National Registry of Myotonic Dystrophy and Facioscapulohumeral Muscular Dystrophy Patients and Family Members

DM Physician Check List

☐☐☐☐☐☐☐☐ Pt Number ☐☐☐ Pt Initials ☐☐ ☐☐ ☐☐☐☐ Date Reviewed

Clinical Diagnostic Criteria for DM:

	Y	N	Not sure
a) Weakness of distal limbs	☐	☐	☐
b) Weakness of proximal hip girdle muscles	☐	☐	☐
c) Weakness of facial muscles	☐	☐	☐
d) Myotonia on EMG	☐	☐	☐
e) Grip or percussion myotonia	☐	☐	☐
f) Evidence of cataracts	☐	☐	☐
g) Facial or limb muscle weakness within the first 4 weeks of life	☐	☐	☐

Molecular Genetic Diagnostic Categories: (Check only one)

1) DNA test positive: CTG repeat >50 ☐ Size of Repeat ☐☐☐☐
2) DNA test negative: CTG repeat <37 ☐
3) DNA test positive: CCTG repeat >75 ☐ Size of Repeat ☐☐☐☐
4) DNA test negative: CCTG repeat <75 ☐
5) DNA test (DM-1 or DM-2) not done ☐
6) DNA test performed on relative ☐ Size of CTG Repeat ☐☐☐☐

Relationship to patient _____ Size of CCTG Repeat ☐☐☐☐

I. Definite DM-1 ☐
- **a. Symptomatic**
- **b. Asymptomatic**

I. Probable DM-1 ☐

III. Congenital DM-1/or childhood DM-1 ☐

IV. Definite DM-2 ☐
- **a. Symptomatic**
- **b. Asymptomatic**

V. Probable DM-2 ☐

VI. Congenital DM-2/or childhood DM-2 ☐

VII. DM like disease with normal DNA (DM-1/DM-2 excluded) ☐

VIII. Possible DM-1 or DM-2 ☐

IX. Unaffected blood relative
- **a. DNA Confirmed** ☐
- **b. DNA not done** ☐

Comments: _____

_____

_____

Signature: _____ **Date:** _____

I) Definite DM-1
DNA analysis of the size of the $[CTG]_n$ repeat size in the DM-1 gene on chromosome 19 is greater than 50 repeats in size (normal range of repeat sizes is 5-37 repeats).

Other definitions of definitely affected individuals include an individual with clinical signs of weakness and evidence of myotonia (clinical/EMG) who has:

- One parent with DNA proven DM-1
  OR
- A child with DNA proven DM-1

II) Probable DM-1
Individual who has not had DNA testing but has weakness of distal limb muscles plus weakness of muscles of swallowing and speech with evidence of myotonia (clinical and/or EMG).

III) Congenital DM-1 or childhood DM-1
Child with facial and limb muscle weakness that is apparent within the first 4 weeks of life and has positive DNA testing or who has a mother with definite DM-1 or probable DM-1.

IV) Definite DM-2
DNA analysis of the size of the $[CCTG]_n$ repeat size in the DM-2 gene on chromosome 3 is greater than 75 repeats in size (normal range is less than 75 repeats).

Other definitions of definitely affected individuals include an individual with clinical signs of weakness and evidence of myotonia (clinical/EMG) who has:

- one parent with DNA proven DM-2
  OR
- a child with DNA proven DM-2.

V) Probable DM-2
Individual who has negative DM-1 DNA testing, weakness of proximal hip girdle muscles, evidence of myotonia (clinical and/or EMG), and cataracts.

VI) Congenital DM-2 or childhood DM-2
Child with muscle weakness that is apparent within the first 4 weeks of life with positive DNA testing for CCTG repeat expansion at DM2 locus or who has a mother with definite DM-2 or probable DM-2.

VII) DM like disease with normal DNA (DM-1/DM-2 excluded)
Individual has a normal size $[CTG]_n$ repeat in the DM-1 gene and normal size $[CCTG]_n$ repeat in DM-2 gene, muscle weakness (as in possible DM-1 and DM-2), and evidence of myotonia (clinical and/or EMG).

VIII) Possible DM-1 or DM-2
Review of clinical records indicate some degree of muscle weakness (no specific distribution required) plus evidence of myotonia (clinical or EMG)

IX) Unaffected blood relative

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