Overview

Useful For
Confirmation of a clinical diagnosis of Duchenne muscular dystrophy (DMD) or Becker muscular dystrophy (BMD)

Distinguishing DMD from BMD in some cases, based on the type of deletion detected (allows for better prediction of prognosis)

Determination of carrier status in family member at risk for DMD or BMD

Prenatal diagnosis of DMD or BMD in at-risk pregnancies

Genetics Test Information
Deletions and duplications only.

If testing is being performed due to family history, documentation regarding the familial mutation before testing an asymptomatic individual or proceeding with carrier testing is preferred.

Reflex Tests

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Reporting Name</th>
<th>Available Separately</th>
<th>Always Performed</th>
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<tbody>
<tr>
<td>CULFB</td>
<td>Fibroblast Culture for Genetic Test</td>
<td>Yes</td>
<td>No</td>
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<tr>
<td>CULAF</td>
<td>Amniotic Fluid Culture/Genetic Test</td>
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<tr>
<td>MATCC</td>
<td>Maternal Cell Contamination, B</td>
<td>Yes</td>
<td>No</td>
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</tbody>
</table>

Testing Algorithm
For prenatal specimens only: If amniotic fluid (nonconfluent cultured cells) is received, amniotic fluid culture/genetic test will be added and charged separately. If chorionic villus specimen (nonconfluent cultured cells) is received, fibroblast culture for genetic test will be added and charged separately. For any prenatal specimen that is received, maternal cell contamination studies will be added.

See Neuromuscular Myopathy Testing Algorithm in Special Instructions.

Special Instructions
- Informed Consent for Genetic Testing
- Molecular Genetics: Neurology Patient Information
- Neuromuscular Myopathy Testing Algorithm
- Informed Consent for Genetic Testing (Spanish)

Method Name
Dosage Analysis by Polymerase Chain Reaction (PCR)/Multiplex Ligation-Dependent Probe Amplification (MLPA)

NY State Available
Yes
Specimen

Specimen Type
Varies

Shipping Instructions
Specimen preferred to arrive within 96 hours of collection.

Specimen Required
Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Submit only 1 of the following specimens:

Preferred:

Specimen Type: Whole blood

Container/Tube:
Preferred: Lavender top (EDTA) or yellow top (ACD)
Acceptable: Any anticoagulant

Specimen Volume: 3 mL

Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tube.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Due to the complexity of prenatal testing, consultation with the laboratory is required for all prenatal testing. Prenatal specimens can be sent Monday through Thursday and must be received by 5 p.m. CST on Friday in order to be processed appropriately. All prenatal specimens must be accompanied by a maternal blood specimen. Order MATCC / Maternal Cell Contamination, Molecular Analysis on the maternal specimen.

Specimen Type: Amniotic fluid

Container/Tube: Amniotic fluid container

Specimen Volume: 20 mL

Specimen Stability Information: Refrigerated (preferred)/Ambient

Specimen Type: Chorionic villi
Test Definition: DBMD
DMD/BMD Deletion/Duplication

Container/Tube: 15-mL tube containing 15 mL of transport media

Specimen Volume: 20 mg

Specimen Stability Information: Refrigerated

Acceptable:

Specimen Type: Confluent cultured cells

Container/Tube: T-25 flask

Specimen Volume: 2 flasks

Collection Instructions: Submit confluent cultured cells from another laboratory.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Forms

1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available in Special Instructions:

- Informed Consent for Genetic Testing (T576)
- Informed Consent for Genetic Testing-Spanish (T826)

2. Molecular Genetics: Neurology Patient Information in Special Instructions

3. If not ordering electronically, complete, print, and send a Neurology Specialty Testing Client Test Request (T732) with the specimen.

Specimen Minimum Volume

Blood: 1 mL
Amniotic Fluid: 10 mL
Chorionic Villus: 5 mg

Reject Due To

No specimen should be rejected.

Specimen Stability Information

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<th>Temperature</th>
<th>Time</th>
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Clinical and Interpretive

Clinical Information

Duchenne muscular dystrophy (DMD) is an X-linked recessive disorder characterized initially by proximal muscle weakness beginning before age 5 years. Affected individuals typically have pseudohypertrophy of the calf muscles.
and exhibit toe-walking, waddling gait, and the Gower sign (climbing up the legs when rising from a seated position on the floor). Not only is skeletal muscle affected in DMD, but also the smooth muscle of the gastrointestinal tract and possibly bladder, as well as cardiac muscle.

Initial symptoms are followed by dramatic progression of weakness leading to loss of ambulation by age 11 or 12. Death is often caused by cardiac failure or by respiratory failure before age 30, unless ventilator support is provided.

The allelic Becker muscular dystrophy (BMD) has a similar presentation, although age of onset is later and the clinical course is much milder. Cardiac involvement can be the only sign and patients are often ambulatory into their thirties.

DMD and BMD are caused by mutations in the DMD gene, which encodes for dystrophin. Approximately 50% to 65% of patients have intragenic deletions and approximately 5% to 10% have intragenic duplications. Less frequently, DMD and BMD result from nondeletion and nonduplication mutations, which are not detected by this assay.

Approximately one-third of sporadic cases of DMD/BMD occur due to new mutations. In sporadic cases, it is possible for the mother of an affected individual to have germline mosaicism. This means that the germ cells may contain a mutation even if the mutation is not detected in peripheral blood. In cases of germline mosaicism, which occurs with a frequency of up to 15%, further offspring are at risk for inheriting a dystrophin mutation.

**Reference Values**

An interpretive report will be provided.

**Interpretation**

An interpretive report will be provided.

**Cautions**

In addition to disease-related probes, the multiplex ligation-dependent probe amplification technique utilizes probes localized to other chromosomal regions as internal controls. In certain circumstances, these control probes may detect other diseases or conditions for which this test was not specifically intended. Results of the control probes are not normally reported. However, in cases where clinically relevant information is identified, the ordering physician will be informed of the result and provided with recommendations for any appropriate follow-up testing.

This test may not detect deletions/duplications present in very low levels of mosaicism

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.

**Clinical Reference**


Performance

Method Description

Multiple ligation-dependent probe amplification (MLPA) is utilized to test for the presence of large deletions and duplications within the DMD gene. (Unpublished Mayo method)

PDF Report

No

Day(s) and Time(s) Test Performed

Batched 1 time per week

Analytic Time

14 days

Maximum Laboratory Time

21 days

Specimen Retention Time

Whole Blood: 2 weeks (if available); Extracted DNA: 3 months

Performing Laboratory Location

Rochester

Fees and Codes

Fees

- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. For assistance, contact Customer Service.

Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

CPT Code Information

81161-DMD (dystrophin) (eg, Duchenne/Becker muscular dystrophy) deletion analysis and duplication analysis, if performed

Fibroblast Culture for Genetic Test

88233-Tissue culture, skin or solid tissue biopsy (if appropriate)

88240-Cryopreservation (if appropriate)

Amniotic Fluid Culture/Genetic Test

88235-Tissue culture for amniotic fluid (if appropriate)
**Test Definition: DBMD**

DMD/BMD Deletion/Duplication

88240-Cryopreservation (if appropriate)

Maternal Cell Contamination, B

81265-Comparative analysis using Short Tandem Repeat (STR) markers; patient and comparative specimen (eg, pre-transplant recipient and donor germline testing, post-transplant non-hematopoietic recipient germline [eg, buccal swab or other germline tissue sample] and donor testing, twin zygosity testing or maternal cell contamination of fetal cells (if appropriate)

**LOINC® Information**

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