MAYO CLINIC LABORATORIES

Test Definition: DBMD

Duchenne/Becker Muscular Dystrophy, DMD Gene, Large Deletion/Duplication Analysis, Varies

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

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
CULFB	Fibroblast Culture for Genetic Test	Yes	No
CULAF	Amniotic Fluid Culture/Genetic Test	Yes	No
MATCC	Maternal Cell Contamination, B	Yes	No
_STR1	Comp Analysis using STR (Bill only)	No, (Bill only)	No
_STR2	Add'l comp analysis w/STR (Bill Only)	No, (Bill only)	No

Genetics Test Information

This test is for genetic deletions and duplications only.

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

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

Additional Testing Requirements
All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

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. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.

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. **Do not** aliquot.
Specimen Stability Information: Ambient (preferred)/Refrigerated

Prenatal Specimens

Due to its complexity, consultation with the laboratory is required for all prenatal testing; call 800-533-1710 to speak to a genetic counselor.

Specimen Type: Amniotic fluid
Container/Tube: Amniotic fluid container
Specimen Volume: 20 mL
Specimen Stability Information: Refrigerated (preferred)/Ambient

Specimen Type: Chorionic villi
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

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Clinical & 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 years 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 variants 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 variants, which are not detected by this assay.

Approximately one-third of sporadic cases of DMD/BMD occur due to new variants. 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 variant even if the variant 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 variant.

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 alterations (ie, 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

1. Thompson MW, McInnes RR, Willard HF: Genetics in Medicine. 5th ed. WB Saunders Company; 1991:367-372
2. Desquerre I, Christov C, Mayer M, et al: Clinical heterogeneity of duchenne muscular dystrophy (DMD): definition of sub-phenotypes and predictive criteria by long-term follow-up. PLoS One. 2009;4(2):e4347. doi: 10.1371/journal.pone.0004347
3. Verma S, Anziska Y, Cracco J: Review of Duchenne muscular dystrophy (DMD) for the pediatricians in the community. Clin Pediatr (Phila). 2010;49(11):1011-1017. doi: 10.1177/0009922810378738

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) Performed

Batched 1 time per week

Report Available

14 to 21 days

Specimen Retention Time

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

Performing Laboratory Location

Rochester

Fees & 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 account representative. 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. It has not been cleared or approved by the US 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)
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

Test ID	Test Order Name	Order LOINC® Value
DBMD	DMD/BMD Deletion/Duplication	75385-5

Result ID	Test Result Name	Result LOINC® Value
55261	Result Summary	50397-9
55262	Result	75385-5
55263	Interpretation	69047-9
55264	Specimen	31208-2
55265	Source	31208-2
55266	Released By	18771-6