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FH (familial hypercholesterolaemia) 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

Pre-treatment Triglyceride: _____ 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

Date Taken: _____ (for internal use only: Date received: _____)

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: Exclusive of each other (i.e. maximum 6 points if both clinical signs are present)	
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	
This patient has a causative variant in the *LDLR*, *APOB* or *PCSK9* gene	8
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

BIOCHEMICAL GENETICS,
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 testing is for disease-causing variants in the *LDLR* gene and also for two specific disease-causing variants in *APOB* and *PCSK9 genes* respectively. Further testing for genetic variants in other genes, which have also been strongly linked with causing FH in rarer circumstances, may be performed depending on an overall assessment of the clinical and biochemical phenotype, and the initial genetic test results.** In addition, testing for an established LDL-Cholesterol Genetic Risk Score (GRS), may be undertaken in some cases, such variants being associated with an increased likelihood of an FH-like Clinical Phenotype (FH-phenocopy) 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: _____