

APPENDIX 1
The Muscle-Biopsy Common Data Element—Revised (MB CDE-R) Form

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:** ___ years ___ months
4. **Symptoms at presentation (check all that apply):**
 Weakness
- Distribution
- | | | | | |
|--------------------------------------|--|--------------------------------------|--|-------------------------------|
| <input type="checkbox"/> Symmetrical | <input type="checkbox"/> Asymmetrical | | | |
| <input type="checkbox"/> Proximal | <input type="checkbox"/> Distal | <input type="checkbox"/> Limb-girdle | <input type="checkbox"/> Facioscapulohumeral | <input type="checkbox"/> Neck |
| <input type="checkbox"/> Paraspinal | <input type="checkbox"/> Finger flexor | <input type="checkbox"/> Other _____ | | |
- Other muscle symptoms
- | | | | |
|--|---|---|--|
| <input type="checkbox"/> Hypotonia | <input type="checkbox"/> Muscle pain | <input type="checkbox"/> Exercise intolerance | |
| <input type="checkbox"/> Episodic muscle pain/cramping | <input type="checkbox"/> Rhabdomyolysis | <input type="checkbox"/> Contractures | |
- Failure to thrive
- Respiratory difficulties
- Skin changes
- Eye symptoms
- Ptosis Ophthalmoplegia
- Joint laxity
- Clinical features of cardiac involvement/Known cardiac disease _____
- Central nervous system disease _____
- Exposure to toxins, supplements, or drugs (and relationship between exposure and biopsy acquisition) _____
- 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 laboratory data:**

