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NHS
Central & South
Genomic Laboratory Hub
Oxford Genetics Laboratories

Familial Hypercholesterolaemia (R134) Genetic Testing Request Form

Patient Details

NHS No:		**Sex*:**	
Surname:			
Forename:		**Address:**	
Date of Birth:			
Hospital:		**Postcode:**	
Ethnicity:		**Hospital No:**	

*Please state if karyotypic and/or phenotypic sex differ from given sex.

Requester Details

Clinician:		**Email:**	
Reporting Address:		**Invoice Address:**	

Clinical Information

CVD History		**Age**	**Clinical signs of FH**		**Lipids**	
ACS/MI	☐		Corneal arcus	☐	**Currently on statins?**	☐
CABG	☐		Xanthelasma	☐	**Dose:**	
PTCA	☐		Tendon xanthoma	☐	**Pretreatment levels:**	
Angina	☐				**Total cholesterol:**	
Stroke/TIA	☐				**LDL-c**	
PVD	☐				**HDL-c**	
Cartoid artery intima-media thickness:					**Triglycerides**	
Other:						
Referral criteria used:	Simon Broome: ☐ Welsh: ☐ Dutch: ☐ Other:				**Score:**	
Family history of CVD, raised cholesterol etc.						

Testing required

Diagnostic testing • 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.	☐	
Familial variant testing Testing for a known familial variant for either:	☐	
• Cascade testing of a pathogenic variant	☐	Affected: ☐ Unaffected: ☐
• Segregation analysis of a variant of uncertain significance	☐	Affected patients only
Index case name:		
Index case date of birth:		
Relationship to the 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		