

APPROVAL FORM FOR FAMILIAL HYPERCHOLESTEROLAEMIA (FH) GENE TEST.

Midlands region – requests referred to Waikato Hospital

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 BOTH the following (TOTAL 2 PAGES for each patient):

1. *Patient details on this page* (all boxes must be checked) **AND, EITHER:**
2. If *no known family mutation*: FH risk scoring table (Table 1 for Adults, or Table 2 for Children <18yrs) **OR**
3. If *known family mutation*: Details of mutation and affected family member(s) for 'cascade screening'

Completion of this form indicates patient agreement to be tested and their information kept securely in a confidential registry. Patients have right to access/correction of data.

Please return **TWO PAGES** (this, and one other page as relevant) as a pdf attachment to the lab.

PATIENT DETAILS:

NHI..... Name dob

Requestor..... Contact Phone/email

1. The patient has no identified likely secondary cause (^see below) **AND**
2. **One** of the following (check which):
 - patient is an adult with Dutch Lipid Clinic Score (DLCS) of at least 6* **OR**
 - under 18yr & meets the criteria in European Atherosclerosis Guidelines **OR**
 - family member with a proven FH genetic mutation (cascade screening) **OR**
 - other patients by individual pathologist pre-approval (*details required*)
3. Patient has had lipoprotein (a) tested (once only) (result & date

^ Secondary causes to exclude: hypothyroidism, liver/renal disease, proteinuria, high dose steroids or immunosuppressants

*If in doubt we strongly recommend discussion with a local specialist in lipid metabolism (Chemical Pathologist at Waikato hospital or Endocrinologist at Waikato Diabetes Service)

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

+Please email TWO PAGES (THIS AND ONE OTHER PAGE) as an attachment with subject containing "FH Gene Test", NHI and "Attention chemical pathologist" to:

In Midlands region: Labadmin@waikatodhb.health.nz

TABLE 1: ADULTS (AGE 18 OR MORE). NO KNOWN FAMILY MUTATION

Please complete all sections, including total score and clinical details

NHI..... Name

d.o.b. 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 %ile 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)	Yes No/NK	2
Early cerebral or peripheral vascular disease (men <55, women <65yr)	Yes No/NK	1
Physical Exam Xanthomas (skin/tendons) Site	Yes No/NK	6
(Note: cholesterol deposits around the eyes do <u>not</u> qualify)		
Arcus cornealis if present at age <45 yrs	Yes No/NK	4
<u>LDL Cholesterol (mmol/L)</u> (<i>if on statin, please correct the LDL by ratio using the table on next page</i>)	RESULT and DATE	
≥ 8.5 mmol/L		8
6.5-8.4 mmol/L		5
5.0-6.4 mmol/L		3
4.0-4.9 mmol/L		1
< 4.0 mmol/L, or not known		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.

TABLE 2: CHILDREN UNDER AGE 18YR. NO KNOWN FAMILY MUTATION.

Please complete, including clinical details

NHI..... Name

d.o.b. Date completed

CHILDREN: European Atherosclerosis Guidelines	Tick one	LDL Results & Dates (2)
<ul style="list-style-type: none"> LDL \geq5.0 mM, no secondary cause (thyroid, liver, renal disease, drugs). <u>OR</u> 	<input type="checkbox"/>	
<ul style="list-style-type: none"> LDL \geq4 mmol/L AND first degree relative with early CVD (men <55yr, women <65yr) <u>OR</u> 	<input type="checkbox"/>	
<ul style="list-style-type: none"> LDL \geq3.5 mmol/L AND family history of first degree relative with genetically proven FH 	<input type="checkbox"/>	
<p>Clinical Details (Family History, Age/type of first event, other risk factors)</p>		

CASCADE TESTING. KNOWN FAMILY MUTATION.

Mutation details in family (gene and pathogenic variant)

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Please provide details of other known affected family members (at least one is required):

NHI..... Name dob

Relationship to person being tested.....

LDL level(mmol/L) Treatment (if any)

NHI..... Name dob

Relationship to person being tested.....

LDL level(mmol/L) Treatment (if any)

NHI..... Name dob

Relationship to person being tested.....

LDL level(mmol/L) Treatment (if any)

NHI..... Name dob

Relationship to person being tested.....

LDL level(mmol/L) Treatment (if any)

Background information

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