

APPROVAL FORM FOR FAMILIAL HYPERCHOLESTEROLAEMIA (FH) GENE TEST.

Genetic testing for familial hypercholesterolaemia (FH) is a very expensive test and the frequency of identification depends on the prior likelihood of finding a genetic cause. The risk scoring system used in Australasia is based on the Dutch Lipid Clinic Score. For Children the European Atherosclerosis Society guidelines are used. With rare exceptions, only requests where this score shows a high probability of FH are performed.

Please complete and email (3 pages below) to: ChemicalPathologist@adhb.govt.nz

1. This summary page (page 1)
2. DLCS scoring page (Adult or child) or cascade testing page (page 2, 4 or 5)
3. Consent page (page 6)

PATIENT DETAILS:

NHI..... Name DoB

Requestor..... Contact Phone/email

1. ☐ Patient has no identified likely secondary cause* **AND**

2. **One** of the following:

- ☐ Adult with Dutch Lipid Clinic Score (DLCS) of at least 6[†] (page 2)
- ☐ Under 18yr & meets the criteria in European Atherosclerosis Guidelines (page 4)
- ☐ Family member with genetically proven FH mutation (cascade testing) (page 5)
- ☐ other patients by pathologist pre-approval **AND**

3. ☐ Genetic testing consent form (page 6)

*Secondary causes to exclude: ketogenic diet, hypothyroidism, liver/renal disease, proteinuria, drugs (esp. high dose steroids, atypical antipsychotics, immunosuppressants)

[†]For online help with DLCS score, refer to Australasian Atherosclerosis calculator tool
<https://www.athero.org.au/fh/calculator/>

Note: We recommend all patients should also have lipoprotein (a) tested ONCE.

Primary care practitioners can request testing, but if in doubt we recommend discussion with a local specialist in lipid metabolism (Chemical Pathologist, Cardiologist, Metabolic Specialist, Endocrinologist).

You can contact the on call Chemical Pathologist through the regional hospital or Awanui.

ADULTS (AGE 18 OR MORE). NO KNOWN FAMILY MUTATION**Please complete all sections, including total score and clinical details**

NHI Name

DoB Date completed

ADULTS: Dutch Lipid Clinic Score (DLCS score) Circle Yes or No/NK (not known) <i>on each line</i> as applicable	CIRCLE NK = not known	SCORE (if Yes)
Family History First degree relative with premature coronary or vascular disease (men <55, women <60yr) OR LDL cholesterol >95 th percentile for age and sex (<i>please provide details in comments section</i>)	Yes No/NK	1
First degree relative with clinical features (tendon xanthoma, arcus senilis <45yr)	Yes No/NK	2
Patient History Early coronary artery disease (men <55, women <60yr) Early cerebral or peripheral vascular disease (men <55, women <65yr)	Yes No/NK Yes No/NK	2 1
Physical Exam Xanthomas (skin/tendons) Site: (Note: cholesterol deposits around the eyes do <u>not</u> qualify) Arcus cornealis if present before age 45 yrs	Yes No/NK Yes No/NK	6 4
<u>LDL</u> Cholesterol (mmol/L) (<i>if on statin, please correct the LDL by ratio using the table on next page</i>) ≥ 8.5 mmol/L 6.5-8.4 mmol/L 5.0-6.4 mmol/L 4.0-4.9 mmol/L < 4.0 mmol/L, or not known	RESULT and DATE	8 5 3 1 0
Clinical Details (Family History, Age/type of first event, other risk factors) If pre-approved by a Pathologist please provide name and discussion outcome.		TOTAL SCORE (sum)

LDL-cholesterol adjustment

To calculate the DLCN score the untreated* LDL-cholesterol concentration is required.

*prior to being treated with cholesterol-lowering medication

If this is unknown, then it can be calculated from the medicated concentration using the adjustment factor to the right.

To calculate the untreated LDL-cholesterol concentration adjusting for cholesterol medication.

**Untreated LDL-cholesterol =
medicated concentration x
cholesterol adjustment
factor for medication/dose**

Example:

A patient's LDL-cholesterol is 3.6mmol/L on Atorvastatin 80mg

Untreated LDL-cholesterol
= 3.6 x 2.150538
= 7.7mmol/L

Therefore the patient would have a LDL-cholesterol of 7.7mmol/L if they were not on medication.

	LDL-cholesterol adjustment factor
Atorvastatin 10mg	1.618123
Atorvastatin 20mg	1.763668
Atorvastatin 40mg	1.937984
Atorvastatin 80mg	2.150538
Rosuvastatin 5mg	1.709402
Rosuvastatin 10mg	1.872659
Rosuvastatin 20mg	2.070393
Rosuvastatin 40mg	2.314815
Rosuvastatin 80mg	2.624672
Simvastatin 10mg	1.37741
Simvastatin 20mg	1.492537
Simvastatin 40mg	1.636661
Simvastatin 80mg	1.818182
Ezetimibe 10mg	1.236094
Ezetimibe/simvastatin 10mg/10mg	1.855288
Ezetimibe/simvastatin 10mg/20mg	2.008032
Ezetimibe/simvastatin 10mg/40mg	2.252252
Ezetimibe/simvastatin 10mg/80mg	2.463054
Ezetimibe/atorvastatin 10mg/10mg	2
Ezetimibe/atorvastatin 10mg/20mg	2.173913
Ezetimibe/atorvastatin 10mg/40mg	2.173913
Ezetimibe/atorvastatin 10mg/80mg	2.5
Ezetimibe/rosuvastatin 10mg/10mg	2.48139
Ezetimibe/rosuvastatin 10mg/20mg	2.739726
Ezetimibe/rosuvastatin 10mg/40mg	3.333333
Pravastatin 10mg	1.251564
Pravastatin 20mg	1.322751
Pravastatin 40mg	1.422475
Other	1.43

Source: Australian Atherosclerosis Society website.

CHILDREN UNDER AGE 18 YR.

Please complete, including clinical details

NHI Name

DoB Date completed

CHILDREN: European Atherosclerosis Guidelines	Tick one	LDL Results & Dates (2 tests)
- LDL ≥ 5.0 mmol/L <i>no secondary cause</i> (thyroid, liver, renal disease, drugs). OR - LDL ≥ 4 mmol/L AND first degree relative with at least 1 of: - early CVD (men < 55yr, women < 65yr) - clearly raised LDL (adult ≥ 5.0 mM, child ≥ 4.0 mM) - clear clinical features (xanthomas, xanthelasma < 45yr) OR - LDL ≥ 3.5 mmol/L AND family history of first degree relative with genetically proven FH (details in page 5) OR - Request by Paediatric Metabolic Specialist or lipidologist outside criteria above (requires detailed reasons below)	☐ ☐ ☐ ☐	
Clinical Details (Family History, Age/type of first event, other risk factors)		

CASCADE TESTING. KNOWN FAMILY MUTATION.

Please provide details of at least one other known affected family member:

NHI..... Name DoB
Relationship to person being tested.....
LDL off treatment (mmol/L)
Mutation
Where was genetic testing done?

Additional family member(s) if known:

NHI..... Name DoB
Relationship to person being tested.....
LDL off treatment (mmol/L)
Mutation
Where was genetic testing done?

NHI..... Name DoB
Relationship to person being tested.....
LDL off treatment (mmol/L)
Mutation
Where was genetic testing done?

Background information



Consent for Genetic Testing and /or DNA Storage

Patient details

For WRGL use only

REC	
DATE / TIME	
SAMPLE	
VOL / CONDITION	
TEST REQUIRED	

PLEASE DO NOT PUT ANYTHING IN THIS BOX

Genetic testing may be used to establish a diagnosis or to determine risk status
Consent is given for:

Test: _____ / DNA Storage Only (circle)

Sample: Blood Urine Tissue Other _____

- The purpose of this genetic testing is:
 - For my care and treatment YES ☐ NO ☐
 - To understand implications for my whānau/family YES ☐ NO ☐
- If I am unable to receive results, they will be discussed with my representative or nominated person
- Genetic testing may reveal information about biological whānau/family relationships
- Genetic testing may reveal incidental findings that are unrelated to the condition being investigated
- GHSNZ may share genetic information to benefit whānau/family accessing genetic healthcare – unless noted overleaf.
- GHSNZ will not further share genetic information unless required or permitted to do so by law
- Genetic testing for some conditions may only be available overseas
- DNA will be stored in laboratories involved in this testing
- Stored DNA/tissue may be returned or destroyed on request
- Stored DNA may be used as a laboratory control sample or for Quality Assurance purposes
- Genetic testing may have insurance implications
- Consent for genetic testing can be withdrawn at any time prior to testing

I understand the information above and overleaf and I have had the opportunity to ask questions

Signed: _____ Date: _____
Patient/Parent/Representative

Signed: _____ Date: _____
Health Professional

Representative or nominated individual to be sent genetic results in the event I am unable to receive them

Name: _____ Relationship: _____

Address: _____ Telephone: _____

Authorised by Clinical Leadership Team: 11/8/2021
Genetic Health Service NZ - genetichealthservice.org.nz

Review date: August 2023

Completion of this form indicates:

- Agreement of patients and family members to be tested
- The demographic and genetic information be kept securely in a confidential registry. Patients have right to access/correct/remove related data