



FAMILIAL/AUTOSOMAL DOMINANT HYPERCHOLESTEROLEMIA (FH) MOLECULAR GENETICS REQUEST FORM

Patient Information or Addressograph

First name: _____ Surname: _____

Patient address: _____

DOB: _____ Sex: _____ Hospital: _____

Ward/Clinic: _____ Hospital No. _____

whole blood EDTA sample required

SJH Laboratory Number

Referral Information

Name of Referrer: _____ Title/Position: _____ Consultant: _____

Address of the referrer: _____ Hospital: _____

Clinical History

Pre-treatment **total** cholesterol: _____ mmol/L

Pre-treatment **LDL** cholesterol: _____ mmol/L

Pre-treatment triglycerides: _____ mmol/L

Relatives genetically tested (Y/N) _____ If yes, indicate the performing laboratory: _____

Ethnic origin: _____ *Ethnic background may assist with interpretation of test results.*

LDLR gene Mutation (if applicable)

LDLR gene known mutation : _____ Probands relationship to patient: _____

Sample Information

Specimen Type: (Whole Blood, Buccal Swab, DNA) _____ Date Taken: _____

Date Received: _____ External Lab No: _____

Informed Consent Information

Patient has signed consent form? (Y/N) _____

Patient signature: _____

Please retain original consent in patient file and attach copy if possible

Minimum criteria required for testing

Please revert page and FILL IN Dutch Lipid clinic network & Simon Broome criteria. Please transfer scores below:

Does patient meet the following criteria?:

Dutch Lipid Clinic Network criteria for Definite FH (Y/N) _____

If yes, indicate score _____

Dutch Lipid Clinic Network criteria for Probable FH (Y/N) _____

If yes, indicate score _____

AND/OR

Simon Broome Criteria for Definite FH (Y/N) _____

Simon Broome Criteria for Possible FH (Y/N) _____

Requesting Clinician Signature: _____ MCRN no: _____

Please note that samples arriving without implied informed consent and completed questionnaire will not be processed

Appendix:**Dutch Lipid Clinic Network Criteria for diagnosis of Familial Hypercholesterolemia (FH) in adults**

	Please circle
	Score
Family history	
First-degree relative with known premature coronary and/or vascular disease (men <55 years, women <60 years) or First-degree relative with known LDL-cholesterol above the 95th percentile for age and sex	1
First-degree relative with tendinous xanthomata and/or arcus cornealis or Children aged less than 18 years with LDL-cholesterol above the 95th percentile for age and sex	2
Clinical history	
Patient with premature coronary artery disease (ages as above)	2
Patient with premature cerebral or peripheral vascular disease (as above)	1
Physical examination	
Tendinous xanthomata	6
Arcus cornealis prior to age 45 years	4
LDL-cholesterol (mmol/L)	
LDL-C ≥ 8.5	8
LDL-C 6.5–8.4	5
LDL-C 5.0–6.4	3
LDL-C 4.0–4.9	1
DNA analysis: functional mutation in the <i>LDLR</i> , <i>APOB</i> or <i>PCSK9</i> gene	8
PATIENT TOTAL SCORE	
STRATIFICATION	Total score
Definite FH	≥ 8
Probable FH	6–7
Possible FH	3–5
Unlikely FH	<3

Modified UK Simon Broome criteria

	Please circle points
Criteria	Point
DNA analysis: functional mutation in the <i>LDLR</i> , <i>APOB</i> or <i>PCSK9</i> gene	1
Tendon xanthomas in patient or first (parent, sibling, child) /second-degree relative (grandparent, uncle, aunt)	2
Family history MI (age <50 years) in second-degree or age <60 years in first-degree relative	3
Family history of cholesterol >7.5 mmol/L in first- or second-degree relative	4
Cholesterol >7.5 mmol/L (adult) or >6.7 mmol/L (age <16 years)	5
LDL-C >4.9 mmol/L (adult) or >4.0 mmol/L (age <16 years)	6
STRATIFICATION	Point
Definite FH	(5 or 6) + 1
Probable FH	(5 or 6) + 2
Possible FH	(5 or 6) + (3 or 4)

ST. JAMES'S HOSPITAL LABMED DIRECTORATE			
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Consent form for Diagnostic Genetic Testing on patient

MOLECULAR DIAGNOSTICS,
BIOCHEMISTRY DEPARTMENT, ST JAMES'S HOSPITAL, DUBLIN
Tel: +44 1 4162935

Patient name: _____

DOB: _____

1. I, _____, request that an attempt be made using genetic material (either DNA, RNA or both) to assess the probability that: I / my child (DELETE WHERE NOT APPLICABLE) might have inherited a disease-causing mutation in the gene(s) ***LDLR, APOB and PCSK9***, such mutations being associated with the medical condition ("disorder") **familial hypercholesterolaemia**.
2. In wishing to proceed with this test I have been fully informed about the test. I understand that the test will show **ONE** of the following:
 - a. **That I do have the disorder or carry a strong genetic susceptibility for the disorder and that other family members may therefore be at risk of developing this condition.**
 - b. **That I do not have the disorder**
 - c. **That the test results are indeterminate or difficult to interpret.**

Name: _____

Address: _____

Hospital: _____

Hospital registration number: _____

Signature of patient/parent/guardian: _____

Date: _____

For Medical Staff:

I have explained in detail to the above patient the principles and implications of genetic testing for the disorder. Given the clinical information available at this juncture I believe this test to be in the best interests of the patient.

Signature: _____ Date: _____

Name (Printed): _____

Medical Council registration number: _____