North Bristol NHS Trust

FSHD Clinical Proforma

(NHSE GTD code R74 / R345)

Name of patient (and name at birth, if different): ……

Date of Birth : ……. **Sex of patient: Male / Female** **Hospital ID/ NHS no**……..

Name of referring Doctor: ……..……..

Clinical specialty of referrer: Neurology/Paediatric Neurology ☐ Clinical Genetics ☐

Postal address of referring Doctor: ……...

Email address for sending results: ……

Billing Information: Who will pay for this test? .....

Full Address *and* email for billing: ……

Basis for testing

	Yes	No	Not known	Details/comment
Are there symptoms in the patient to be tested?				
Is this patient's clinical presentation typical for FSHD?				
Is this a primary Diagnostic test?				
Alternatively, do you wish to exclude FSHD as a possibility?				
Have the symptoms progressed over time?				**Age at first onset:**
Has another family member already been diagnosed with FSHD?				
If yes, how is the affected person related to this patient? (e.g. son of mother's uncle)				**Relationship:**
If possible, please can you provide the name and date of birth (DOB) of the affected relative(s)?				**Name and DOB:**
Is the clinical diagnosis in the relative supported by a typical positive DNA test result?				
Do you know which laboratory did the test in the relative? *Please provide a copy of the report if possible*				

Test requested:	Yes	No
FSHD1 primary test		
FSHD1 permissive haplotype analysis (4qA/4qB)		
FSHD2		

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Distribution of muscle involvement :

Please tick , or add Yes (Y) / No (N) / unknown (unk) , or right side (R) or left side (L) as appropriate

	Face	Shoulders / scapulae	Upper arm / elbow	Forearm/ wrists	Hand	Pelvic girdle	Knee	Lower leg or ankle
Current distribution of wasting or weakness								
Site of 1st symptom								
Symmetrically involved muscles								
Asymmetric involvement (e.g. R>L)								
Contractures								

Current facial & scapular weakness in more detail *(please tick or put Y/ N/ unk) :*

	Weakness / wasting present	No wasting / weakness
Peri-orbital (eye closure)		
Peri-oral (cheek puff)		
Ptosis (exclusion criteria)		
Extra ocular (exclusion criteria)		
Scapulae : - winging		
- overriding		
Limited abduction of arms		

If both scapular and pelvic girdle are affected, which was affected first? *(Please tick)*

Scapular then pelvic	Pelvic then scapular	not known

Please add any further available relevant information:

CK result (IU/l)	Muscle biopsy report	Other

Are you testing this patient's DNA for any other conditions simultaneously: if so, which ones?

Myotonic Dystrophy	EDMD X-Linked	EDMD Aut.Dom	Xp21BMD	SMA	Mitochondrial	LGMD	Nemaline Myopathy	Other

Please send Samples (DNA/EDTA blood) and Requests to:
Bristol Genetics Laboratory (SWGLH)
Pathology Sciences
Southmead Hospital
Bristol
BS10 5NB, UK

Contact

Tel: +44 (0)117 414 6168
Email: nbn-tr.geneticsenquiries@nhs.net