

Overview

Useful For

Obtaining a rapid, expert opinion on muscle biopsy specimens for neuromuscular disease

Testing Algorithm

A battery of enzyme histochemical stains will be performed on frozen tissue; additional histochemical stains or immunostains may be performed on frozen tissue; other tests can be performed as indicated and will be charged separately. The reviewing neuromuscular pathologist will determine the need for additional testing.

For all consultations, ancillary testing necessary to determine a diagnosis is ordered at the discretion of the Mayo Clinic neuromuscular pathologist. An interpretation, which includes an evaluation of the specimen and determination of a diagnosis, will be provided within a formal pathology report.

Frozen tissue sent for consultation: Appropriate additional stains may be performed at an additional charge.

Slides sent for consultation: Special stains and studies performed on the case should be sent with the case for review. In order to determine an accurate diagnosis, some of these stains or studies may be deemed to warrant repeat testing at an additional charge at the discretion of the reviewing Mayo Clinic neuromuscular pathologist. The interpreting neuromuscular pathologist may also request frozen tissue to perform additional studies that may be considered necessary for diagnosis.

Note: Testing requested by the referring physician (immunostains, etc) may not be performed if deemed unnecessary by the reviewing Mayo neuromuscular pathologist. Electron microscopic studies **are** not performed on muscle biopsy specimens. For more information see [Why Electron Microscopy is Not Performed on Muscle Biopsy Specimens](#) in Special Instructions. See [Pathology Consultation Ordering Algorithm](#) in Special Instructions.

Special Instructions

- [Muscle Biopsy Specimen Preparation](#)
- [Muscle Histochemistry Patient Information](#)
- [Pathology Consultation Ordering Algorithm](#)
- [Why Electron Microscopy is Not Performed on Muscle Biopsy Specimens](#)

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
IHPCI	IHC Initial	No	No
IHPCA	IHC Additional	No	No
IFPCI	IF Initial	No	No
IFPCA	IF Additional	No	No
SS2PC	SpecStain, Grp II, other	No	No
SS3PC	SpecStain, Grp III, enzyme	No	No
HCFPC	SpecStain, frozen	No	No
COSPC	Consult, Outside Slide	No	No
CSPPC	Consult, w/Slide Prep	No	No
CUPPC	Consult, w/USS Prof	No	No
CRHPC	Consult, w/Comp Rvw of His	No	No
LV4RP	Level 4 Gross and Microscopic, RB	No	No

Method Name

Muscle Biopsy Surgical Pathology Consultation and/or Review of Outside Material

NY State Available

Yes

Specimen**Specimen Type**

Varies

Ordering Guidance

This test is not appropriate for inhalation-transmission diseases such as tuberculosis, *Brucella*, measles, and varicella zoster. This test is also not appropriate for suspected Creutzfeldt-Jacobs Disease (CJD).

Additional Testing Requirements

Biopsies from different sites require separate orders and separate specimen vials.

Necessary Information

Failure to provide this information may delay the specimen processing significantly.

1. Collection date and patient date of birth are required.
2. The name and phone number of the ordering physician and a brief history are essential to achieve a consultation fully relevant to the ordering physician's needs.
3. [Muscle Histochemistry Patient Information](#) must accompany all specimens.

Specimen Required

Biopsies from different sites require separate orders and separate specimen vials.

Preferred: Frozen muscle biopsy tissue

Supplies: Muscle Biopsy Kit (T541)

Specimen Type: Muscle biopsy tissue (frozen) and/or slides

Collection Instructions:

1. Prepare and transport specimen per instructions in [Muscle Biopsy Specimen Preparation](#) in Special Instructions.
2. Patient history and requests should be clearly labeled with correct patient identifiers and pathology accession/case number.
3. All specimens must be labeled with specimen type.

Additional Information: Contact the Mayo Clinic Muscle Laboratory for special problems to maximize benefit of the muscle biopsy.

Acceptable: Stained muscle biopsy slides

1. Submit all stains performed on the case.
2. All specimens must be labeled with specimen type.

Forms

1. [Muscle Histochemistry Patient Information](#) (T361) is required, see Special Instructions.
2. [Why Electron Microscopy is Not Performed on Muscle Biopsy Specimens](#) in Special Instructions.

Reject Due To

No specimen should be rejected.

Specimen Minimum Volume

1.5 cm biopsy

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Frozen (preferred)		
	Ambient		

Clinical & Interpretive
Clinical Information

This consultative practice strives to bring the client the highest quality of diagnostic neuromuscular pathology, aiming to utilize only those ancillary tests that support the diagnosis in a cost-effective manner, and to provide a rapid turnaround time for diagnostic results.

Reference Values

An interpretive report will be provided.

Interpretation

Results are reported in a formal neuromuscular pathology report that includes diagnosis and an interpretive comment, if necessary. The formal pathology report is faxed or sent by mail according to the preference of the referring institution.

Cautions

Be aware that poor freezing and improper handling of the muscle tissue may hinder the neuromuscular pathologist's interpretation of the biopsy. It is crucial to provide a properly handled muscle specimen. In this regard, see directions for [Muscle Biopsy Specimen Preparation](#) sheet in Special Instructions. A shipping kit is available, Muscle Biopsy Kit (T541) and is recommended for collecting and shipping muscle specimens. It includes a container for the tissue, collection instructions, the [Muscle Histochemistry Patient Information](#) sheet, and a box to ship the specimen.

Clinical Reference

- Engel AG: The muscle biopsy. In: Engel AG, Franzini-Armstrong C, eds. Myology. 3rd ed. McGraw-Hill; 2004:681-690
- Liewluck T, Sorenson EJ, Walkiewicz MA, Rumilla KM, Milone M: Autosomal dominant distal myopathy due to a novel ACTA1 mutation. Neuromuscul Disord. 2017 Aug;27(8):742-746
- Engel AG, Redhage KR, Tester DJ, Ackerman MJ, Selcen D: Congenital myopathy associated with the triadin knockout syndrome. Neurology. 2017 Mar 21;88(12):1153-1156
- Niu Z, Pontifex CS, Berini S, et al: Myopathy with SQSTM1 and TIA1 variants: Clinical and pathological features. Front Neurol. 2018 Mar 19;9:147
- Nicolau S, Liewluck T, Shen XM, Selcen D, Engel AG, Milone M: A homozygous mutation in GMPPB leads to centronuclear myopathy with combined pre- and postsynaptic defects of neuromuscular transmission. Neuromuscul Disord. 2019 Aug;29(8):614-617
- Nicolau S, Liewluck T, Tracy JA, Laughlin RS, Milone M: Congenital myopathies in the adult neuromuscular clinic: Diagnostic challenges and pitfalls. Neurol Genet. 2019 Jun 4;5(4):e341
- Liewluck T, Niu Z, Moore SA, Alsharabati M, Milone M: ACTA1-myopathy with prominent finger flexor weakness and rimmed vacuoles. Neuromuscul Disord. 2019 May;29(5):388-391

8. Nicolau S, Liewluck T, Elliott JL, Engel AG, Milone M: A novel heterozygous mutation in the C-terminal region of HSPB8 leads to limb-girdle rimmed vacuolar myopathy. *Neuromuscul Disord.* 2020 Mar;30(3):236-240

Performance

Method Description

All requests will be processed as a consultation case first. Special studies will be performed only if deemed to be diagnostically indicated. Histochemical and immunohistochemical stains are only performed if frozen muscle biopsy tissue is provided. (Unpublished Mayo method)

PDF Report

No

Specimen Retention Time

6 months

Performing Laboratory Location

Rochester

Fees & Codes

Test Classification

This test has been cleared, approved, or is exempt by the US Food and Drug Administration and is used per manufacturer's instructions. Performance characteristics were verified by Mayo Clinic in a manner consistent with CLIA requirements.

CPT Code Information

88342 (if appropriate)

88341 (if appropriate)

88346 (if appropriate)

88350 (if appropriate)

88305 (if appropriate)

88313 (if appropriate)

88319 (if appropriate)

88314 (if appropriate)

88321 (if appropriate)

88323 (if appropriate)

88323-26 (if appropriate)

88325 (if appropriate)