

**SCOTTISH MOLECULAR GENETICS CONSORTIUM**

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**FAMILIAL HYPERCHOLESTEROLAEMIA DNA TEST REQUEST FORM**

<b>Patient details</b> Name Address  Dob CHI Hospital No Family No	<b>Referrer details</b> Name Address for report  Contact No/E-mail  Specialty
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**PLEASE ENSURE PATIENT DETAILS ARE ALSO RECORDED ON SAMPLE**

*The following clinical details are required to enable us to develop the service in an evidence based manner targeted for our population, and are therefore required. Free testing is available for index cases who fulfil definite or probable Simon Broome criteria (see [www.nice.org.uk](http://www.nice.org.uk)), and are Scottish residents. Test requests in other circumstances should be discussed with the laboratory.*

**Simon Broome diagnostic criteria for index individuals**

Diagnose a person with **definite** FH if they have:

§ Total cholesterol >7.5mmol/l (>6.7mmol/l in child <16yrs) or LDL cholesterol >4.9mmol/l (>4.0mmol/l in child <16yrs) **Plus** tendon xanthomas in patient, or in a 1<sup>st</sup> or 2<sup>nd</sup> degree relative.

**OR**

§ DNA-based evidence of an LDL-receptor mutation, familial defective apo B-100, or a PCSK9 mutation.

Diagnose a person with **possible** FH if they have:

Cholesterol concentrations as defined above **and** at least one of the following:-

§ Family history of myocardial infarction: <50 yrs in a 2<sup>nd</sup> degree relative, or <60 yrs in a 1<sup>st</sup> degree relative.

§ Family history of raised total cholesterol: >7.5mmol/l in adult 1<sup>st</sup> or 2<sup>nd</sup> degree relative or >6.7mmol/l in child or sibling <16 yrs.

**TYPE OF TEST REQUESTED**

Mutation screen of LDL-R and common mutations in Apo B/PCSK9 for index case in family

Test for mutation already found in family (genetics access only)

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**FAMILY HISTORY**

Myocardial infarction <50yrs in a 2 <sup>nd</sup> degree relative or <60yrs in a 1 <sup>st</sup> degree relative	Y	N
1 <sup>st</sup> or 2 <sup>nd</sup> degree relative with total cholesterol >7.5mmol/l	Y	N
Chol >6.7mmol/l in child or sibling <16yrs	Y	N
1 <sup>st</sup> degree relative with tendon xanthomata and/or arcus cornealis	Y	N

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**PATIENT CLINICAL HISTORY**

Premature coronary artery disease (men <55yrs; women <60yrs)	Y	N
Premature cerebral or peripheral vascular disease (men <55yrs; women <60yrs)	Y	N

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**PATIENT PHYSICAL EXAMINATION**

Tendon xanthomata	Y	N
Arcus cornealis prior to age 45yrs	Y	N
Total cholesterol (fasting Y N )	<input type="text"/>	mmol/l
LDL-cholesterol (fasting Y N )	<input type="text"/>	mmol/l

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**PREVIOUS DNA RESULT IN FAMILY (if Y state how related to patient)**

Functional mutation in LDLR gene	Y...../N
Functional mutation in ApoB/PCSK9 gene	Y...../N

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**RULE OUT SECONDARY CAUSES OF HYPERCHOLESTEROLAEMIA  
(Please circle below. If abnormal state levels)**

Renal function	normal/abnormal.....
Liver function	normal/abnormal.....
Thyroid function	normal/abnormal.....

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**If patient does not fulfil Simon Broome criteria**, please write details below about history/presentation/reason as to why you think the sample should be tested and the lab will contact you to discuss further.