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

## FSHD PHYSICIAN CHECKLIST

Reg ID Number ☐☐☐☐☐☐ Initials ☐☐☐ Date Reviewed ☐☐ ☐☐ ☐☐☐☐

**CLINICAL DIAGNOSTIC CRITERIA:**

| | 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/affected relative suggesting alternative diagnosis | ☐ | ☐ | ☐ |
| e) EMG in patient/affected relative with 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 ☐

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**MOLECULAR DIAGNOSTIC CRITERIA:**

Smallest allele: ☐☐ kb If >38, check here ☐

A/B Variant: ☐

SMCHD1/DNMT3B mutation: ☐

D4Z4 hypomethylation (≤20 %): ☐☐ %

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DNA test performed on relative, check: ☐ Relationship to subject: _____

Smallest allele on affected relative: ☐☐ kb If >38, check here ☐

**MOLECULAR DIAGNOSTIC CATEGORIES:** (Check only one)

A) FSHD1: one allele ≤38 Kb ☐
B) FSHD2 ☐
C) FSHD1 DNA test negative (both alleles >38 Kb) ☐
D) DNA testing not done or incomplete ☐
E) Negative FSHD1 and 2 DNA testing ☐
F) FSHD1 and FSHD2 testing positive ☐

**DIAGNOSTIC CLASSIFICATION:** (Based on combined clinical categories 1 - 5 and molecular categories A - F; use Diagnostic Classification Table below.)

**I. Definite FSHD1**
- 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. Definite FSHD2**
- a. Symptomatic (1B or 2B) ☐
- b. Asymptomatic blood relative (3B)** ☐
- c. DNA positive but clinically uncertain (4B) ☐
- d. DNA positive but clinically atypical (5B) ☐

**III. Possible FSHD2**
Symptomatic (1C, 2C, or 4C) ☐

**IV. FSHD: 4q35 association unknown**
Symptomatic (1D or 2D) ☐

**V. Unaffected blood relative**
- a. DNA confirmed: DNA test negative (3C or 3E) ☐
- b. DNA not done or unknown (3D) ☐

**VI. Uncertain**
Unknown (4D) ☐

**VII. Not FSHD**
**Clinically not consistent with FSHD;**
DNA negative or unknown
(1E, 2E, 4E, 5C, 5D, or 5E) ☐

**VIII. FSHD1 & FSHD2**
- a. Symptomatic (1F, 2F, 4F, 5F) ☐
- b. Asymptomatic (3F) ☐

* 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 | III | VII |
| D | IV | IV | Vb | VI | VII |
| E | VII | VII | Va | VII | VII |
| F | VIIIa | VIIIa | VIIIb | VIIIa | VIIIa |

**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 ☐

**D. Uncertain** ☐

**COMMENTS:** _____

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_____

**SIGNATURE:** _____ **DATE:** _____

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