Sonic Genetics

Service available through Sonic Healthcare practices:

Douglass Hanly Moir Pathology
Sullivan Nicolaides Pathology
Melbourne Pathology
Barratt & Smith Pathology
Capital Pathology
Clinipath Pathology
Bunbury Pathology
Clinpath Pathology
Hobart Pathology
Launceston Pathology
North West Pathology
Southern.IML Pathology

Familial hypercholesterolaemia | Request form

FOR THE DOCTOR

Patient details

First name _____

Surname _____

Date of birth ____/____/____ Sex _____

Address _____

_____

_____

Phone (mobile) _____

Medicare No. ☐☐☐☐☐☐☐☐☐☐

PATIENT STATUS AT TIME OF SERVICE OR SPECIMEN COLLECTION
(Required by law for all patients) Was the patient a:

Private patient in a private hospital or approved day hospital?	☐ Yes	☐ No
Private patient in a recognised hospital?	☐ Yes	☐ No
Public patient in a recognised hospital?	☐ Yes	☐ No
Outpatient of a recognised hospital?	☐ Yes	☐ No

Test requested

Diagnostic test (Specialist only)

☐ Comprehensive panel (LDLR, PCSK9, APOB, LDLRAP1)
Please select criteria that apply:
- ☐ Dutch Lipid Clinic Network (DLCN) score of ≥6
- ☐ LDL-cholesterol level ≥6.5 mmol/L in the absence of secondary causes
- ☐ LDL-cholesterol level >5.0 mmol/L with signs of premature/accelerated atherogenesis
- ☐ None of the above (MBS rebates will not apply)

Familial cascade testing

☐ A copy of the relevant relative's report must be provided for testing to be performed.

Clinical information

Highest LDL-c: _____ (mmol/L) DLCN score: _____

_____

_____ ☐ SD

Genetic counselling REQUIRED sonicgenetics.com.au/consent

Informed consent is mandatory for testing to be performed

Written informed consent attested by the requesting specialist (see overleaf) ***OR***

Informed consent: (please select one)
- ☐ Sonic Genetics to complete
- ☐ Completed by an appropriately qualified specialist or certified genetic counsellor

Requesting doctor

Name _____

Address _____

_____

_____

Phone _____ Provider No. _____

I confirm that this patient has been counselled about the purpose, scope and limitations of the test and has given consent.

DOCTOR SIGNATURE

X Date

Copy reports to

Name _____

Address _____

_____

_____

FOR THE PATIENT – Patient and Financial Consent

I confirm that I have been informed about the purpose, scope and limitations of the test.

If I do not fulfil the Medicare criteria

ACCOUNT STATEMENT
I understand that as the test requested is not eligible for a Medicare rebate, I will pay in full prior to the sample being tested. Cancellation fees may apply.

If I do fulfil the Medicare criteria

MEDICARE ASSIGNMENT (Section 20A of the Health Insurance Act 1973):
I offer to assign my right to benefits to the Approved Pathology Practitioner who will render the requested pathology service(s) and any eligible pathologist determinable service(s) established as necessary by the practitioner.

PATIENT SIGNATURE

X Date

Practitioner's Use Only (Reason for patient being unable to sign)

For pricing, please refer to our website – sonicgenetics.com.au
For any enquiries, please contact Sonic Genetics on 1800 010 447.

FOR THE COLLECTOR

I certify that I established the identity of the patient named on this request, collected and immediately labelled the accompanying specimen(s) with the patient's name, DOB and date/time of collection.

Collector initials	☐ 1 x 4 mL EDTA	Patient initials
Location code	Date collected / /	PAY CAT
Collection type	Time collected :	

Your doctor has recommended that you use one of the subsidiaries affiliated with Sonic Healthcare Limited, an Approved Pathology Authority. You are free to choose your own pathology provider. However, if your doctor has specified a particular pathologist on clinical grounds, a Medicare rebate will only be payable if that pathologist performs the service. You should discuss this with your doctor.

Sonic Genetics

Familial hypercholesterolaemia

Information for patients

Purpose

Genetic testing for familial hypercholesterolaemia (FH) is used to provide a definitive diagnosis of this condition. Having a genetic diagnosis of FH allows for more targeted clinical management, access to medications on the Pharmaceutical Benefits Scheme (PBS) and testing of relevant family members to determine their need for clinical management and monitoring.

Diagnostic testing is performed for a person with clinical features suggestive of FH. These clinical features include a high level of cholesterol in the blood, a family or personal history of high cholesterol and cardiovascular events, and other features associated with excess cholesterol. Diagnostic testing will analyse genes known to cause FH. If an abnormality is found, a genetic diagnosis of FH is confirmed.

Cascade testing (predictive testing) is performed for a first- or second-degree biological relative of a person with a genetic diagnosis of FH. This relative may not have any clinical features of FH. If they have the abnormal gene, they will develop clinical features of FH in the future if untreated. By testing for the presence of the abnormal gene already identified in the family, relatives who have FH can be identified and offered early and effective treatment. Cascade testing only tests for the abnormal gene already identified in the family.

Limitations

There are currently more than 2,000 different genetic abnormalities that cause FH. Our clinical and scientific experts use advanced technology and computer analysis to identify abnormalities in the four genes commonly associated with FH: LDLR, PCSK9, APOB and LDLRAP1. The interpretation of the genetic analysis may be dependent on your clinical features and family history.

The genetic test may identify a genetic change that is not definitely abnormal. These genetic changes are called 'variants of uncertain significance', or VUS. If a VUS is found, this does not provide a definitive diagnosis of FH. A VUS cannot be used for cascade testing of family members.

Approximately 20% of individuals with definite clinical features of FH do not have a genetic abnormality identified in the FH genes, that is, the genetic diagnosis of FH cannot be confirmed. Hypercholesterolaemia still needs to be treated, even if there is not a genetic cause identified. A doctor can determine if there are further tests, lifestyle changes or medications recommended in this situation.

Genetic counselling

Your doctor will be able to provide you with information and advice regarding this test. Your doctor may also recommend that you seek genetic counselling before or after the test.

A genetic counsellor provides information and support to patients as they make decisions about their genetic health. Sonic Genetics can provide professional genetic counselling via phone or video link before and/or after testing on request by your doctor. There is no additional cost associated with this service.

Family communication

If you receive a genetic diagnosis of FH, it is important that this information be shared with relevant family members so they can decide whether they wish to have cascade testing. Cascade testing is recommended for first-degree relatives (parents, siblings, children) and second-degree relatives (grandparents, grandchildren, aunts, uncles, nieces and nephews). Children should be considered for FH cascade screening by the age of 10 years. Early diagnosis and appropriate clinical care in relatives reduces the risk of serious complications, through early and effective treatment.

Sharing this information with relatives can be difficult. Sonic Genetics is available to help you share this information with the family. We can provide a letter to give to each relative, and we can (with your assistance) send a letter to the relevant relatives. If you have a genetic diagnosis of FH, and have given consent to Sonic Genetics, we will get in touch with you to discuss this.

Results

Diagnostic testing

- If an abnormal gene is detected, this confirms you have FH. Your doctor will recommend further tests and treatment. Cascade testing of family members is recommended.
- If no abnormal gene is detected, the genetic diagnosis of FH cannot be confirmed. However, it is still important that your doctor recommends tests and treatment for your raised cholesterol and other clinical features. Cascade testing is not available to your family members; it may be appropriate for them to have their cholesterol level checked.

Cascade testing

- If you have inherited the abnormal gene, this confirms that you have a genetic diagnosis of FH. Your doctor will recommend further tests and treatment. Cascade testing of family members is recommended.
- If you do not have the abnormal gene, you do not have a genetic diagnosis of FH. Your doctor may still need to check your cholesterol level, and will advise you if any further tests or treatment are recommended, according to general cardiovascular risk guidelines.

Results will be delivered to your doctor, typically within 4–6 weeks. Your doctor can contact our genetic pathologists and senior medical scientists to discuss your result.

Non-rebated testing

Non-rebated testing is available for patients who are willing to cover the full cost of the test themselves. It is important that such patients seek advice from a doctor experienced with lipid disorders to ensure that the test will be worthwhile.

Further information about FH genetic testing is available on our website;
sonicgenetics.com.au/fhp

Patient and Financial Consent

This genetic test has been ordered by your doctor following a discussion with you. You have access to a number of sources of information about this test, including your doctor, the Sonic Genetics website, a genetic counsellor and this information. Before the test is performed, you have the opportunity to ask questions, seek further information, and request genetic counselling. If a genetic abnormality is detected, this may be important information to share with family members. They may have the same genetic abnormality and develop familial hypercholesterolaemia. Genetic tests are unable to identify every genetic abnormality that can cause a disease. A person may have a genetic condition even if a genetic abnormality is not found. For this reason, a genetic test result must be considered in the light of other information that a doctor has about a patient's health and history. Medicare covers all or part of the cost of some genetic tests under certain circumstances. Your doctor will advise what fee (if any) you will need to pay.

Medicare criteria (as of July 2021)

Indication	Item #	MBS rebate requirements
Testing an individual with clinically confirmed or suspected FH	**73352**	Characterisation of germline variants causing familial hypercholesterolaemia (which must include the LDLR, PCSK9 and APOB genes), requested by a specialist or consultant physician, for a patient for whom no familial mutation has been identified, and who has any of the following: a Dutch Lipid Clinic Network score of at least 6; an LDL-cholesterol level of at least 6.5 mmol/L in the absence of secondary causes or an LDL-cholesterol level of between 5.0 and 6.5 mmol/L with signs of premature or accelerated atherogenesis. Applicable only once per lifetime.
Cascade testing of family members	**73353**	Detection of a familial mutation for a patient who has a first- or second-degree relative with a documented pathogenic germline gene variant for familial hypercholesterolaemia. Applicable only once per lifetime.