

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

## Physician Checklist FSHD

     

Reg ID Number

  

Initials

     

Date Reviewed

### CLINICAL DIAGNOSTIC CRITERIA FOR FSHD:

Y   N   Insufficient data

#### Inclusion:

a) Weakness of facial muscles

  

b) Either weakness of scapular stabilizers or foot dorsiflexors

  

#### Exclusion:

c) Presence of ptosis or weakness of extraocular muscles

  

d) Muscle biopsy in patient or affected relative with features suggesting an alternative diagnosis

  

e) EMG in patient or affected relative showing myotonia or neurogenic Changes

  

### CLINICAL DIAGNOSTIC CATEGORIES: (Check only one)

1. **Clinically definite:** all inclusion **Y** and exclusion criteria **N** met

2. **Clinically probable:**

- Inclusion criteria **a or b** met in addition to all exclusion criteria met
- Inclusion criteria met but exclusion criteria **d and/or e** not available

3. **Unaffected:** No clinical signs of muscle disease.

4. **Uncertain:** Muscle weakness present but clinical information insufficient

5. **Not FSHD:** not consistent with FSHD

### MOLECULAR DIAGNOSTIC CRITERIA FOR FSHD:

Subject's smallest allele on DNA test:   Kb. If >50, check here

DNA test performed on relative, check:

Relationship to subject: \_\_\_\_\_

Smallest allele on affected relative:   Kb. If >50, check here

### MOLECULAR GENETIC DIAGNOSTIC CATEGORIES: (Check only one)

A) DNA test positive: one allele  $\leq 35$  Kb

B) DNA test indeterminate: one allele >35-50 Kb

C) DNA test negative: all alleles >50 Kb

D) DNA test not done

**Diagnostic Classification:** (based on combined clinical categories 1-5 and molecular categories A-D; use diagnostic classification table below.)

**I. Definite 4q35 FSHD**

- a. Symptomatic (1A or 2A)
- b. Asymptomatic blood relative (3A)\*
- c. DNA positive but clinically uncertain (4A)
- d. DNA positive but clinically atypical (5A)

**II. Indeterminate 4q35 FSHD**

- a. Symptomatic (1B or 2B)
- b. Asymptomatic blood relative (3B)\*\*
- c. DNA indeterminate but clinically uncertain (4B)
- d. DNA indeterminate but clinically atypical (5B)

**III. Non-4q35 FSHD (1C or 2C)**

**IV. FSHD: 4q35 association unknown**

- a. Clinically definite (1D)
- b. Clinically probable (2D)

**V. Unaffected blood relative**

- a. DNA confirmed: DNA test negative (3C)
- b. DNA not done or unknown (3D)

**VI. Uncertain**

unknown (4C or 4D)

**VII. Not FSHD**

Clinically not consistent with FSHD;  
DNA negative or unknown (5C or 5D)

\* Must have blood relative with diagnostic classification Ia or Ib

\*\* Must have blood relative with diagnostic classification IIa or IIb

Diagnostic Classification Table					
Molecular Diagnostic Categories	Clinical Diagnostic Category				
	1	2	3	4	5
A	Ia	Ia	Ib	Ic	Id
B	IIa	IIa	IIb	IIc	IId
C	III	III	Va	VI	VII
D	IVa	IVb	Vb	VI	VII

**Inheritance Pattern:** (Check only one)

**A. Dominant**

- 1. DNA confirmed
- 2. By history, not DNA confirmed

**B. Non-dominant familial (affected siblings with unaffected parents)**

- 1. DNA confirmed
- 2. By history, not DNA confirmed

**C. Sporadic**

- 1. DNA confirmed
- 2. By history, not DNA confirmed

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**D. Uncertain**



**Comments:** \_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

**Signature:** \_\_\_\_\_ **Date:** \_\_\_\_\_

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