (cont.)





1. National Library of Medicine. Genetic testing registry. https://ncbi.nlm.nih.gov/gtr/all/tests/?term=muscular+dystrophy.
2. INVITAE. Invitae Comprehensive Neuromuscular Disorders Panel. https://www.invitae.com/en/providers/test-catalog/test-03280.

Types of Genetic Tests: Panel Testing

Utility of Panel Testing

Pros

- Less costly than traditional single-gene analysis and WES/WGS
- Designed to ensure high coverage of all regions of interest
  - Wide coverage to detect single nucleotide variants, small insertions/deletions
- Lower chance of incidental findings compared to WES/WGS
- Allows for testing of multiple conditions with phenotypic overlap
- Can detect mosaicism
- Faster turn-around-time than WES/WGS

Cons

- More genes= more VUS
- Less customizable
- Family member sample not often included in initial analysis
- Needs in-depth interpretation
- Results can be hard for patients and providers to interpret, given all reportable variants in genes analyzed included, regardless of phenotype

VUS, variant of unknown significance; WES, whole-exome sequencing; WGS, whole-genome sequencing.

1. Ng KWP, et al. *Front Neurol*. 2022;13:997551. 2. Barp A, et al. *Diagnostics (Basel)*. 2021;11(4):701.

Types of Genetic Tests: Whole Exome/Genome Sequencing[1–3]

Whole-Exome Sequencing

- Analysis limited to sequencing of the exonic/protein-coding regions of the genome
- May include mitochondrial DNA

Whole-Genome Sequencing

- Covers both coding (exonic) and noncoding (intronic) regions of the genomic DNA, including nuclear and mitochondrial DNA
- Can also detect CNVs and repeat expansion disorders**

- Detailed clinical information needed for proper variant filtering and interpretation
  - Phenotype-driven analysis
- Trio testing strongly recommended to inform segregation
- Increased likelihood of incidental findings
- In-depth consent process is necessary due to the complexities of testing and possible incidental results
- Highest yield in complex pediatric cases

CNV, copy number variation.

1. Berisha SZ, et al. *Birth Defects Res*. 2020;112:293-306. 2. Barp A, et al. *Diagnostics (Basel)*. 2021;11:701. 3. Lalonde E, et al. *Front Pediatr*. 2020;8:373.

Types of Genetic Tests: WES/WGS Consent and Special Considerations

High cost, with variable insurance approval policies

Incidental finding considerations

- Nonpaternity
- Consanguinity
- Medically actionable variants
  - Can typically opt-in or out
  - Insurance implications

Strongly recommended to involve a genetic counselor in the discussion of WES/WGS testing and consent

WES, whole-exome sequencing; WGS, whole-genome sequencing.

Types of Genetic Tests: Limitations of Panel/WES/WGS[1,2]

Might not include large CNV analysis
(particularly WES and NGS)

Does not include methylation studies
May be necessary for imprinted genes
(PWS, Angelman syndrome)

WES cannot detect expansions/ contractions in repeat regions
(DM1, DM2, FSHD, SCA, C9orf72, OPMD, etc. need separate assay)

Decreased coverage in genes with pseudogenes or homologous regions
(SORD, GBA, and others)

Does not provide expression data

WES/WGS interpretation limited by clinical information provided

C9orf72, chromosome 9 open reading frame 72; CNV, copy number variation; DM1, myotonic dystrophy type 1; DM2, myotonic dystrophy type 2; FSHD, facioscapulohumeral dystrophy; GBA, glucocerebrosidase; OPMD, oculopharyngeal muscular dystrophy;
PWS, Prader-Willi syndrome; SCA, spinocerebellar ataxias; SORD, sorbitol dehydrogenase deficiency; WES, whole-exome sequencing; WGS, whole-genome sequencing.

1. Barp A, et al. *Diagnostics (Basel)*. 2021;11:701. 2. Ng KWP, et al. *Front Neurol*. 2022;13:997551.

Types of Genetic Tests: New and Emerging Technologies

- **RNA sequencing**
- **Metabolomics**
- **Epigenomics**
- **Long read sequencing**
- **Optical genome mapping**

Method	Advantages	Disadvantages	Cost
RNA sequencing	Functional readout of RNA products for expression and splicing	Relevant tissues often not available Highly variable profiles across control cohorts Lack of appropriate cohorts for different demographics	$$
Metabolomics	Functional readout of metabolome Can be open or targeted	Highly variable profiles across control cohorts Lack of appropriate cohorts for different demographics	$
Proteomics	Functional readout of proteome Can be open or targeted	Highly variable profiles across control cohorts Lack of appropriate cohorts for different demographics	$
Lipidomics	Functional readout of lipidome Can be open or targeted	Highly variable profiles across control cohorts Lack of appropriate cohorts for different demographics	$
Methylation array	Functional readout of DNA methylation across the genome	Limited resolution. Lack of appropriate cohorts for different demographics Published signatures are often not available to compare with	$$
Methylation-sensitive MLPA	Inexpensive, rapid targeted analysis	Only practical for very limited targeted analysis	$
MAVE	Functional readout of genomic variants	Need a suitable assay for genes or variants of interest	$, $$

MAVE, multiplexed assays of variant effect; MLPA, multiplex ligation-dependent probe amplification. $, $$, $$$, $$$$ denote relative cost.

Kernohan and Boycott. The expanding diagnostic toolbox for rare genetic diseases. Nat Rev Genet. 2024 Jun;25(6):401-415. (PMID: 38238519)

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Test Your Knowledge

True or False:

- Q1: Exome sequencing can detect myotonic dystrophy type 1
  - ➢ False

- Q2: On exome sequencing, secondary findings are optional.
  - ➢ True....
    ...but they are not optional to discuss and must be reviewed with every patient and each person to be included in testing

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Panel Discussion

- What are the most common points of confusion around different test types?
- What type of testing should always be referred to a genetic counselor?

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Considerations When Ordering a Genetic Test

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When Testing May Be Inappropriate

Unstable emotional or psychological state of the patient

Example:
Presymptomatic HD testing

Patient unable to properly provide informed consent

Example:
Patient with ID, overt substance abuse, etc.

Improper ordering provider

Testing should be ordered by clinician comfortable interpreting complex genetic testing results

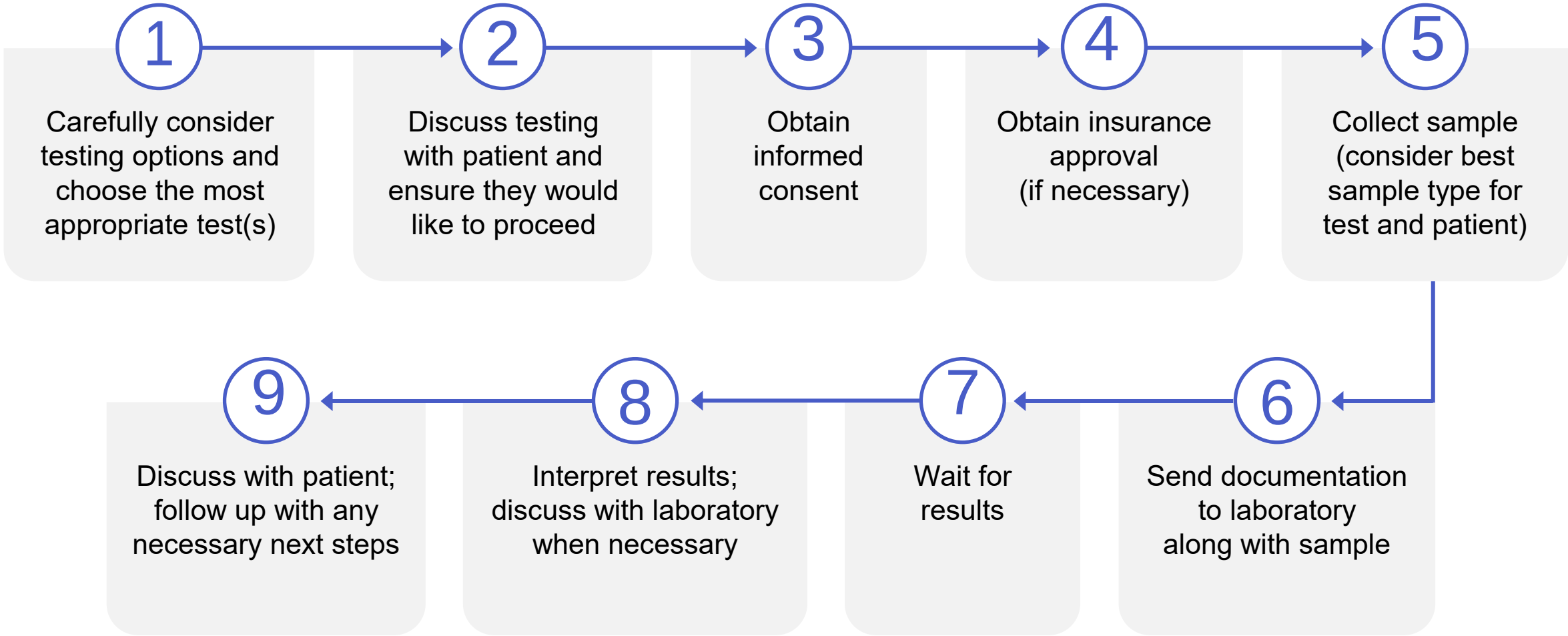
Testing of minors for adult-onset conditions and/or carrier status

Prenatal testing (ongoing pregnancy) for adult-onset disorders

Prohibitive cost of testing

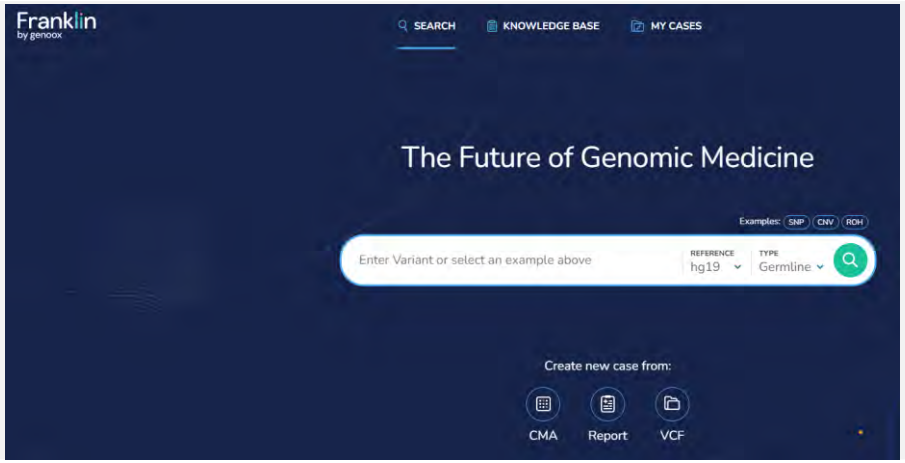
HD, Huntington disease; ID, intellectual disability.

How Do I Order a Genetic Test?



Resources

To find a genetic counselor in your area, consult the National Society of Genetic Counselors: https://findageneticcounselor.nsgc.org/



Variant assessment tool: https://franklin.genoox.com/clinical-db/home

Sponsored Programs for Neuromuscular Diseases*

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
  - DMD as first test
  - Reflex to neuromuscular panel if negative

Carrier testing

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

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

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

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

Sponsored Programs for Neuromuscular Diseases* (cont.)

Neuropathy

PreventionGenetics Alnylam Act hATTR Amyloidosis[1]

Includes TTR analysis only

PreventionGenetics NavigATTR[2]

Friedreich Ataxia

PreventionGenetics FA Identified[3]

- Includes analysis of FXN
- Offered to US and Puerto Rico residents aged 16 and older

Spinal Muscular Atrophy

Invitae SMA Identified[4]

Athena Diagnostics SMA Testing[5]

ALS

Invitae ALS Identified[6]

Includes C9orf72 and ALS panel

PreventionGenetics ALS Testing Program[7]

Includes analysis of ATXN2 as well as C9orf72 and ALS panel

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

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

1. Prevention Genetics. AlnylamAct hATTR Amyloidosis. https://www.preventiongenetics.com/sponsoredTesting/ttr 2. Prevention Genetics. https://www.preventiongenetics.com/sponsoredTesting/navigattr 3. Prevention Genetics. https://www.preventiongenetics.com/sponsoredTesting/faidentified 4. INVITAE. Spinal Muscular Atrophy (SMA) Identified Program. https://www.invitae.com/en/sponsored-testing/sma-identified. 5. Athena Diagnositics https://www.athenadiagnostics.com/view-full-catalog/sma-diagnostic-test1 6. INVITAE. Amyotrophic Lateral Sclerosis Identified Program. https://www.invitae.com/en/sponsored-testing/als-identified. 7. Prevention Genetics. Amyotrophic Lateral Sclerosis (ALS) Testing Program. https://www.preventiongenetics.com/sponsoredTesting/Ionis_ALS

Sponsored Programs for Neuromuscular Diseases* (cont.)

Mitochondrial Disease

PreventionGenetics No-Cost Genetic Testing Program for TK2 Deficiency[1]

Periodic Paralysis

Invitae Uncovering Periodic Paralysis[2]

Lysosomal Storage Diseases

Lantern Project[3]

Only available to patients aged ≥18 years

Test options

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

PreventionGenetics No-Cost Genetic Testing for Fabry Disease[4]

Includes sequencing and deletion/duplication of GLA gene

- Enzyme activity assay optional

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

TK2, thymidine kinase 2; UMDF, United Mitochondrial Disease Foundation.

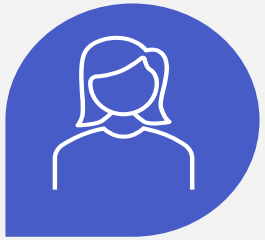
1. Prevention Genetics. Zogenix No-Cost Genetic Testing Program for Thymidine Kinase 2 Deficiency. https://www.preventiongenetics.com/sponsoredTesting/Zogenix. 2. INVITAE. Uncovering Periodic Paralysis. https://www.invitae.com/en/uncoveringperiodicparalysis/. 3. PerkinElmer Genetics. The Lantern Project. https://www.perkinelmergenomics.com/lanternproject/. 4. https://aakp.org/center-for-patient-research-and-education/fabry-disease/

Panel Discussion

- How commonly do “when not to test” scenarios occur?
  - How do the discussions tend to evolve?
  - What role does each provider play?

- What advice do you have for providers who don’t feel comfortable interpreting a test?
  - When should they reach out to a genetic counselor?

Case Study



- Patient diagnosed with DYSF MD based on a VUS and path variant
- VUS later downgraded to benign and "took the diagnosis away" from the patient

Do not immediately assume that VUS = diagnosis

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Key Takeaways

- Each genetic test has its own pros and cons
- Ideally, involve a genetic counselor when ordering any genetic test
- Resources are available to facilitate access

This presentation and a pdf of sponsored testing programs are available for download at: MDA.org/MedEd

Thank You

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