

[Study Name/ID pre-filled]

Site Name: _____

Date Form Completed: ___/___/___ (mm/dd/yyyy)

Subject ID: _____

Visit: [pre-populated by database]

1. Clinical Diagnosis of CMD to LGMD spectrum disorder? Yes No
2. CMD to LGMD spectrum diagnosis confirmed by muscle or skin biopsy? Yes** No*
 - a. *If No, recommended for skin biopsy? Yes No
 - b. *If No recommended for muscle biopsy? Yes No
 - c. **If Yes, date CMD to LGMD spectrum diagnosis confirmed by muscle or skin biopsy?
___/___ (MM/YYYY)
 - d. **If Yes, additional testing needed? Western blot IHC
3. Muscle MRI performed? Yes** No
**If Yes, date muscle MRI performed? ___/___ (MM/YYYY)
4. Genetic confirmation of CMD to LGMD spectrum disorder? Yes** No*
 - a. *If No, recommended for additional testing? Yes No
 - b. **If Yes, date CMD to LGMD spectrum diagnosis confirmed by genetic testing ___/___ (MM/YYYY)
 - c. Type of genetic testing performed:
 - Sequencing of genomic DNA
 - Sequencing of cDNA
 - SNP assay
 - CGH array
 - Next generation sequencing
 - Dedicated exome
 - Whole Exome
 - Whole Genome
 - d. Recommended for additional genetic testing for parents and siblings? Yes No
 - e. Would the family like to have blood and tissue samples banked for CMD research? Yes No
If Yes, referral to CMD BioBank? Yes No
5. Specify CMD to LGMD Subtype: _____ (see chart below)

CMD to LGMD Subtype
C = Collagen VI/Bethlem/Ullrich
D = Dystroglycanopathy
A7 = Alpha 7 integrin
L = Lamin A/C
M = Merosin deficient
S = SEPN1
U = Undiagnosed CMD
O = Other

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6. Which CMD gene? _____ (see below chart)

CMD Genes				
Col6A1	Fukutin	LMNA	SEPN1	DPM3
Col6A2	ITGA7	POMT1	DAG1	Unknown
Col6A3	LARGE	POMT2	DPM1	Other, specify
FKRP	LAMA2	POMgnt1	DPM2	

7. Muscle Immunohistochemistry (check all that apply):

- Alpha dystroglycanopathy with secondary merosin deficiency
- Alpha dystroglycanopathy with normal merosin
- Partial merosin deficiency
- Complete merosin deficiency
- Partial collagen VI deficiency
- Complete collagen VI deficiency
- Lamin A/C

8. Fibroblast Immunohistochemistry (check all that apply):

- Collagen VI - Decreased matrix expression
- Collagen VI - Intracellular retention

9. Western Blot (check if Abnormal, meaning reduced or absent):

- Alpha dystroglycan
- Merosin
- Collagen VI

10. Creatine Kinase Levels (only first and last tests are of interest)

Lab #	Collected?	Date Collected (MM/YYYY)	Age Collected	Result	Abnormal?	Normal Range
1	<input type="checkbox"/> Yes <input type="checkbox"/> No	____/____	[derived field]	_____ U/l	<input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	_____ U/l
2	<input type="checkbox"/> Yes <input type="checkbox"/> No	____/____	[derived field]	_____ U/l	<input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	_____ U/l

GENERAL INSTRUCTIONS

This form contains data elements that are collected from various diagnostic procedures used to assess progression of disease.

Responses to categories are obtained from health professionals performing the procedure and laboratory tests results.

SPECIFIC INSTRUCTIONS

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

- **Date of CMD to LGMD spectrum diagnosis first confirmed by muscle or skin biopsy** - The preferred format for recording date is MM /YYYY. 99/9999 can be used to indicate an unknown date.
- **Date CMD to LGMD spectrum diagnosis confirmed by genetic testing** - The preferred format for recording date is MM /YYYY. 99/9999 can be used to indicate an unknown date.
- **CMD to LGMD Subtype** – Choose only one, if known
- **CMD gene** – Choose only one, if known
- **Muscle Immunohistochemistry** – Check all that apply.
- **Fibroblast Immunohistochemistry** – Check all that apply.
- **Western blot** – Check if Abnormal, i.e. reduced or absent.
- **Creatine Kinase levels** – Only first and last tests performed are of interest.
- **Creatine Kinase levels date collected** - The preferred format for recording date is MM/YYYY. 99/9999 can be used to indicate an unknown date.
- **Creatine Kinase Age Collected** – This is a derived field based on the Date of Birth and Date Collected
- **Creatine Kinase Result** – International System of Units (SI) or American Units are preferred.
- **Creatine Kinase Normal Range** – International System of Units (SI) or American Units are preferred.