

CCEF Section 3: Clinical Diagnostic Form

	TYPICAL FEATURES	UNCOMMON FEATURES
1. ONSET OF MUSCLE WEAKNESS	<input type="checkbox"/> Facial weakness of orbicularis oculi or oris <input type="checkbox"/> Scapular weakness with altered ability to abduct arms <input type="checkbox"/> Humeral muscles (biceps/triceps)	<input type="checkbox"/> Distal lower limbs onset with triceps surae weakness <input type="checkbox"/> Distal upper limbs onset <input type="checkbox"/> Pelvic girdle onset
2. AXIAL MUSCLES INVOLVEMENT	<input type="checkbox"/> Hyperlordosis <input type="checkbox"/> Beevor's sign	<input type="checkbox"/> Camptocormia <input type="checkbox"/> Dropped head
3. FACIAL INVOLVEMENT	<input type="checkbox"/> Weakness of Orbicularis oculi (facial score ≥ 1) <input type="checkbox"/> Weakness of Orbicularis oris (facial score ≥ 1)	<input type="checkbox"/> Weakness of extra-ocular muscles <input type="checkbox"/> Weakness of masticatory muscles (persistent dysphagia)
4. SCAPULAR GIRDLE INVOLVEMENT	<input type="checkbox"/> Impairment of upper limb abduction with winged scapula or limitation of forward flexion (scapular FSHD score ≥ 1)	<input type="checkbox"/> Isolated distal upper limb muscle weakness <input type="checkbox"/> Impairment of arms abduction ($<90^\circ$) without winged scapula at rest and/or on attempted shoulder abduction or forward flexion
5. PELVIC GIRDLE INVOLVEMENT	-----	<input type="checkbox"/> Isolated and/or prevailing pelvic girdle muscle weakness
6. LOWER LIMBS INVOLVEMENT	<input type="checkbox"/> Weakness of tibialis anterior muscles weakness	<input type="checkbox"/> Early gastrocnemius and/or soleus atrophy/weakness
7. BLOOD CPK LEVEL (at least two samples 1 month apart)	<input type="checkbox"/> Normal range <input type="checkbox"/> $< 4x$ normal value (<1000 U/L)	<input type="checkbox"/> Value $> 4x$ normal value (>1000 U/L)
8. OTHER SIGNS	<input type="checkbox"/> Shoulders winging on attempted shoulder abduction or forward flexion <input type="checkbox"/> Horizontal clavicles <input type="checkbox"/> Forward sloping of the shoulders at rest <input type="checkbox"/> Sunken or flattened appearance of the chest <input type="checkbox"/> Atrophy of pectoralis muscles <input type="checkbox"/> Orbicularis oris hypokinesia during speech	<input type="checkbox"/> Myotonic phenomenon <input type="checkbox"/> Rippling <input type="checkbox"/> Eyelid ptosis <input type="checkbox"/> Extra-ocular muscle weakness <input type="checkbox"/> Early muscle contractures <input type="checkbox"/> Cardiomyopathy <input type="checkbox"/> Early respiratory insufficiency (Non Invasive Ventilation, NIV; FSHD score <12) <input type="checkbox"/> Pes cavus <input type="checkbox"/> Myoglobinuria

Importantly: Indicate the presence of comorbidities / results of previous injuries / illnesses that can possibly affect the neurological examination: _____

Extra-muscular involvement: hearing loss, epilepsy, retinal involvement, cognitive impairment