Appendix 3 Cascade Testing Package

Familial Hypercholesterolaemia Clinical Support Service
Royal Prince Alfred Hospital

LIPID SPECIALIST **GENETIC COUNSELLORS** **RESEARCH NURSES**

Dear Doctor,

Your patient has recently been identified as being at risk of having Familial Hypercholesterolaemia (FH). GPs can arrange MBS-funded (Item 73353) genetic testing for relatives of individual's confirmed to have FH, referred to as "cascade testing". Below are some key points to assist with arranging this testing.

When to arrange FH cascade testing:
1. All first and second degree relatives of an individual with genetically confirmed FH are eligible for testing of the family variant (cascade testing)
2. Genetic testing can be helpful to determine whether to commence lipid-lowering treatment
3. Genetic testing is available for children as treatment can commence from around age 10
4. Genetic testing is optional but all at risk relatives should have their LDL-cholesterol levels checked.

Points to discuss with your patient considering genetic testing for FH:
1. What is FH – FH causes high levels of LDL-cholesterol and when left untreated increases the risk of cardiovascular disease
2. FH is a hereditary condition – FH is autosomal dominant, meaning first degree relatives are at 50% risk and second degree relatives are at 25% risk of having FH
3. Potential insurance considerations for genetic testing (link to further information over page)
4. All patients undergoing genetic testing require written consent, please use the form provided.

How to arrange FH cascade screening:
Option A – Blood Collection
1. We have enclosed a prefilled request form. Please complete with your patient's details and the relative's report reference number: MD-XX-XXXXX
2. The patient should be made aware that the request form also includes an LDL-cholesterol check
3. The patient should take the completed form to a **hospital pathology service** to ensure they are not charged for the collection.

Option B – Saliva Collection
1. FH testing can also be done with a saliva sample. You or your patient can request a saliva collection kit by contacting RPA Hospital Vascular Health Clinic
2. We have enclosed a prefilled request form. Please complete with your patient's details and the relative's report reference number: MD-XX-XXXXX. Please cross out the LDL-cholesterol check as this cannot be performed on a saliva sample. If you wish to also check your patient's LDL-cholesterol this will need to be done on a blood sample
3. The request form can either be sent with the completed saliva collection kit directly to RPA Hospital Molecular Genetics, or you can email/fax the form separately.

If you choose to use a private pathology service, please Cc RPA Hospital Vascular Health Clinic in the results at [clinic email address]

Receiving and delivering results:
1. Turnaround time for FH genetic testing is approximately 2-3 months
2. Once the result is available, please arrange an appointment to discuss this with your patient:
   a. Family variant identified – genetically confirmed to have FH. Management guidelines available at HealthPathways https://sydney.communityhealthpathways.org/
   b. Family variant not identified – not confirmed to have FH. Manage as per general population.

If you would like additional support, please contact your local genetics service or one of our genetic counsellors at the RPA Hospital Vascular Health Clinic on [phone number] or email [address]

Yours sincerely,

FH Clinical Support Service

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Royal Prince Alfred Hospital

LIPID SPECIALIST **GENETIC COUNSELLORS** **RESEARCH NURSES**

Familial Hypercholestorolaemia Resources for Providers

Several resources are available to assist GPs in identifying and managing FH which include:

- FH Health Pathway - https://sydney.communityhealthpathways.org/
- An online calculator for the Dutch Lipid Clinic Network (DLCN) criteria score for phenotypic diagnosis can be accessed through the Australian Atherosclerosis Society Calculator at - https://www.athero.org.au/fh/calculator/
- Centre for Genetics Education FH Factsheet - https://www.genetics.edu.au/PDF/Familial_hypercholesterolaemia_fact_sheet-CGE.pdf
- Insurance Considerations with Genetic Testing in Australia - https://www.genetics.edu.au/SitePages/Life-insurance-products-and-genetic-testing-in-Australia.aspx
- The National FH Registry – an electronic database where FH patients can provide consent for their medical information, family history and other related information to be collected for research purposes - https://www.athero.org.au/fh/wp-content/uploads/FH-Registry-Brochure_v4.pdf

Appendix 3 Cascade Testing Package

Familial Hypercholesterolaemia Genetic Testing

Patient Details

Surname First Name

MRN Date of birth Sex

Street Address Phone

Requesting Practitioner Information

Surname Initials Telephone

Address Email

Fax

I confirm that the patient has been informed of the process, scope and limitations of this test, and that the patient is aware they may receive a bill if they do not fulfil the Medicare rebate criteria.

Signature Provider number

Preference for delivery of results: _ Mail _ Fax _ Email

Copy To

Medicare Authorisation

TO BE COMPLETED BY THE PERSON ASSIGNING BENEFITS FOR THE SERVICES ON THIS FORM. I understand that my medical practitioner has requested test(s) that may not be covered by Medicare, for which I may receive an account which I will pay in full. I understand that I will receive an invoice from the pathology service performing this test which may be a different laboratory who reported the original pathology. I agree to accept responsibility for the full payment of the fees for the test[s] that are not rebatable by Medicare. The genetic testing may involve more than one test.

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's Signature: ..... Date: .....

Medicare Number: ☐☐☐☐☐☐☐☐☐☐

Practitioner use only:
(Reason patient cannot sign) .....

☐ Verbal consent was provided by patient to submit unpaid account to Medicare.

☐ A private patient in a private hospital, or approved hospital facility
A public patient in a recognised hospital

☐ An outpatient of a recognised hospital
A private patient in a recognised hospital

Sample Requirements

Please collect two independent samples, one 4 ml EDTA tube each from two separate venepuncture time points, five minutes apart e.g. one at 1310 hrs and then another at 1315 hrs.

Sample One (1 x 4 mL EDTA)

Collection Date Collection Time Collector Signature

Sample Two (1 x 4 ml EDTA)

Collection Date Collection Time Collector Signature

Send To:

Your doctor has recommended that you use NSW Health Pathology. 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. Accredited for compliance with NPAAC Standards and ISO15189.

Appendix 3 Cascade Testing Package

Familial Hypercholesterolaemia Genetic Testing

Clinical Details

Total Cholesterol LDL Cholesterol Triglycerides

Lipid Lowering Treatment and Adherence *(at time of sample collection)*

Clinical Information *(including pedigree and any genetic results for affected family members)*

Test Requested

☐ **Comprehensive Analysis of FH Genes *LDLR, APOB, PCSK9***

Patient 1st the first individual in the family having genetic testing for FH (item 73352).

Must be ordered by a **Consultant Physician.**

Dutch Lipid Clinic Network Score (required) *see https//www.othero.org.au/fh/colculator/*

- ☐ Patient has no previously identified FH familial variant AND
- ☐ Dutch lipid Clinic Network score of 6 and above OR
- ☐ LDL-c, holesterol of 6.5 mmol/L or above in the absence of secondary causes OR\
- ☐ In the presence of premature or accelerated atherosclerosis, LDL-cholesterol of 5 mmol/L or above

X **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.**

Cascade testing (item 73353). May be ordered by **GP or Consultant Physician.** Please attach copy of family member's genetic report (familial variant) and pedigree (relationship to family member).

Appendix 3 Cascade Testing Package

NSW GOVERNMENT Health

Facility:

FAMILY NAME	MRN
GIVEN NAME	__ MALE __ FEMALE
D.O.B. _ _ /_ _ /_ _ M.O.	
ADDRESS	
LOCATION / WARD	
COMPLETE ALL DETAILS OR AFFIX PATIENT LABEL HERE	

CONSENT: GENETIC TESTING

(for all types of genetic and genomic testing for ADULTS, MATURE MINORS and MINORS

CONSENT FOR GENETIC TESTING is provided by (please tick an option below):

__ **An adult** (a patient with capacity)

__ **A mature minor** (a patient with capacity)
I *(the health practitioner)* have assessed this patient to be a minor with capacity to give consent as they have demonstrated sufficient maturity and intellect to fully understand what is proposed.

__ **A parent / guardian of a minor without capacity**

PROVISION OF INFORMATION TO PATIENT/ PARENT/ GUARDIAN To be completed by Health Practitioner

_____
INSERT NAME OF HEALTH PRACTITIONER

have discussed with *this patient/parent/guardian* the reason for conducting the proposed genetic test*. I have informed *this patient/parent/guardian* of the nature, possible results, limitations and material risks of the proposed genetic test*, as confirmed on this form by this *patient/parent/guardian.*
This patient/parent/guardian has been offered additional written information and/or reference to on line resources about the genetic testing.

Genetic testing is being conducted for Familial hypercholesterolaemia

_____
INSERT NAME OF CONDITION(S) OR CLINICAL INDICATIONS

***TYPE OF GENETIC TEST (please tick an option below):**

__ **Carrier Testing:** a genetic test performed on a person to identify if they carry a gene change.

__ **Diagnostic Testing:** a genetic test performed on a person to identify a specific genetic condition.

X **Predictive/Presymptomatic Testing:** a genetic test performed on a person with a family history of a genetic condition, who does not usually have symptoms at the time of testing, to determine if they have inherited that condition or susceptibility to that condition.

Prenatal Testing: a genetic test to identify possible genetic conditions in an unborn baby.

Other (please s p e c i f y) : -

_____

INTERPRETER PRESENT _ Yes __No

_____ _____
INSERT NAME OF INTERPRETER SIGNATURE

DATE _____ TIME **AM/PM** _____
EMPLOYEE ID / PROVIDER NUMBER

_____ _____ / / / / / /
SIGNATURE OF HEALTH PRACTITIONER DATE

NO WRITING

Page 1 of 2

CONSENT: GENETIC TESTING
(for all types of genetic and genomic testing for ADULTS MATURE MINORS and MINORS)

Appendix 3 Cascade Testing Package

NSW GOVERNMENT Health

Facility:

CONSENT: GENETIC TESTING

(for all types of genetic and genomic testing for ADULTS, MATURE MINORS and MINORS)

FAMILY NAME	MRN
GIVEN NAME	__ MALE __ FEMALE
D.O.B. _ _ /_ _ /_ _	M.O.
ADDRESS	
LOCATION / WARD	

COMPLETE ALL DETAILS OR AFFIX PATIENT LABEL HERE

PATIENT / PARENT / GUARDIAN CONSENT

To be completed by Patient / Parent / Guardian

I understand and acknowledge that:

- A blood, saliva or tissue sample will be used to test DNA;
- I will be told the results by a health practitioner;
- This is not a "general health test";
- Results are based on current knowledge that may change in the future;
- This test will not predict all future health problems;
- I can change my mind about having the test performed or about receiving genetic test results at any time by contacting the health practitioner;
- There are a number of different possible results from the testing and these can have implications for *me/my child* and *my/my child's* family;
- The results may be of "unknown or uncertain significance", which means they cannot be understood based on current knowledge;
- There is a chance that some genetic tests could identify other medical conditions (or susceptibility to other medical conditions) as an incidental finding;
- The genetic test results may identify unexpected family relationships;
- The genetic test results may affect *my/my child's* ability to obtain some types of insurance (for example, life insurance);
- Further testing may be needed to finalise the result;
- The reason for testing and the potential benefits, consequences and limitations involved in the testing have been explained in a way I understand;
- I have had an opportunity to discuss the information, ask questions and have any concerns addressed and I am satisfied with the explanations and answers to my questions;
- *My/my child's* results are confidential and will only be released with my consent or as required or permitted by law.

RELEASE OF GENETIC TESTING RESULTS (please tick YES or NO)

► *My/my child's* test results can be shared with relevant health practitioners involved in the care of *my/my child's* family members (genetic relatives): __ Yes __ No

Genetic relatives are people who are related to an individual by blood, for example, a sibling, parent or descendant of the individual.
Please note: Genetic information can be used and disclosed without consent in order to lessen or prevent a serious risk to the life, health or safety of a genetic relative no further removed than third degree; and, only where the disclosure is made in accordance with the guidelines issued by the Information and Privacy Commission NSW

► If I cannot be contacted, details of *my/my child's* test results can be released to a nominated individual: __ **Yes** __ **No**

Please provide contact details for an appropriate person:

Name: _ Phone: _ _ _ _ _ _ _ _ _ _ _

Relationship to Patient: -

ADULT AND MATURE MINOR CONSENT (a patient with capacity)

I consent to genetic testing as discussed with _
INSERT NAME OF HEALTH PRACTITIONER

_____ INSERT NAME OF PATIENT _____ SIGNATURE OF PATIENT _____ DATE

PARENT/GUARDIAN CONSENT (a parent / guardian of a minor without capacity)

I consent to genetic testing as discussed with _
INSERT NAME OF HEALTH PRACTITIONER

for _ _ _ _ _ _ _ _ _ _ _ _ _ _ _ _ _ _ _
INSERT NAME OF MINOR

_____ INSERT NAME OF PARENT/GUARDIAN _____ SIGNATURE OF PARENT/GUARDIAN __ __ /__ __ DATE

_____ RELATIONSHIP TO MINOR OF PARENT/GUARDIAN _____ ADDRESS

NO WRITING

Appendix 3 Cascade Testing Package

Familial Hypercholesterolaemia

△ inherited (runs in families) △ high △ cholesterol △ in your blood

Familial Hypercholesterolaemia (FH) is an inherited condition that causes high levels of 'bad' (LDL) cholesterol starting at birth.

FH is not caused by an unhealthy lifestyle. FH is caused by a 'faulty' gene which is passed from parent to child. This 'faulty' gene stops 'bad' cholesterol from being removed from the blood.

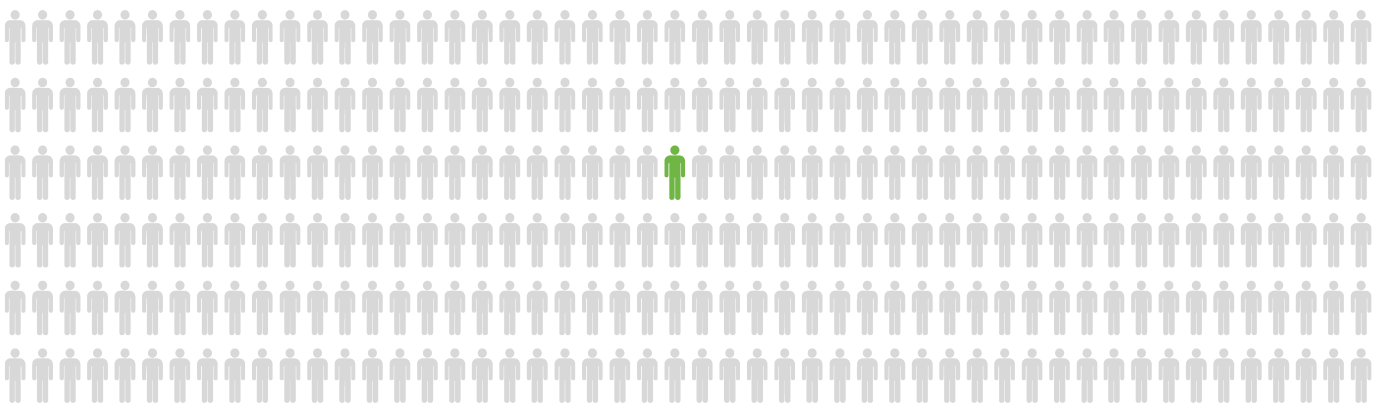
Over time 'bad' cholesterol can build-up in the arteries causing blockages. Blockages in the arteries of the heart (heart disease) cause heart attacks.

People with undiagnosed and untreated FH are at 20 times greater risk of having a heart attack.

20x

People with undiagnosed and untreated FH can have heart attacks and even die at a young age, as early as their 20s.

Early diagnosis and early treatment to lower the 'bad' cholesterol will stop its build-up in the arteries and help ensure a normal life expectancy.



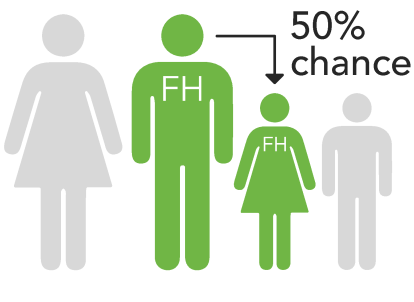
In Australia FH affects 1 in 300 people

90%

adults remain undiagnosed

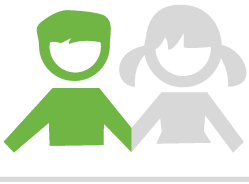
98%

children remain undiagnosed



FH runs in families.
All close family members (parents, siblings and children) of a person with FH should have their cholesterol tested.
They have a 50% (1 in 2) chance of also having FH.

Children with an FH parent should be tested around the age of 10.



FH will be suspected if a person has:

- high 'bad' cholesterol
- heart disease/attacks at a young age*
- a close family member with high 'bad' cholesterol or FH
- a close family member with heart disease/attacks at a young age*
- visible cholesterol deposits; 'lumps' in the hands, legs or eyes

* young age is men before the age of 55 and women before the age of 60

Treatment for FH includes:

- lifelong medication
- healthy diet
- physical activity
- healthy weight
- no alcohol or in moderation
- no smoking

fH Australasia Network

Early Diagnosis
Early Treatment
Saves Hearts

www

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