Familial Hypercholesterolaemia Genotyping Request Form

Version 8

Family number/Genetics record number .....

Full Name of Individual .....

Address .....

Post Code .....

Date of Birth .....

Sex *(tick)* Male Female

NHS Number | | | | | | | | | | |

Name of Consultant requesting test (BLOCK CAPS) .....

Signature .....

Contact address of requesting clinician/email .....

Contact telephone number of requesting clinician .....

Date of request .....

Full Address for return of report .....

Hospital .....

Fasting Lipid Profile

Date: .....

Total cholesterol ..... mmol/L

Triglyceride ..... mmol/L

HDL-cholesterol ..... mmol/L

LDL cholesterol ..... mmol/L

Was the patient taking any lipid lowering medication when this test was taken? YES / NO

If YES, state name of lipid medication and dosage .....

Pedigree - please draw in the box below

- Family pedigree should show 1st degree relatives as a minimum.
- If any family member has had blood taken previously for FH genotyping, even if result is not reported, it is important to state this.
- If you think there is a possibility that the patient has inherited FH from both parents and could be homozygous or compound heterozygote, please indicate this.

Clinical Diagnosis and narrative giving clinical reasons for genotype request…

Genotype scoring criteria for patients with a clinical diagnosis of FH **Please note these criteria only apply for index cases, not family members of known genotype positive patients.**		**Points** **(Please only circle a single highest score from each box)**
Family History	**1st/2nd degree relative:** • known with premature (<60yrs) CHD • known with premature (<45yrs) CHD • known with LDL-C > 4.9mmol/l (or total chol > 7.5mmol/l) • <18yrs with LDL-C > 4.0mmol/l (or total chol > 6.7mmol/l) ***Please specify relation to index case***	 1 2 1 2 ……
Physical Examination	• Tendon xanthomata (in patient **or 1st/2nd** degree relative) • Premature corneal arcus (no score for arcus senilis)	6 4
Clinical History	• Patient with premature CHD (<45 yrs) • Patient with premature CHD (<50 yrs) • Patient with premature CHD (<60 yrs) • Patient with premature (<60yrs) strokes and/or peripheral vascular disease	4 3 2 1
Untreated or corrected LDL- Cholesterol Concentrations (mmol/l)	• LDL-C ≥ 8.5 • LDL-C 6.5 – 8.4 • LDL-C 5.0 – 6.4 • LDL-C 4.0 – 4.9 ***If untreated LDL- C values are unobtainable see attached sheet (Correction Factor Table) and calculate estimated value.***	8 5 3 1
Fasting Triglycerides (mmol/l)	• Triglyceride 2.5 – 3.4 • Triglyceride 3.5 – 4.9 • Triglyceride ≥ 5.0 ***Please record in the narrative box any 2° causes that predispose to raised triglycerides, e.g. diabetes.***	minus 2 minus 3 minus 4

Eligibility for FH genotyping is based on total points score
(Only one score from each box)

6 or above eligible for genotyping

5 or less usually not unless exceptional circumstances (if applicable, please state these in the clinical diagnosis and narrative box)

Forms which are unclear, incomplete or not eligible for genotyping will be returned to the requesting clinician and the sample stored for at least 6 months for possible future use.

LDL-C Correction Factor Table for patients on cholesterol lowering medication

If untreated LDL-C levels are unobtainable, then the following table can be used to estimate untreated values. To achieve this, multiply the treated LDL-C value by the appropriate correction factor.

Statin / dose (mg)	Correction Factor
Ezetimibe	
10	1.2
Pravastatin	
10	1.2
20	1.3
40	1.5
Pravastatin + Ezetimibe	
10 + 10	1.5
20 + 10	1.6
40 + 10	1.7
Simvastatin	
10	1.4
20	1.6
40	1.7
80	1.9
Simvastatin + Ezetemibe	
10 + 10	1.9
20 + 10	2.0
40 + 10	2.3
80 + 10	2.4
Atorvastatin	
10	1.6
20	1.8
40	2.0
80	2.2
Atorvastatin + Ezetemibe	
10 + 10	2.0
20 + 10	2.2
40 + 10	2.2
80 + 10	2.5
Rosuvastatin	
5	1.8
10	1.9
20	2.1
40	2.4
Rosuvastatin + Ezetimibe	
10 + 10	2.5
20 + 10	2.7
40 + 10	3.3

N.B. The above figures are calculated from a number of different research projects and clinical trials.

CONSENT FORM FOR GENETIC TESTING

Patient Name:…… **D.O.B.** ……..

1. Test - I consent to my/my child's sample to be analysed for:.....

.....The test has been explained to me by .....I have had the opportunity to ask questions and I understand the implications of the test for me/my child and the rest of the family.

*Name of interpreter (if appropriate): .....

2. Sharing Information - I consent to the results of my/my child's test(s) to be shared with other relatives and healthcare professionals providing testing for my relatives Yes ☐ No ☐

3. Results (if applicable) - I understand that, where possible, the results of my/my child's test(s) will be shared with me/my doctors/health professionals involved in my/my child's care. If I am unable to receive the results of the test(s), I would like the results to be given to:

Name: .....Relationship to you: .....

Address: .....Telephone No: .....

Information

The following leaflet has been provided:.....

I understand that:

- I can change my mind at any stage and can choose not to receive the result.
- My/my child's sample may be used anonymously for the development of new tests and quality assurance.
- My/my child's sample will be stored in a DNA bank but no guarantee can be given that it will be available indefinitely.
- On occasion there can be a problem with the sample or test and a resample may be required delaying the result.
- The result may be difficult to interpret.
- Some genetic tests may occasionally reveal unexpected information e.g. information about paternity.

I have read and accept the above information.

....

Name of person giving consent **Date** **Signature**

If giving consent on behalf of a child please indicate your relationship to the child: .....

....

Name of person taking consent **Date** **Signature**

CONSENT TO BE CONTACTED FOR RESEARCH

We hope that you have found your contact with the All Wales Familial Hypercholesterolemia (FH) testing service to be helpful. As you may be aware, this is a service funded by the Welsh Government and the British Heart Foundation. Therefore, we are grateful for patient feedback about the service, and plan to contact you in the future to assess your satisfaction and find out whether it met your needs.

In addition, we would like to let you know about research related to FH, and give you the opportunity to be involved in such projects.

If you are happy for us to do this, please complete consent form below:

Please tick

I am happy to be contacted about research connected to the

All Wales FH testing service which has been ethically approved. ☐

Please complete or affix label:

Name: _____

Date of Birth: _____ Reference No: _____

Address: _____

_____

_____

Signature: _____**Date:** _____