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.