Oxford University Hospitals NHS Foundation Trust

Central and South Genomic Laboratory Hub

Familial Hypercholesterolaemia (R134) Genetic Testing Request Form

All queries to: ouh-tr.fhservice.oxfordgenetics@nhs.net Tel.: 01865 226022
https://www.ouh.nhs.uk/services/referrals/genetics/genetics-laboratories/default.aspx

Attach patient label here

Name _____

Date of birth _____

NHS number _____

Referring clinician:

Specialty:

Contact details (email and/or tel. no.):

Please complete this section as fully as possible for all cases

CVD History

ACS/MI **Yes / No** Age___
CABG **Yes / No** Age___
PTCA **Yes / No** Age___
Angina **Yes / No** Age___
Stroke/TIA **Yes / No** Age___
PVD **Yes / No** Age___
Carotid artery intima–media thickness _____
Other_____

Ethnicity_____

Clinical signs of FH

Corneal arcus **Yes / No**
Xanthelasma **Yes / No**
Tendon xanthoma **Yes / No**

Lipids

Currently on statins? **Yes / No**
Dose _____
Pre-treatment levels:
Total chol. _____
LDL-c _____
HDL-c _____
Triglycerides _____

Referral criteria used and score obtained (circle appropriate and note score)
Simon Broome / Welsh / Dutch / Other (specify):

Family history of CVD, raised cholesterol etc. **Yes / No**
Details

Testing required (please tick box)

☐ **1) Diagnostic testing**
Includes:
- Full sequence analysis of *LDLR, APOB, PCSK9, LDLRAP1, APOE*
- Dosage analysis of *LDLR*
- Polygenic LDL-C-raising SNP score

Please note dosage analysis may be less reliable on DNA from buccal swabs. Blood is the preferred tissue type for this analysis.

☐ **2) Familial variant testing**
Testing for a known familial variant for:
Cascade testing of a pathogenic variant (**affected / unaffected – please circle**)
OR Segregation analysis of a variant of uncertain significance (**affected patients only**)

Index case name _____
Index case DOB _____
Relationship to this patient _____

Please provide a copy of the relative's diagnostic genetic report or as much information as possible regarding where and when testing was carried out, including the variant if known.