Bristol Genetics Laboratory

FSHD proforma for further DNA testing

Name of patient (and maiden name):……..

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

Referring Clinician: Name……..……..

Clinician specialty e.g. Neurologist/ Clinical Geneticist (please delete as appropriate)

Clinician's full address and email: ……

……

Billing Information: Will this bill be paid by the clinician or the patient? .....

Full Address for billing…….

Type of test.

Please fill in the relevant answer as appropriate :

	yes	no	n.k.	info.
Are there symptoms in the patient to be tested?				
Do you feel that this patient's presentation is typical for FSHD?				
Is this a Predictive test? i.e. Has another family member already been diagnosed with FSHD ?				
If so, how are they related to this patient? (e.g. maternal uncle's son)				
Can you, if possible, please provide their name and dob.				
Is the diagnosis in the relative supported by a typical DNA test result?				
If so, which lab did the test ?				
Is this a primary Diagnostic test?				
Alternatively, do you wish to exclude FSHD as a possibility?				
Age at first (retrospective) onset of symptoms: (e.g.…….years.)				
Test requested:				
FSHD1				
Permissive haplotype analysis				
FSHD2				

(Please continue on to page 2)

Distribution of muscle involvement :

Please tick , or add Yes (Y) / No (N) / not known (nk) , or R or L as appropriate

	Face	Shoulders or scapulae	Upper arm or elbow	Forearm or wrists	Hand	Pelvic girdle	Knee	Lower leg or ankle
Site of 1st symptom								
Current distribution of wasting or weakness								
Symmetrically involved muscles								
Asymmetric involvement (put 'R' or 'L' as weaker)								
Contractures								

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

	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 DNA for any other conditions simultaneously: if so, which ones?

Myotonic Dystrophy	EDMD X-Linked	EDMD Aut.Dom	Xp21 BMD	SMA	Mitochondrial	LGMD	Nemaline Myopathy	Other

PLEASE SEND REQUESTS TO
BRISTOL GENETICS LABORATORY
PATHOLOGY SCIENCES
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