Biochemical Genetics,
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FH (familial hypercholesterolemia) GENETICS REQUEST FORM

Patient Information or Addressograph

First name: _____Surname: _____

Patient address: _____

_____

DOB::_____ Sex: _____

Ward/Clinic: _____ Hospital No._____

2 whole blood EDTA samples required

SJH Laboratory number

Referral Information:

Consultant's name: _____

Address of requesting consultant: _____ Hospital: _____

Name of referrer _____ Title/position: _____ Ext/Bleep: _____

Clinical Information:

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

Lipoprotein (a) if known: _____ nmol/L

Is patient on Lipid lowering treatment? (Y/N) _____ If yes state name of lipid medication, dosage and duration of treatment: _____

Current Total cholesterol: _____ mmol/L Current LDL cholesterol: _____ mmol/L

Current Triglyceride: _____ mmol/L

Does the patient have xanthomata? (Y/N) _____ If yes indicate location? _____

Ethnic origin: _____

Relative with known FH-causing variant? (Y/N) _____ If yes, provide known variant detail: _____

Informed Consent Information: *Please retain original consent form in patient file.*

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

Specimen Information:

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

Minimum criteria required for genetic testing:

Please revert page and <u>FILL IN</u> Dutch Lipid Clinic Network (DLCN) criteria. Please transfer scores below:

DLCN criteria score_____

Please note genetic analysis will be performed if DLCN score is ≥ 6 (Probable/Definite FH).

Requesting Clinician Signature: _____ MCRN no: _____
Date: _____

Please note that samples arriving without confirmed informed consent by signature and completed questionnaire will not be processed.

P.T.O

Appendix:

Dutch Lipid Clinic Network Criteria for diagnosis of Familial Hypercholesterolemia in Adults

Criteria	**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 (men <55 years, women <60 years)	2
Patient with premature cerebral or peripheral vascular disease (men <55 years, women <60 years)	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 *LDLR*, *APOB* or *PCSK9* gene	8
***Exclusive of each other (i.e. maximum 6 points if both clinical signs are present)**	
PATIENT TOTAL SCORE:	

STRATIFICATION	**Total score**
Definite FH	>8
Probable FH	6–8
Possible FH	3–5
Unlikely FH	<3

P.T.O

Consent form for Diagnostic Genetic Testing on patient

MOLECULAR DIAGNOSTICS,
BIOCHEMISTRY DEPARTMENT, ST JAMES'S HOSPITAL, DUBLIN
Tel: +353 1 4162935

Patient name:

DOB:

Address:

Hospital:

Hospital registration number:

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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 genetic predisposition for the medical condition ("disorder") **Familial Hypercholesterolaemia (FH)**. This includes testing for well documented disease-causing mutations in the gene(s) ***LDLR, APOB and PCSK9,*** and also an LDL-Cholesterol Genetic Risk Score (GRS), such variants being associated with an increased likelihood of an FH-like Clinical Phenotype and/or polygenic hypercholesterolaemia.

2. In wishing to proceed with this test I have been fully informed about the nature of the genetic tests involved. I understand that the test will show **ONE** of the following:

a. That I have a genetic variant predisposing to Familial Hypercholesterolaemia and that other family members may therefore be at risk of developing this condition
b. That I do not have genetic evidence of Familial Hypercholesterolaemia
c. That I carry a strong genetic susceptibility for developing a polygenic hypercholesterolaemia which can produce an FH-like clinical phenotype
d. That the test results are indeterminate or difficult to interpret

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: _____