# Geisinger Health Plan Policies and Procedure Manual

**Policy: MP378**

**Section: Medical Policy**

**Subject: Genetic Testing for Neuromuscular Disorders**

**Applicable line of business:**

| | | | |
|---|---|---|---|
| **Commercial** | **x** | **Medicaid** | **x** |
| **Medicare** | **x** | **ACA** | **x** |
| **CHIP** | **x** | | |

**I. Policy:** Genetic Testing for Neuromuscular Disorders

**II. Purpose/Objective:** To provide a policy of coverage regarding Genetic Testing for Neuromuscular Disorders

**III. Responsibility:**

A. Medical Directors
B. Medical Management

**IV. Required Definitions**

1. Attachment – a supporting document that is developed and maintained by the policy writer or department requiring/authoring the policy.
2. Exhibit – a supporting document developed and maintained in a department other than the department requiring/authoring the policy.
3. Devised – the date the policy was implemented.
4. Revised – the date of every revision to the policy, including typographical and grammatical changes.
5. Reviewed – the date documenting the annual review if the policy has no revisions necessary.

Commercial

Geisinger Health Plan may refer collectively to health care coverage sponsors Geisinger Health Plan, Geisinger Quality Options, Inc., and Geisinger Indemnity Insurance Company, unless otherwise noted. Geisinger Health Plan is part of Geisinger, an integrated health care delivery and coverage organization.

Medicare

Geisinger Gold Medicare Advantage HMO, PPO, and HMO D-SNP plans are offered by Geisinger Health Plan/Geisinger Indemnity Insurance Company, health plans with a Medicare contract. Continued enrollment in Geisinger Gold depends on contract renewal. Geisinger Health Plan/Geisinger Indemnity Insurance Company are part of Geisinger, an integrated health care delivery and coverage organization.

CHIP

Geisinger Health Plan Kids (GHP Kids) is a Children's Health Insurance Program (CHIP) offered by Geisinger Health Plan in conjunction with the Pennsylvania Department of Human Services (DHS). Geisinger Health Plan is part of Geisinger, an integrated health care delivery and coverage organization.

Medicaid

Geisinger Health Plan Family (GHP Family) is a Medical Assistance (Medicaid) insurance program offered by Geisinger Health Plan in conjunction with the Pennsylvania Department of Human Services (DHS). Geisinger Health Plan is part of Geisinger, an integrated health care delivery and coverage organization

**V. Additional Definitions**
Medical Necessity or Medically Necessary means Covered Services rendered by a Health Care Provider that the Plan determines are:

a. appropriate for the symptoms and diagnosis or treatment of the Member's condition, illness, disease or injury;
b. provided for the diagnosis, and the direct care and treatment of the Member's condition, illness disease or injury;
c. in accordance with current standards of good medical treatment practiced by the general medical community.
d. not primarily for the convenience of the Member, or the Member's Health Care Provider; and
e. the most appropriate source or level of service that can safely be provided to the Member. When applied to hospitalization, this further means that the Member requires acute care as an inpatient due to the nature of the services rendered or the Member's condition, and the Member cannot receive safe or adequate care as an outpatient.

**Medicaid Business Segment**
Medically Necessary — A service, item, procedure, or level of care that is necessary for the proper treatment or management of an illness, injury, or disability is one that:

- Will, or is reasonably expected to, prevent the onset of an illness, condition, injury or disability.
- Will, or is reasonably expected to, reduce or ameliorate the physical, mental or developmental effects of an illness, condition, injury or disability.
- Will assist the Member to achieve or maintain maximum functional capacity in performing daily activities, taking into account both the functional capacity of the Member and those functional capacities that are appropriate for Members of the same age.

**DESCRIPTION:**
Neuromuscular disorders consist of a genetically and phenotypically heterogenous group of diseases, disrupting any component of the neuroaxis of the peripheral nervous system. These disrupted components can be skeletal muscle, neuromuscular junction, or nerves. The causes can be genetic (single gene disorder, polygenic disorder), nongenetic (infective, autoimmune, autoinflammatory), or yet to be identified.

**INDICATIONS:**

## TARGETED VARIANT ANALYSIS
Targeted mutation analysis for a known familial variant associated with a neurodegenerative or neuromuscular disease is considered medically necessary when the member has a close blood relative with a known pathogenic variant for the disease.

## COMPREHENSIVE NEUROMUSCULAR DISORDERS PANEL
Comprehensive neuromuscular panel testing is considered medically necessary to establish a diagnosis when the following criteria are met:
At least one of the following conditions are present:

- Neonatal stridor, respiratory insufficiency and episodic cyanotic and/or apneic episodes; **OR**
- Neonatal feeding difficulties, poor suck or choking; **OR**
- Neonatal generalized weakness, bulbar palsy, facial weakness or ptosis; **OR**
- Generalized or focal motor weakness; **OR**
- Muscle fatigability and/or atrophy with evidence of neuromuscular transmission defect(s) on electromyography: **OR**
- Ptosis or extraocular muscle weakness;

**AND**

- The member has undergone targeted analysis with non-diagnostic results; **OR**
- The member’s presentation is inconsistent with a neuromuscular disorder for which a targeted gene analysis is considered to be the standard of care.

## COMPREHENSIVE ATAXIA PANEL
Comprehensive ataxia panel testing is considered to be medically necessary in members who exhibit any of the following:

- Progressive dysarthria; **OR**
- Eye muscle weakness; **OR**
- Eye movement abnormalities; **OR**

- Progressive coordination difficulties with hand and/or finger movement; **OR**
- Progressive gait disturbance

**AND**

Non-genetic etiologies such as paraneoplastic disease, vascular disorders, alcoholism, nutritional deficiencies, and multiple sclerosis have been ruled out.

## PARKINSON DISEASE

Multigene panel testing for Parkinson disease is considered to be medically necessary when:

- The member exhibits clinical symptoms of Parkinson disease before age 50; **OR**
- The member exhibits clinical symptoms pf Parkinson disease**; AND**
    - The member has a family history of at least one relative with Parkinson disease, movement disorder or dementia

## MYOTONIC DYSTROPHY

CNBP repeat analysis and/or DMPK repeat expansion analysis is considered to be medically necessary in the following circumstances:

1. Members at any age with evidence of myotonic discharges on electromyography ( EMG);

**OR**

2. Members at any age with evidence of grip myotonia

**OR**

3. Members at any age with at least one of the following:
    - Distal extremity weakness; **OR**
    - Face and neck weakness

**AND at least one of the following**

- Insulin insensitivity; **OR**
- Hypogammaglobulinemia; **OR**
- Posterior subcapsular cataracts; **OR**
- Cardiomyopathy or conduction defects

4. Neonates with at least two of the following presentations:
    - Respiratory insufficiency
    - Generalized weakness
    - Hypotonia
    - Facial weakness
    - Positional malformations (e.g., clubfoot deformity, etc)

5. Asymptomatic adult members with a close blood relative with known myotonic dystrophy

## INHERITED PERIPHERAL NEUROPATHIES (e.g. Charcot-Marie-Tooth, etc)

Multigene panel testing for inherited peripheral neuropathies is considered to be medically necessary when the member exhibits one or more of the following:

- Distal muscle weakness and atrophy; **OR**
- Diminished tendon reflexes; **OR**
- Diminished ankle dorsiflexion (foot drop); **OR**
- Pes cavus deformity of the foot; **OR**
- Recurrent acute focal sensory and/or motor neuropathies; **OR**
- Painless nerve palsy after minor trauma or compression; **OR**
- Distal sensory loss

## FRIEDREICH'S ATAXIA

Testing for FXN repeat analysis or sequencing analysis is considered to be medically necessary the:

1. The member is at least 18 years of age, is asymptomatic and has biological sibling diagnosed with Friedreich's ataxia

**OR**

2. The member exhibits at least two of the following:
    - Progressive ataxia; **OR**
    - Dysarthria; **OR**
    - Diminished sense of position and/or vibration in the lower extremities; **OR**
    - Pyramidal weakness of the lower extremities; **OR**

- Muscle weakness; **OR**
- Optic atrophy or deafness; **OR**
- Extensor plantar signs (Babinski sign); **OR**
- Pes cavus deformity of the foot; **OR**
- Diabetes or glucose intolerance; **OR**
- Hypertrophic nonobstructive cardiomyopathy

**AND**

Non-genetic etiologies such as paraneoplastic disease, vascular disorders, alcoholism, nutritional deficiencies, and multiple sclerosis have been ruled out.

## HEREDITARY SPASTIC PARAPLEGIA

Multigene panel testing is considered to be medically necessary to confirm clinical diagnosis in members who exhibit any of the following:

- Lower extremity spasticity; **OR**
- Lower extremity hyperreflexia and Extensor plantar signs (Babinski sign); **OR**
- Diminished sense of vibration in the lower extremities; **OR**
- Weakness of the hamstring, tibialis anterior or iliopsoas

## AMYOTROPHIC LATERAL SCLEROSIS (ALS)

Multigene panel testing is considered to be medically necessary in members meeting the following criteria:

The member is at least 18 years of age and exhibits **ALL** of the following:

- Signs and symptoms of lower motor neuron degeneration; **AND**
- Signs and symptoms of upper motor neuron degeneration; **AND**
- Progressive worsening of symptoms; **AND**
- Absence of other disease states to account for the upper and lower motor neuron degeneration

## DUCHENNE and BECKER MUSCULAR DYSTROPHY

Genetic testing in for variants the DMD gene is considered to be medically necessary in the following circumstances:

- To confirm the diagnosis in member exhibiting signs and symptoms of a dystrophinopathy; or
- To confirm or exclude the need for surveillance in at-risk relatives; or
- For at-risk first and second-degree relative of an individual with a dystrophinopathy and results of testing will allow informed reproductive decision making; or
- Prenatal testing if at least one parent is known to be a carrier or has a close blood relative who has the disease or is a known carrier.
- To inform appropriate gene therapy opportunities if DMD testing was completed greater than one year ago AND was negative or uninformative

## HUNTINGTON DISEASE

Genetic testing is considered to be medically necessary in members meeting the following criteria:

1. The member is
    - An adult with unexplained progressive choreatic movement disorder and neuropsychiatric disturbance; or
    - An adult with a 1st, 2nd or 3rd degree family member with a confirmed molecular diagnosis of the disease; or
    - A pediatric ( <18 yrs) member with either:
        - A known family history of the disease; or
        - Two or more of the following symptoms:
            - Seizures
            - Rigidity
            - Gait disturbance
            - Oral motor dysfunction

    **AND**
2. The member has a documented assessment by a licensed clinical psychologist or social worker regarding implications of this testing

    **AND**
3. Informed consent for testing from the member or member's legal guardian has been obtained by the ordering provider or care team

**EXCLUSIONS:**
Targeted variant analysis in the absence of signs or symptoms of the disease, or in the absence of a blood relative with a known pathogenic variant is considered to be of **UNPROVEN** values and is therefore **NOT COVERED**.

**Note: A complete description of the process by which a given technology or service is evaluated and determined to be experimental, investigational or unproven is outlined in MP 15 - Experimental Investigational or Unproven Services or Treatment.**

## Medicaid Business Segment:
Any requests for services, that do not meet criteria set in the PARP, may be evaluated on a case by case basis.

**CODING ASSOCIATED WITH:**
***The following codes are included below for informational purposes and may not be all inclusive. Inclusion of a procedure or device code(s) does not constitute or imply coverage nor does it imply or guarantee provider reimbursement. Coverage is determined by the member specific benefit plan document and any applicable laws regarding coverage of specific services. Please note that per Medicare coverage rules, only specific CPT/HCPCS Codes may be covered for the Medicare Business Segment. Please consult the CMS website at*** www.cms.gov ***or the local Medicare Administrative Carrier (MAC) for more information on Medicare coverage and coding requirements.***

81161 DMD (dystrophin) (eg, duchenne/becker muscular dystrophy) deletion analysis, and duplication analysis, if performed,
81179 ATXN2 (ataxin 2) (eg, spinocerebellar ataxia) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
81185 CACNA1A (calcium voltage-gated channel subunit alpha1 A) (eg, spinocerebellar ataxia) gene analysis; full gene sequence
81187 CNBP (CCHC-type zinc finger nucleic acid binding protein) (eg, myotonic dystrophy type 2) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
81188 CSTB (cystatin B) (e.g., Unverricht-Lundborg disease) gene analysis; evaluation to detect abnormal (e.g., expanded) alleles
81189 CSTB (cystatin B) (e.g., Unverricht-Lundborg disease) gene analysis; full gene sequence
81190 CSTB (cystatin B) (e.g., Unverricht-Lundborg disease) gene analysis; known familial variant(s)
81284 FXN (frataxin) (eg, Friedreich ataxia) gene analysis; evaluation to detect abnormal (expanded) alleles,
81285 FXN (frataxin) (eg, Friedreich ataxia) gene analysis evaluation; characterization of alleles (eg, expanded size)
81286 FXN (frataxin) (eg, Friedreich ataxia) gene analysis; full gene sequence,
81324 PMP22 (peripheral myelin protein 22) (eg, Charcot-Marie-Tooth, hereditary neuropathy with liability to pressure palsies) gene analysis; duplication/deletion analysis,
81325 full sequence analysis, known familial variant
81326 family variant
81239 SMN1 (survival of motor neuron 1, telomeric) (eg, spinal muscular atrophy) gene analysis; dosage/deletion analysis (eg, carrier testing), includes SMN2 (survival of motor neuron 2, centromeric) analysis, if performed ,
81401 Molecular pathology procedure, Level 2 (eg, 2-hyphen10 SNPs, 1 methylated variant, or 1 somatic variant [typically using nonsequencing target variant analysis], or detection of a dynamic mutation disorder/triplet repeat)
81448 Hereditary peripheral neuropathies (e.g., Charcot-Marie-Tooth, spastic paraplegia), genomic sequence analysis panel, must include sequencing of at least 5 peripheral neuropathy-related genes (e.g., BSCL2, GJB1, MFN2, MPZ, REEP1, SPAST, SPG11, SPTLC1)
81286 FXN (frataxin) (eg, Friedreich ataxia) gene analysis; full gene sequence,
81403 Molecular pathology procedure, Level 4 (eg, analysis of single exon by DNA sequence analysis, analysis of >10 amplicons using multiplex PCR in 2 or more independent reactions, mutation scanning or duplication/deletion variants of 2-5 exons)
81404 Molecular pathology procedure, Level 5 (eg, analysis of 2-hyphen5 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 6-10 exons, or characterization of a dynamic mutation disorder/triplet repeat by Southern blot analysis)
81405 Molecular pathology procedure, Level 6 (eg, analysis of 6-10 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 11-25 exons, regionally targeted cytogenomic array analysis)
81406 Molecular pathology procedure, Level 7 (eg, analysis of 11-25 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 26-50 exons, cytogenomic array analysis for neoplasia
81407
81408 Molecular pathology procedure, Level 9 (e.g., analysis of >50 exons in a single gene by DNA sequence analysis) --includes DMD (dystrophin) (e.g., Duchenne/Becker muscular dystrophy), full gene sequence
0216U {Genomic Unity Ataxia Repeat Expansion Analysis}
0217U {Genomic Unity Comprehensive Ataxia Analysis}
0218U Neurology (muscular dystrophy), DMD gene sequence analysis, including small sequence changes, deletions,

duplications, and variants in non-uniquely mappable regions, blood or saliva, identification and characterization of genetic variants {Genomic Unity DMD Gene Analysis}

0233U FXN (frataxin) (eg, Friedreich ataxia), gene analysis, including small sequence changes in exonic and intronic regions, deletions, duplications, short tandem repeat (STR) expansions, mobile element insertions, and variants in non-hyphenuniquely mappable regions {Genomic Unity FXN Analysis}



**LINE OF BUSINESS:**
**Eligibility and contract specific benefits, limitations and/or exclusions will apply. Coverage statements found in the line of business specific benefit document will supersede this policy. For Medicare, applicable LCD's and NCD's will supercede this policy. For PA Medicaid Business segment, this policy applies as written.**

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This policy will be revised as necessary and reviewed no less than annually.

**Devised:** 9/24

**Revised:**

**Reviewed:**

**CMS UM Oversight Committee Approval:** 11/8/24

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Coverage for experimental or investigational treatments, services and procedures is specifically excluded under the member's certificate with Geisinger Health Plan. Unproven services outside of an approved clinical trial are also specifically excluded under the member's certificate with Geisinger Health Plan. This policy does not expand coverage to services or items specifically excluded from coverage in the member's certificate with Geisinger Health Plan. Additional information can be found in MP015 Experimental, Investigational or Unproven Services.

Prior authorization and/or pre-certification requirements for services or items may apply. Pre-certification lists may be found in the member's contract specific benefit document. Prior authorization requirements can be found at https://www.geisinger.org/health-plan/providers/ghp-clinical-policies