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. Age at presentation: ( ) years; ( ) months
2. Symptoms at presentation (check all that apply):

Weakness

Hypotonia

Muscle pain

Cardiac disease

Central nervous system disease

Unknown

1. Elevated creatine kinase:  Yes  No  Unknown (please specify) Value

## 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: (yyyy-mm-dd)
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 1 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 |

## Histological Findings in Muscle Biopsy or Autopsy Specimens

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

H and E

Gomori trichrome

NADH

SDH/COX

PAS

Oil Red O

ATPase 4.3

ATPase 4.6

ATPase 9.4

Other, specify:

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

Fatty replacement, mild

Fatty replacement, moderate

Fatty replacement, severe

Endomysial fibrosis, mild

Endomysial fibrosis, moderate

Endomysial fibrosis, severe

Myofiber degeneration, mild

Myofiber degeneration, moderate

Myofiber degeneration, severe

Myofiber regeneration

Abnormalities of fiber type

\*Specify: Type 1 predominance, specify % of Type 1 fibers

Type 2 predominance, specify % of Type 2 fibers

Fiber type grouping (of both fiber types)

Hypertrophic fibers

Atrophy/Hypotrophy

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

Specify:  Angulated  Round

Myopathy-associated pathological structures, specify:

Central nuclei

Specify estimated % of fibers (include eccentric nuclei):

Eccentric nuclei

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

Inclusion bodies/ Rimmed vacuoles

Nemaline rods

Specify:  Restricted to one fiber type, specify which:

Nuclear rods present

Ragged red fibers, Estimated number: (please specify)

COX- negative fibers, Estimated number: (please specify)

Central cores

Specify:  Structured  Unstructured

Minicores

Marked hypotrophy of type 1 fibers

Inflammation, mild  Inflammation, moderate  Inflammation, severe

Specify:

Perivascular

Specify:

Evidence of vascular damage

Thrombi identified in blood vessels

Diffuse

Associated with myofiber damage

Inflammatory cells identified

Specify (choose all that apply):

Lymphocytes

Neutrophils

Macrophages

Eosinophils (as a prominent component)

Microorganisms identified, specify which:

Abnormal storage material

Specify:

Abnormal cell types found in the biopsy

Specify which ones:

Excessive glycogen

Specify severity:  Mild  Severe

Excessive intracellular lipid

Specify severity:  Mild  Severe

Liver biopsy performed

Describe results:

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

Myosin immunohistochemistry

Dystrophin panel (list stains in question 4)

Other stains for limb-girdle or congenital muscular dystrophy (list stains in question 4)

Inflammatory myopathy panel (list stains in question 4)

1. Immunohistochemical/ Immunofluorescence assays performed: (please specify)

List name of antibodies used

Antibodies Used, data table

| Name of antibodies used: | Check if not known |
| --- | --- |
| Data to be entered by site |  |

1. List the Western Blot assays performed and corresponding results:
2. #1
3. Name of assay:
4. Result:
5. : #2
6. Name of assay:
7. Result:
8. Assays with normal immunoreactivity: (please specify)
9. Assays with reduced immunoreactivity: (please specify)
10. Assays with absent immunoreactivity: (please specify)
11. Other abnormalities noted on immunohistochemistry: (please specify)

## Epon-Embedded Tissue/Electron Microscopy (Muscle Biopsy/Autopsy Specimens)

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

Both – Light microscopy and Electron microscopy

1. Abnormalities noted in:  Contractile apparatus

Sarcotubular organization

Mitochondria, specify (choose all that apply):

Abnormal shape

Abnormal numbers

Abnormal location

Abnormal architecture

1. Describe any pathological inclusions noted, or indicate Not applicable:  N/A
2. Describe any abnormal storage material identified, or indicate Not applicable:  N/A

## 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.