## FAMILIAL HYPERCHOLESTEROLAEMIA GENE TEST REQUEST

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Essential Patient Demographics (Patient label can be used)			
Forename:	Surname:	Date of Birth:	
CHI No.:	Pedigree no. (if relevant):	Male / Female (Circle as appropriate)	
Address (must include postcode):			
		Postcode:	
Essential Referrer Details			
Referring Clinician(s):	erring Clinician(s): Department & Address for report:		
Email address (generic departmental where possible):			
Essential Sample Information			
Sample Type:	Date Taken:	Time Taken:	
High Risk: YES / NO If yes, please state risk Notify lab in advance if high risk	Urgent analysis required: YES / NO	DNA Storage only: YES / NO	)
INDEX CASES: see Diagnosing familial hypercholesterolaemia (guidelines)   Right Decisions Indicate how patient meets FH Wales Criteria: https://fhwalescriteria.co.uk/assistant Choose the highest relevant score in each box and add together. Score of 6 or above is eligible for genetic testing. Score of 5 or less not eligible unless exceptional circumstances.			
1 Family history ie 1 <sup>st</sup> or 2 <sup>nd</sup> degree relative with			
CHD aged <60 yrs CHD aged <45 yrs Adult: LDL cholesterol > 4.9 mmol/L (or total cholesterol > 7.5 mmol/L) Age <18 yrs: LDL cholesterol > 4.0 mmol/L (or total cholesterol > 6.7 mmol/L)			1 2 1 2
2 Physical examination  Tendon xanthomata (in patient or 1st /2nd degree relative)  Premature corneal arcus (<50 yrs)			6 4
Patient clinical history  Patient with premature CHD (<45yrs) Patient with premature CHD (<50yrs) Patient with premature CHD (<60yrs) Patient with premature CHD (<60yrs) Patient with premature (<60yrs) strokes and/or peripheral vascular disease  Patient untreated or corrected LDL-cholesterol (https://fhwalescriteria.co.uk/ldl_estimator)			4 3 2 1
Consider age of patient. Secondary causes should be excluded prior to genetic testing.  LDL-cholesterol 8.5 mmol/L or higher  LDL-cholesterol 6.5 to 8.4 mmol/L  LDL-cholesterol 5.0 to 6.4 mmol/L  LDL-cholesterol 4.0 to 4.9 mmol/L			8 5 3 1
5 Patient fasting triglycerides  Triglycerides 2.5 to 3.4 mmol/L  Triglycerides 3.5 to 4.9 mmol/L  Triglycerides 5.0 mmol/L or higher		-2 -3 -4	
Choose <u>ONE score only</u> per category (Score ≥6 eligible for genetic testing) Total			
Additional details if applicable			
Discussed with Clinical Genetics or Lipidologist? Y / N / NA If yes name of contact:			
CASCADE TESTING OF RELATIVES: complete where family variant has already been detected			
Pedigree reference no.:  Name of relative with known variant:  Variant details including lab. report reference:			
If index variant found in another lab please supply a copy of the report where possible			
CONSENT: It is the responsibility of the referring clinician to obtain informed consent from the patient / carer for the test and for the sample to be stored for future diagnostic testing.			
Referring Clinician Signature:	Print name:	Date:	