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

DM Physician Check List

Pt Number Pt Initials Date Reviewed

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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 s4-1( )6(o)-4(f)3(d)-8.868 /P <</M0.0019TEMC /Pa

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