NHS
South West Genomic Laboratory Hub

Clinical Proforma

Facioscapulohumeral muscular dystrophy (FSHD)
NHS England GTD code R74 / R345

1. Patient and referrer identification

First name of patient	Surname / family name of patient	Date of birth	Sex of patient

NHS / National health care ID number			
Full name of requesting doctor			
Clinical specialty of requesting doctor (please tick)	**Genetics**	**Neurology**	**Pediatric Neurology**
Clinic address of requesting doctor			
Email address to send test results			
Provider of payment for the test (if applicable)			
Email address to send invoice for payment (if applicable)			
Postal address to send invoice for payment (if applicable)			

2. Testing requested (please tick all requested)

Please contact the laboratory for current prices if applicable

FSHD1 diagnostic test (linear gel Southern blot)	
FSHD1 predictive / presymptomatic test (linear gel Southern blot) ***** Clinical Genetics request only *****	
FSHD1 haplotype test (linear gel Southern blot)	

FSHD2 D4Z4 DR1 methylation analysis (pyrosequencing)	
FSHD2 *SMCHD1* sequencing analysis	
If considering prenatal diagnosis for FSHD1, please contact the laboratory for discussion	

3. Family history

Is there a family history of clinical symptoms consistent with FSHD?	Yes	No	Not known
Is there a genetic diagnosis of FSHD in the family?	Yes	No	Not known
Details of family member with genetic diagnosis of FSHD *(please supply copy of laboratory results if possible)*			
Relationship to current patient			

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4. Clinical history of patient (for Diagnostic test requests only)

After clinical examination, do you consider that the patient has a typical presentation for FSHD?	
Age of patient at onset of symptoms	
Have symptoms progressed over time?	

Clinical features

	Yes	No	Asymmetric?
Scapular winging			
Facial muscle weakness			
Upper arm weakness			
Lower leg weakness			
Foot drop			
Hip / limb girdle weakness			
Abdominal muscle weakness			
Wrist / hand / finger weakness			

	Yes	No
Hearing loss		
Respiratory involvement		
Retinal involvement		
Cardiac arrythmia		
Other:		

Other investigations completed

	Results	Not done
Serum creatine kinase (CK)		
Muscle biopsy		
EMG		
Genetic testing for other neuromuscular disorders		

5. Consent for genetic testing

It is the referring clinician's responsibility to ensure that the patient/carer knows the purpose of the test and that the DNA sample will be retained in long term storage and may be used for future diagnostic tests. Completing this form is an indication that the clinician has obtained consent for testing and storage. The patient should be advised that the sample may be used anonymously for quality assurance and training purposes. Please advise us of any relevant restrictions.

6. Samples and contact details

Sample requirements	**FSHD1** Southern blot tests: >30 micrograms high molecular weight DNA in liquid format *OR* >5ml whole blood in EDTA, to be received at Bristol laboratory within 7 days after sampling **FSHD2** tests: sender's standard export quantity of DNA in liquid format *OR* whole blood in EDTA
Post address	Bristol Genetics Laboratory (SWGLH), Pathology Sciences, Southmead Hospital, Bristol, BS10 5NB, UK
Contact	Email: SWGLHenquiries@nbt.nhs.uk Telephone +44 (0)117 414 6168
Website	https://www.nbt.nhs.uk/south-west-genomic-laboratory-hub