For autopsy specimens, a common checklist can be filled out, but the presence of specific findings in specific muscles and nerves should be evaluated and reported.

**Clinical History**

**1. Gender of patient:**  male  female

**2. Age at presentation:** \_\_\_ years \_\_\_ months

**3. Age at biopsy:** \_\_\_ years \_\_\_ months **Age at prior muscle biopsies** \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**4. Symptoms at presentation (check all that apply):**

Weakness

If experiencing weakness, then indicate distribution:

Symmetrical  Asymmetrical  Limb-girdle

Proximal  Distal  Facioscapulohumeral

Paraspinal  Finger flexor  Neck  Other \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Other muscle symptoms:

Hypotonia  Muscle pain  Exercise intolerance

Episodic muscle pain/cramping  Rhabdomyolysis  Contractures

Failure to thrive

Respiratory difficulties

Skin changes

Eye symptoms:

Ptosis  Ophthalmoplegia

Joint laxity

Clinical features of cardiac involvement/Known cardiac disease, Specify:\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Central nervous system disease, Specify:\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Exposure to toxins, supplements, or drugs (and relationship between exposure and biopsy acquisition), Specify:\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Others (see item 9)

**5.** **Laboratory findings**

*Elevated creatine kinase:*  Yes  No  Unknown \_\_\_\_\_\_\_ Patient Value \_\_\_\_\_\_\_\_\_(Normal Range)

*Elevated erythrocyte sedimentation rate (ESR):*

Yes  No  Unknown \_\_\_\_\_\_\_ Patient Value\_\_\_\_\_\_ (Normal Range)

*Elevated C-reactive protein (CRP):*  Yes  No  Unknown \_\_\_\_\_Patient Value\_\_\_\_\_\_\_ (Normal Range)

*Known autoantibodies in patient:* **\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_**

**6. EMG Findings:**  Not known  Myopathic  Neuropathic

**7. Imaging findings (brain/muscle): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_**

**8. Familial Inheritance:**  None  Autosomal Recessive  Autosomal Dominant

X-linked  Maternal

**9. Other symptoms, signs, known diseases, and lab data: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_**

## Muscle Biopsy and Autopsy Tissue Information

1. \*Is this a biopsy or autopsy specimen?  Biopsy  Autopsy

If this is an autopsy specimen, what is the approximate postmortem interval? *(please specify)*

1. Tissue collected: (please specify)
2. \*Size of tissue collected: ( )x( )x( ) cm
3. \*Date of tissue collection: (mm/dd/yyyy)
4. Biopsy method:  Open  Needle
5. Name of laboratory where pathology was performed: (please specify)
6. Name of laboratory director: (please specify)  Unknown
7. Name of pathologist who diagnosed the case: (please specify)
8. \*Freezing or Fixation Used?

Frozen: Amount: *(please specify)* mg  Not known

Formalin-fixed: Amount: *(please specify)* mg  Not known

Paraffin-embedded: Amount: *(please specify)* mg  Not known

Epon-embedded: Amount: *(please specify)* mg  Not known

1. Was electron microscopy performed?  Yes  No
2. Was subsequent biochemical or genetic testing performed?  Yes  No

If Yes, record results in table below:

Table to input subsequent biochemical or genetic testing data

| Test Name | Results (including units) |
| --- | --- |
| *(data to be entered by site)* | *(data to be entered by site)* |
| *(data to be entered by site)* | *(data to be entered by site)* |
| *(data to be entered by site)* | *(data to be entered by site)* |

**Microscopic Description**

1. **Which standard histochemical stains were used\*? (choose all that apply)**

H and E  Gomori trichrome  NADH  COX  SDH

COX/SDH  PAS  Oil Red O  ATPase 4.3  ATPase 4.6

ATPase 9.4  Acid phosphatase  Phosphorylase  Myoadenylate deaminase

Esterase  Phosphofructokinase  Sudan black  Other, specify: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

1. **Which of the following diagnostic abnormalities were noted on histochemical stains (choose all that apply)\*?**

Fatty infiltration  absent  mild  moderate  severe

Endomysial fibrosis  absent  mild  moderate  severe

Myofiber degeneration  absent  mild  moderate  severe

Necrosis  absent  mild  moderate  severe

Myophagocytosis  absent  present in \_\_\_\_ fibers

Basophilic fibers, large nuclei  absent  present in \_\_\_\_\_ fibers

Hypertrophic fibers  absent  present in \_\_\_\_\_ fibers

Approximate fiber size (largest)\_\_\_\_\_\_\_\_\_\_\_

Atrophy/Hypotrophy  absent  present

Approximate fiber size (smallest) \_\_\_\_\_\_\_\_

Specify:

All fibers within the specimen

Subsets of fibers, leading to excessive variation in fiber size

Specify (choose all that apply):  single fibers  groups of fibers

type 1 fibers only  type 2 fibers only

Perifascicular distribution

Atrophic/hypotrophic fiber shape  angulated  round

Nuclear bags/clumps  absent  present

Myopathy-associated pathological structures, specify:

Central nuclei  absent  present

Specify estimated % of fibers (include eccentric nuclei): \_\_\_\_\_

Internal nuclei  absent  present

Specify estimated % of fibers (if not quantified above): \_\_\_\_\_

Marked hypotrophy of type 1 fibers  absent  present

Nemaline rods/bodies  absent  sparse  moderate (present in \_\_\_\_\_% of fibers)

Specify:  Restricted to one fiber type, specify which: \_\_\_\_\_

Red inclusions on trichrome  absent  sparse  moderate (present in \_\_\_\_\_% of fibers)

Rimmed vacuoles  absent  sparse  moderate (present in \_\_\_\_\_% of fibers)

Non-rimmed vacuoles  absent  sparse  moderate (present in \_\_\_\_\_% of fibers)

Ragged red fibers  absent  sparse  moderate (present in \_\_\_\_\_% of fibers)

COX- negative fibers  absent  sparse  moderate (present in \_\_\_\_\_% of fibers)

Strongly SDH-reactive blood vessels (SSV’s)  absent  present

Central cores  absent  sparse  moderate (present in \_\_\_\_\_% of fibers) Minicores  absent  sparse  moderate (present in \_\_\_\_\_% of fibers)

Core-like lesions  absent  sparse  moderate (present in \_\_\_\_\_% of fibers)

Targetoid fibers  absent  sparse  moderate (present in \_\_\_\_\_% of fibers)

Moth-eaten fibers  absent  sparse  moderate (present in \_\_\_\_\_% of fibers)

Tubular aggregates  absent  present in \_\_\_\_\_% of fibers)

only seen in type \_\_\_\_ fibers

Ring fibers  absent  sparse  moderate (present in \_\_\_\_\_% of fibers)

Split fibers  absent  sparse  moderate (present in \_\_\_\_\_% of fibers)

Lobulated fibers  absent  sparse  moderate (present in \_\_\_\_\_% of fibers)

Blood vessel deposits suggestive of amyloid  absent  present

Abnormalities of fiber type  absent  present

Specify\*:  Type 1 predominance \_\_\_\_\_\_ % Type 1 fibers

Type 2 predominance \_\_\_\_\_\_% Type 2 fibers

Fiber type grouping (of both fiber types)

Absent staining for a histochemical stain:\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Inflammation  absent  mild  moderate  severe

Specify:

Distribution

Perivascular

Evidence of vascular damage  Thrombi identified in blood vessels

Focal

Diffuse

Endomysial

Perimysial

Involving fascia

Associated with myofiber damage

Associated with non-necrotic myofiber

Granulomas

Necrotizing Non-necrotizing Giant cells present  Foreign material present

Inflammatory cells identified

Specify (choose all that apply):

Lymphocytes

Neutrophils

Macrophages

Eosinophils (as a prominent component)

Microorganisms identified, specify which: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Abnormal storage material

Specify:

Excessive glycogen  absent  mild  marked

Excessive intracellular lipid  absent  mild  marked

Intramuscular nerve branches  absent  present

Specify:

Decreased axonal density  Increased endoneurial fibrosis

Abnormality of myelination \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Abnormal structures \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Muscle spindles  absent present

Myotentinous insertion sites absent present

Additional observations

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

1. **Which immunohistochemical stains were used? (choose all that apply)**

Myosin immunohistochemistry

Fast myosin \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Slow myosin \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Embryonic myosin \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Fetal myosin \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Dystrophin panel

Specify:

Dystrophin rod domain (DYS1)  absent  reduced  cytoplasmic  normal

Dystrophin C terminus (DYS2)  absent  reduced  cytoplasmic  normal

Dystrophin N terminus of rod domain (DYS3)

absent  reduced  cytoplasmic  normal

Dystrophin (BMD Hotspot)  absent  reduced  normal

Spectrin  absent on necrotic fibers  normal

Utrophin  normal  increased at sarcolemma

Other stains for limb-girdle or congenital muscular dystrophy

Specify:

Laminin 2/Merosin (80 kDa)  absent  reduced  normal

Laminin 2/Merosin (300 kDa)  absent  reduced  normal

Alpha dystroglycan (VIA4)  absent  reduced  normal

Alpha dystroglycan (IIH)  absent  reduced  normal

Beta dystroglycan  absent  reduced  normal

Alpha sarcoglycan  absent  reduced  normal

Beta sarcoglycan  absent  reduced  normal

Delta sarcoglycan  absent  reduced  normal

Gamma sarcoglycan  absent  reduced  normal

Dysferlin  absent  reduced  cytoplasmic  normal

Emerin  absent  normal

Collagen VI  absent  reduced  normal

Caveolin 3  absent  reduced  normal

Integrin 7  absent  reduced  normal

nNOS  absent  reduced  normal

Inflammatory myopathy panel

CD3  absent  present in \_\_\_ % of lymphocytes

CD4  absent  present in \_\_\_ % of lymphocytes

CD8  absent  present in \_\_\_ % of lymphocytes

CD20  absent  present in \_\_\_ % of lymphocytes

CD138  absent  present in \_\_\_ % of lymphocytes

CD45  absent  present in \_\_\_\_% of mononuclear cells

CD68  absent  present in \_\_\_\_% of mononuclear cells

C5b-9  absent  present on endomysial capillary walls

cytoplasmic staining of necrotic fibers

Major Histocompatability Complex  absent  focal  diffuse

sarcolemmal  cytoplasmic

Protein aggregate myopathy panel

Desmin  normal  increased

Myotilin  normal  increased

B crystallin  normal  increased

Ubiquitin  normal  increased

1. **Additional immunohistochemical/immunofluorescence assays performed**: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_
2. **Other abnormalities noted on immunohistochemistry: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_**

**Epon-Embedded Tissue/Electron Microscopy**

1. **Abnormalities seen on:**  Light microscopy (Toluidine blue staining)  Electron microscopy

Both – Light microscopy and Electron microscopy

1. **Abnormalities noted in:**  Nuclei

Contractile apparatus

Sarcotubular organization

Mitochondria, specify (choose all that apply):

Abnormal size

Large  Small

Abnormal shape

Abnormal numbers

Abnormal location

Abnormal architecture

Basal lamina

Satellite cells

Intramuscular nerve twigs

1. **Describe any pathological inclusions noted:**   N/A \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

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1. **Describe any abnormal storage material identified:**  N/A \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

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## General Instructions

This form contains data elements that are collected when performing various muscle biopsies.

Important note: The data elements included in this CRF module span the range of diagnostic abnormalities seen in both pediatric and adult neuromuscular biopsy specimens. While each of these specific elements does not need to be included in every clinical biopsy report, this checklist provides a list of potentially pertinent positive and negative findings that should be considered when reporting a muscle biopsy. While the usefulness of these specific findings will depend on the differential diagnosis on a clinical case, all of these findings can be clinically important in specific situations. In cases where a specific diagnosis is not clear, it is recommended to evaluate and report the presence or absence of these findings to facilitate subsequent attempts to select biopsies for genetic testing or enrollment in research studies.

## Specific Instructions

Please see the Data Dictionary for definitions for each of the data elements included in this CRF module.

* Clinical History: These elements should be included, when available, to communicate the understanding the pathologist had of the participant/ subject’s clinical symptoms.
* Size of tissue collected –This information may not be available for autopsy tissue.