BMJ Open

Implementation of a primary-tertiary shared care model to improve the detection of familial hypercholesterolaemia (FH): a mixed methods pre-post implementation study protocol

Karen Birkenhead ,[1,2] David Sullivan,[3,4] Claire Trumble,[5] Catherine Spinks,[5] Shubha Srinivasan,[6,7] Andrew Partington,[8] Luke Elias,[9] Charlotte Mary Hespe ,[10] Gabrielle Fleming,[5] Stephen Li,[11] Madeline Calder,[5] Elizabeth Robertson,[4,12] Ronald Trent,[5,13] Mitchell N Sarkies [1,2]

To cite: Birkenhead K, Sullivan D, Trumble C, *et al*. Implementation of a primary-tertiary shared care model to improve the detection of familial hypercholesterolaemia (FH): a mixed methods pre-post implementation study protocol. *BMJ Open* 2024;**14**:e082699. doi:10.1136/bmjopen-2023-082699

► Prepublication history and additional supplemental material for this paper are available online. To view these files, please visit the journal online (https://doi.org/10.1136/bmjopen-2023-082699).

Received 30 November 2023
Accepted 12 April 2024

For numbered affiliations see end of article.

Correspondence to
Dr Karen Birkenhead; karen.birkenhead@sydney.edu.au

ABSTRACT

Introduction Familial hypercholesterolaemia (FH) is an autosomal dominant inherited disorder of lipid metabolism and a preventable cause of premature cardiovascular disease. Current detection rates for this highly treatable condition are low. Early detection and management of FH can significantly reduce cardiac morbidity and mortality. This study aims to implement a primary-tertiary shared care model to improve detection rates for FH. The primary objective is to evaluate the implementation of a shared care model and support package for genetic testing of FH. This protocol describes the design and methods used to evaluate the implementation of the shared care model and support package to improve the detection of FH.

Methods and analysis This mixed methods pre-post implementation study design will be used to evaluate increased detection rates for FH in the tertiary and primary care setting. The primary-tertiary shared care model will be implemented at NSW Health Pathology and Sydney Local Health District in NSW, Australia, over a 12-month period. Implementation of the shared care model will be evaluated using a modification of the implementation outcome taxonomy and will focus on the acceptability, evidence of delivery, appropriateness, feasibility, fidelity, implementation cost and timely initiation of the intervention. Quantitative pre-post and qualitative semistructured interview data will be collected. It is anticipated that data relating to at least 62 index patients will be collected over this period and a similar number obtained for the historical group for the quantitative data. We anticipate conducting approximately 20 interviews for the qualitative data.

Ethics and dissemination Ethical approval has been granted by the ethics review committee (Royal Prince Alfred Hospital Zone) of the Sydney Local Health District (Protocol ID: X23-0239). Findings will be disseminated through peer-reviewed publications, conference presentations and an end-of-study research report to stakeholders.

STRENGTHS AND LIMITATIONS OF THIS STUDY

⇒ The mixed methods design of our study will form a comprehensive picture of the shared care model benefits and the success of the implementation support package.
⇒ We will engage multiple stakeholders to codesign the process and ensure it meets the needs of those who will be impacted most.
⇒ By embedding implementation science into the study design, our protocol brings a systematic and evidence-based approach to improving the detection of familial hypercholesterolaemia.
⇒ Our observational study design introduces the risk of potential confounding variables influencing our outcomes of interest, which will be considered in any analysis and interpretation of findings.
⇒ The 12-month time frame may limit the sample size and exclude patients who decide to undergo cascade testing after the study period.

INTRODUCTION

Background

Familial hypercholesterolaemia (FH) is an autosomal dominant genetic condition that causes elevated low-density lipoprotein (LDL)-cholesterol from birth. The cumulative effects of lifelong exposure to elevated LDL-cholesterol can lead to premature atherosclerotic cardiovascular disease (ASCVD) and associated complications.[1 2] If left untreated, 50% of men and 60% of women will suffer premature ASCVD before 50 and 60 years, respectively.[3] Starting lipid-lowering therapy as early in life as possible can prevent cardiovascular disease and survival can be similar to persons without FH.[4] FH is a tier

1 genomic application which means it is a preventable cause of premature cardiovascular disease supported by evidence-based guidelines.[5] FH is a significant global health concern estimated to affect 1 in 311 people in the general population.[6] Unfortunately, diagnosis rates for FH in most countries are unknown, and most individuals with FH remain undetected and untreated with less than 10% of the estimated 35 million people worldwide with the condition being diagnosed.[2 7]

The detection and management of FH differs between countries which may be attributed to differences in healthcare system delivery.[8] In Australia, detection is primarily driven by opportunistic identification of index cases with some stand-alone programmes for systematic and cascade testing.[9 10] A diagnosis of FH can be made using phenotypic criteria and/or genetic testing, the former being largely driven by an elevated LDL-cholesterol level and confirmed using the Dutch Lipid Clinic Network criteria or other internationally used methods.[2 5] The proband, or first detected FH case in a family, is referred to as the 'index' case. Parents, siblings and children of an index case have a 50% chance of having the condition.[11] Genetic testing of first-degree and second-degree relatives of confirmed index cases is referred to as 'cascade' testing. Cascade testing is one of the most cost-effective approaches for detecting people with FH as it supports early detection and treatment, thereby reducing the lifelong burden of elevated LDL-cholesterol in both individuals and families.[12] However, despite the benefits of cascade testing, uptake remains low in several countries, including Australia, due to barriers associated with direct contact, complex family dynamics and limited practice infrastructure.[13 14]

Genetic testing allows for a more accurate diagnosis and can open pathways for people to access more suitable treatments, such as proprotein convertase subtilisin/kexin type 9 inhibitors, especially for those that have statin intolerance.[11] In 2020, genetic testing for FH was added to the Medicare Benefits Schedule (MBS), which is a list of services subsidised by the Australian government under Medicare, the country's universal health insurance scheme. However, as of August 2023 only 1886 rebates (1654 for diagnostic and 232 for cascade testing) had been made since the MBS Items were introduced. This equates to less than 2% of the estimated 100 000 individuals in Australia likely to have FH, with only a fraction coming from cascade testing. Explanations for the low utilisation of the MBS Items, particularly for cascade testing, may be due to a general lack of awareness or urgency about FH and the availability of the MBS Items, concerns related to potential exclusions from future insurance coverage of a confirmed genetic diagnosis, and accessibility/waiting times for appointment scheduling.[13] Increasing clinician awareness regarding available MBS-funded services and insurance coverage, and reducing wait times through greater general practice involvement in care may address these identified barriers. Increasing detection rates for FH, up to the level of expected cases forecasted when screening was first introduced to the system, would bring substantial individual, community and economic benefits, and have a significant impact on morbidity and death related to ASCVD.

Clinical practice guidelines outline the best available evidence for the delivery of healthcare and include not only what care should be delivered, but how best to deliver that care.[15] Although there are well-established guidelines for the care of FH, the ideal model of care for diagnosing and managing FH in Australia has not yet been established. FH is too common to rely on tertiary referral centres to manage all patients, especially for those with only routine management needs. Almost 90% of Australians visit their general practitioner (GP) at least once a year, making this a valuable setting to help diagnose and manage individuals with FH.[16] Most tests (90%) for LDL-cholesterol are ordered by GPs, further supporting the benefits of screening and diagnosing patients who may have FH.[10] Preliminary feedback from stakeholder interviews conducted by this research team suggests GPs could successfully be engaged in a shared care model, provided that sufficient and appropriate supports are in place.[13] GPs can play an important role in increasing the detection of FH with support from tertiary centres for central coordination to facilitate the process. Overall, the primary care setting provides an opportunity to identify and manage more patients with FH.

It is important to develop implementation strategies to effectively create and operationalise new models of care for the translation of FH clinical practice guidelines into practical application.[17] Furthermore, models of care should be adaptable to suit local needs and be acceptable to patient and provider.[5 18] Implementation science is the study of methods to improve the systematic update of evidence-based guidelines into routine practice and is an essential approach to implementing FH models of care.[8 19] There is a clear gap between evidence and practice in the identification and management of FH, including the underutilisation of genetic testing and family cascade testing.[17] Barriers include a lack of awareness and understanding of FH among many healthcare providers including cardiologists and GPs.[20] Implementation science can help bridge these gaps through well-designed research protocols that use strategies such as providing education and resources to support implementation.[18 21] Implementation science frameworks help to understand the underlying contextual factors that may impact the success of an intervention, thereby helping to identify appropriate strategies to address those barriers.[15] Our study will provide evidence to support the successful implementation of a model of care to improve the detection and management of FH into routine practice across primary and tertiary settings. This model of care could then be scaled up to support the timely

diagnosis, treatment and management of a range of genetic conditions.

Research question

How can existing tertiary health services support the implementation of a primary-tertiary shared care model to improve detection of FH?

Aim

Our aim is to implement a primary-tertiary shared care model to improve the detection of FH in New South Wales (NSW), Australia.

Objectives

The primary objective of this study is to evaluate the implementation of a shared care model and support package for genetic testing of FH. The secondary objective is to evaluate the patient experience of the shared care model.

METHODS

Study design

The approach to implement the primary-tertiary shared care model has been guided by the Exploration, Preparation, Implementation, Sustainment (EPIS) framework.[22] The EPIS framework guides projects through key stages of the implementation process and highlights important factors influencing implementation success with the broader 'outer context' (system) and proximal 'inner context' (organisation) across each EPIS stage. The exploration and preparation stages have been conducted by this research team and involved identifying barriers and enablers to implementation of the shared care model (ie, exploration), followed by tailoring the shared care model and implementation supports to local needs (ie, preparation). The third phase (ie, implementation) of the study will use a mixed methods pre-post implementation study design conducted over a 12-month period.[23] A combination of quantitative (ie, surveys and patient data) and qualitative (ie, semistructured interviews) outcome measures will be collected. Using this design, the impact of the primary-tertiary shared care model for FH will be compared against a historical usual care model for FH. The mix of quantitative and qualitative outcome measures will help form a comprehensive picture of the shared care model and the success of implementation supports tailored to match the contextual needs of healthcare providers and patients. Additionally, the pre-post study design offers an ideal approach to examining implementation under real-world conditions.

Study setting

The study will be conducted at NSW Health Pathology and Sydney Local Health District through the Vascular Health Clinic at Royal Prince Alfred Hospital (RPAH). The Vascular Health Clinic receives approximately 120 FH testing request referrals per year. NSW Health Pathology employs over 4000 staff and operates more than 60 laboratories, where it conducts more than 61 million tests per year from 200 pathology collection services in NSW public hospitals and community health facilities. Sydney Local Health District is located in the centre and inner west of Sydney. Around 16 000 staff are employed at Sydney Local Health District, responsible for the health and well-being of more than 700 000 people living locally, and in rural and remote parts of NSW and Australia. There are 577 general practices within the 587 km^2 area of the Central and Eastern Sydney region providing services to 1.5 million individuals living in the area.

Eligibility criteria

Healthcare providers

All internal and external healthcare providers associated with the Vascular Health Clinic at RPAH including referring GPs, nurses, physicians (eg, cardiologists, geneticists, pathologists, paediatricians) and genetic counsellors.

Patients

The patient sample will include confirmed index cases and their relatives referred to the Vascular Health Clinic at RPAH in Sydney, Australia. Detailed information on the patient sample can be found in figure 1.

Intervention

The intervention is described according to the Template for Intervention Description and Replication (TIDieR) guidelines[24] (online supplemental appendix 1). The shared care model begins following the diagnosis of a genetically confirmed index case within the Vascular Health Clinic (figure 2). A family letter is provided to the index case to give to their relatives or, if the index case prefers, and with their consent, Vascular Health Clinic staff will contact relative(s) on the index case's behalf (online supplemental appendix 2). The family letter has been translated into different languages to ensure FH cascade genetic testing is offered to all relatives of index cases. The Research Electronic Data Capture (REDCap) online database will be used to seek an expression of interest (EOI) from the relatives, where they can provide their contact details if they would like to learn more about cascade testing. The family letter includes a URL and a QR code with a link to REDCap and can be sent via email or provided in hard copy. When an EOI form is completed, staff at the Vascular Health Clinic will contact the relative by telephone or email within 1 week and provide more detailed information about cascade testing. If the relative cannot be reached by either telephone or email, clinic staff will make a second attempt to contact them requesting they reach out to the clinic at their convenience. Relatives are then emailed a cascade testing package that includes supporting paperwork to allow consent and ordering of FH cascade genetic testing with their GP. This package is also sent directly to the relatives preferred GP and includes a one-page cascade screening guide for the GP, a prefilled pathology request form, a genetic testing consent form, a list of educational resources and access to additional support through a

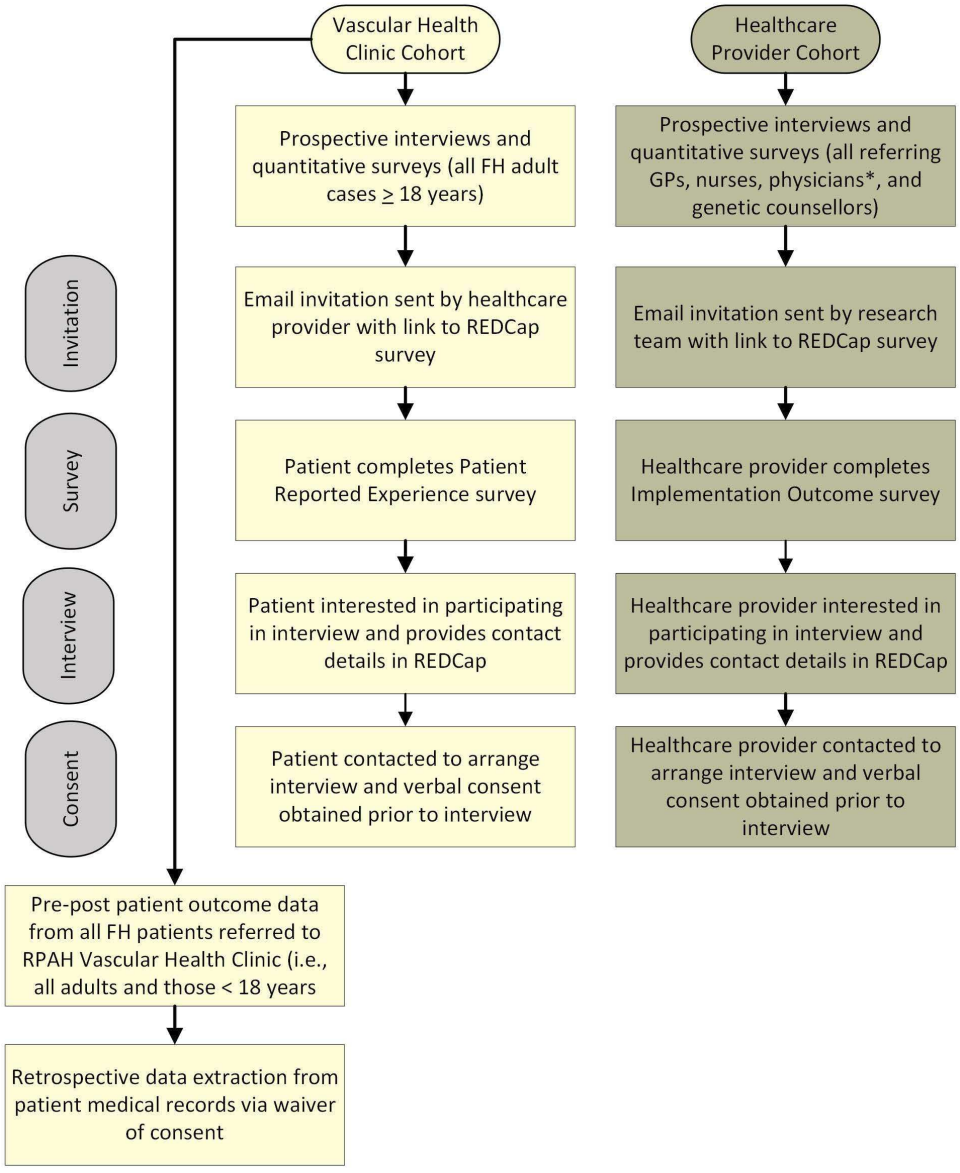


Figure 1 Study protocol flow diagram for evaluating the implementation of the primary-tertiary shared care model. *Cardiologists, geneticists, pathologists and paediatricians. FH, familial hypercholesterolaemia; GP, general practitioner; REDCap, Research Electronic Data Capture; RPAH, Royal Prince Alfred Hospital.

direct line for GPs to the Vascular Health Clinic (online supplemental appendix 3). Once genetic test results are received by the clinic a post results package is sent to the GP providing guidance on what to do with the results (online supplemental appendix 4). General practice and other specialist engagement with tertiary hospital clinics can be challenging, particularly when pathways for referral are not clear and where referring doctors have had previous negative experiences. To address this, our intervention will focus on promoting respectful, two-way communication of the benefits of early detection and preventive treatment. Additionally, the research team conducted preliminary work with key stakeholders to tailor the shared care model to local needs.[13] This includes providing further support through an online health information portal for GPs (ie, HealthPathways), a patient information booklet, a fact sheet on FH with the NSW Centre for Genetics Education and continuous professional development (CPD) opportunities for GPs through an online CPD Journal programme (ie, *Medicine Today*, a peer-reviewed journal of clinical practice).

Recruitment

Quantitative surveys

Healthcare providers will be recruited via an email sent to a purposive sample of key stakeholders associated with the Vascular Health Clinic at RPAH, containing a link to the online REDCap form where they will be asked to complete the implementation outcomes survey.[25] The recruitment of healthcare providers will take an arms-length approach whereby initial contact will be made by an investigator with legitimate access to potential participants' contact details but is not in an unequal workplace relationship with potential participants. The investigator

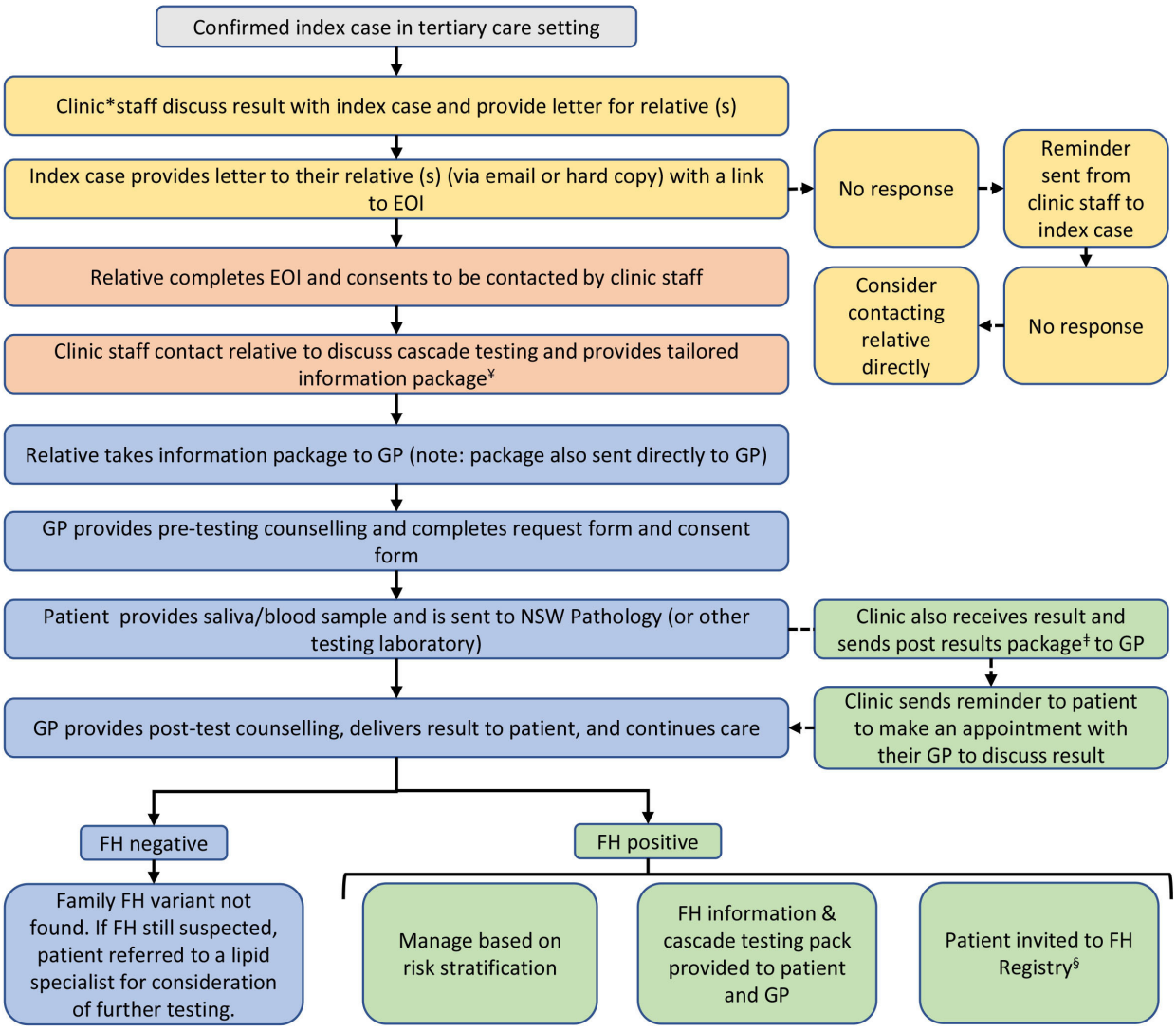


Figure 2 FH intervention for the primary-tertiary shared care model. *Refers to Royal Prince Alfred Hospital Vascular Health Clinic. ¥Includes one-page cascade screening guide for GP, prefilled pathology form, genetic testing consent form, patient FH fact sheet and FH-specific resources for primary care. ‡For positive results, the package includes results letter for GP, results letter for patient, family letter and FH registry consent form. §The National FH Registry is an electronic database where patients can provide consent for their medical information, family history and other related information to be collected for research purposes. EOI, expression of interest; FH, familial hypercholesterolaemia; GP, general practitioner; REDCap, Research Electronic Data Capture.

will approach the healthcare provider by email invitation to request their participation in the study, indicating that they have been identified as a key stakeholder involved in implementing the primary-tertiary shared care model.

Patients referred to the Vascular Health Clinic will be identified by the clinic staff and invited to participate in the study by email. The email will be sent by the patients' physician or a member of their treating team and will include a link to REDCap where they will be asked to complete the patient-reported experience measure (PREM).

Semistructured interviews

At the end of the survey, participants will be asked if they would like to participate in an interview to explore, in more detail, their experience and/or thoughts on the shared care model.

A flow diagram for the study protocol is presented in figure 1.

OUTCOMES

Primary outcomes

The evaluation of the shared care model implementation will be guided by a modified implementation outcome taxonomy.[26 27] Implementation outcomes are conceptually different from health service and clinical effectiveness outcomes. The implementation outcome taxonomy distinguishes between three interrelated types of outcomes: implementation/process,

health service and patient health. While the end goal is improvements in health service and patient outcomes, implementation/process outcomes precede these, with the latter outcomes being impacted by changes in implementation processes (ie, changes in clinical practice to deliver the intervention). A summary of primary outcome measures is presented in table 1.

Secondary outcomes

Patient experience will be captured using a PREM during the 12-month implementation period. PREMs are routine state-wide surveys used to gather patient feedback across all levels of the health system to improve patient care.

Outcomes will compare the impact of the new shared care model over a 12-month period against a historical usual care control condition.

Quantitative outcome data

Quantitative pre-post data will be collected from TrakGene, a clinical genetics database, and routine electronic medical record data collected at RPAH.[28 29] Quantitative outcome data will be collected through surveys (ie, using REDCap) sent to providers and patients. The surveys will be administered at a single time point and will take approximately 5 min to complete.

Qualitative outcome data

Semistructured interviews will be conducted both during the implementation and at the 12-month follow-up. Semistructured interviews will be conducted online or in person, depending on the participant's preference. The online semistructured interviews will be conducted via the Zoom or Microsoft Teams videoconference platforms. The interview guide will be informed by the EPIS framework[22] and literature on implementing genetic testing in primary care. The interview structure will be guided by the constructivist framework, allowing the conversation to be shaped by feedback from interviewees. The interview will be approximately 60 min in duration.

Implementation cost

A separate protocol is planned for a model-based health economic evaluation to represent the 'intervention' and 'usual care' comparator cohorts, and their resulting costs and effects.

Planned sample size

For the quantitative data, our statistical power estimation is based on the difference in the rate of cascade tests per index case before and after the adoption of our model. An estimated total sample size of 124 index cases will provide 80% power for a two-sided α=0.05, where there is a rate of 0.30 during the historical control and 0.60 after adoption of the shared care model (calculated using G*Power 3.1.9.7). The RPAH laboratory received 123 FH testing request referrals in 2021, which would provide a sufficient sample if similar numbers were received in 2022–2024.

For qualitative data pertaining to the implementation of the shared care model, we will use the concept of theoretical saturation to determine the interview sample size. From our experience, we anticipate conducting approximately 20 interviews throughout the 12-month study period.

DATA ANALYSIS

Quantitative data analysis

Quantitative pre-post data and implementation outcome surveys will be analysed using SAS V.9.4 (SAS Institute). Descriptive statistics will be used to summarise demographic data, such as mean and SD, median and IQR, and sample maximum and minimum for continuous data, and frequencies and proportions for categorical data. Statistical significance will be set at $p \leq 0.05$. Correlations between the acceptability, appropriateness and feasibility scales will be estimated using Pearson and Spearman rank-order correlation coefficients. Evidence of delivery data will be analysed using a Poisson regression model. Fidelity data will be analysed using logistic regression with potential confounders (eg, age, ethnicity, education level) included in the model. Goodness-of-fit diagnostics will be conducted, and if overdispersion of count data is identified then a negative binomial regression will be used. Timely initiation from referral to genetic testing and diagnosis will be assessed by linear regression.

Qualitative data analysis

Qualitative data will be coded and thematically analysed using QSR NVivo software according to a constructivist framework approach. This approach seeks to understand a social phenomenon and construct theories through participants' experiences, using iterative data collection and analysis.[30] EPIS constructs will be coded in relation to how they influence the implementation outcomes, such as how organisational staffing processes affect the feasibility of the model. Sample sizes provided are estimates, as data collection will continue until thematic saturation is achieved.

Patient and public involvement

Patient and public involvement in the design of this research study commenced before the research grant funding application stage. A range of key stakeholders with policy and practice expertise from NSW, Australia, were consulted to help codesign the shared care model. Essential partners involved in the codevelopment of the protocol included GPs, genetic counsellors, consumers, pathologists, paediatricians, geneticists and policymakers. Patients with lived experience of FH were involved in the design of this study from the initial planning stages and provided their input through focus groups, interviews and participation on the Project Steering Committee. The research question was informed through consultations with key stakeholders whose expertise and experience directly apply to the study protocol. A Project Advisory Group was established with representatives from local networks within the Sydney Local Health District to

Table 1 Summary of primary outcome measures

Outcome	Definition	Measure	Description
Acceptability	Acceptability is the degree to which the shared care model is agreeable, palatable or satisfactory to key healthcare professionals and patient stakeholders.	Measured using the Acceptability of Intervention Measure (AIM)[25] and qualitative interviews.	Four-item 5-point scale* and qualitative interview.
Appropriateness	Appropriateness is the perceived fit, relevance and compatibility of the model to solve the issue of FH detection and management.	Measured using the Intervention Appropriateness Measure (IAM)[25] and qualitative interviews.	Four-item 5-point scale* and qualitative interview.
Feasibility	Feasibility is considered the extent to which the new model is able to be successfully implemented.	Measured using the Feasibility of Intervention Measure (FIM)[25] and qualitative interviews.	Four-item 5-point scale* and qualitative interview.
Evidence of delivery	Use, provision or receipt of an intervention.	Measured by the rate of people genetically tested for FH and the rate of confirmed FH diagnoses.	Rate of genetic test results returned to NSW Pathology or other testing laboratory and the rate of confirmed gene changes indicating an FH diagnosis.
Fidelity	Degree to which the model is implemented as prescribed.	Measured by the adherence to key steps in the primary-tertiary model of care protocol.	Provider level ► Proportion of index cases provided with family letter. ► Proportion of index cases sent text message reminders when EOI is not received. ► Proportion of relatives who receive an attempted clinic staff contact to discuss cascade testing. ► Proportion of relatives and their GPs who are sent the cascade testing package. ► Proportion of relatives sent text message reminders when test results are not received by the Vascular Health Clinic. ► Proportion of GPs contacted once test results received. Patient level ► Proportion of index cases that provide the family letter to their relatives. ► Proportion of index cases that generated an EOI from a relative. ► Proportion of relatives genetically tested for FH. ► Proportion of relatives who followed up with their GP to receive test results.
Implementation cost	Implementation costs are those related to the execution of the model of care and implementation supports.	Measured directly from the service, and rates of uptake and adherence.	Implementation cost outcomes will be reported in a separate health economic modelling study.
Timely initiation	Extent to which a newly implemented treatment is initiated in a timely manner.	Measured by the mean time from referral to genetic testing and diagnosis.	Length of time between EOI response and post results package sent to GP.

*Responses include: completely disagree, disagree, neither agree nor disagree, agree and completely agree.
EOI, expression of interest; FH, familial hypercholesterolaemia; GP, general practitioner.

help design the project, support recruitment, maximise input while minimising participant burden and identify pathways for disseminating research findings.

Ethics and dissemination

This study was approved by the Ethics Review Committee (RPAH Zone) of the Sydney Local Health District (Protocol ID: X23-0239). Findings from this study will be disseminated through peer-reviewed publications, conference presentations and reports for key stakeholders and partners in the field.

DISCUSSION

This study will evaluate a primary-tertiary shared care model for increasing the detection and management of FH. Our study addresses an important gap in the care of people with FH by increasing detection of FH and enhancing access to genetic testing.[5] This study embeds implementation science into the design and methods as an important and necessary component to address and overcome barriers to improving the care of FH.[8] For an intervention to be successfully implemented and ongoing, it must be acceptable to the end user (ie, those delivering and/or receiving the intervention).[15] By interviewing patients and providers, we are addressing this important element by understanding, from the perspective of the end user, why the primary-tertiary shared care model may or may not be successful. Despite the strong evidence base behind FH care, efforts to translate guidelines into practice are often unsuccessful, likely related to a lack of relevant or appropriate implementation strategies.[17 31] The design of this study protocol has taken into consideration previously identified barriers and enablers by this research group allowing for the selection of appropriate implementation strategies tailored to local needs.[13] A fundamental element of all FH models of care is early detection and initiation of treatment to avoid the lifelong burden of elevated LDL-cholesterol, and engaging with GPs will support this process.[8] Usual shared care models typically involve GPs initiating collaboration with lipid specialists by referring individuals with suspected FH for diagnosis and treatment.[32 33] In contrast, our primary-tertiary shared care approach represents a departure from usual care whereby the GP is actively approached and supported as an integral component of the model. This unique perspective offers an innovative way of evaluating the approach, underscoring its potential success, particularly when overcoming the initial challenge of promoting genetic testing by GPs. This protocol also incorporates an important aspect of patient-centred care by empowering, and actively involving, index cases as care partners responsible for reaching out to family members for cascade testing. Additionally, it emphasises the importance of establishing linkages and collaboration with primary care providers, contributing to the achievement of integrated and partnered care during the early stages of case management. Successful implementation of our primary-tertiary shared care model will support early detection of FH as cascade testing extends across age groups, in particular children and young adults who have the most to gain from early detection.[34]

Findings from this study will be reviewed by the research team to identify core elements of the shared care model and implementation supports. Based on these core elements, recommendations will be made to support the long-term sustainment of the shared care model at the local level and broader scaling to other local health districts through the state-wide reach provided by NSW Health Pathology. A scale-up model will be formulated, which will include recommendations for model protocols and knowledge translation resources that could be used across NSW, Australia and internationally. The model has been designed to be incorporated into existing care workflows and make use of available funding reimbursement for genetic testing, enhancing the ability for scale-up to other tertiary hospitals in Australia.

There are some potential limitations to this study. Findings may not be generalisable to other settings due to differences in staff, patient populations, leadership and organisational culture. For example, fidelity could vary based on staff engagement levels with the new model of care. Measures for acceptability, appropriateness and feasibility may be subjective, influenced by providers' perceptions on how the model fits within their healthcare setting. Qualitative interviews will provide individualised feedback that could vary across settings and patient populations. However, steps will be taken to address these limitations to ensure core elements and implementation supports, and a well-planned implementation handover, are in place to support scale-up and implementation into policy and practice.

Results of this study have significant potential to improve patient and clinical outcomes by identifying and testing family members at risk. Our model will increase capacity building in general practice by supporting GPs to implement genetic medicine as part of standard practice. Long term, this model will lead to the prevention of early-onset coronary artery disease by improving the therapeutic management of FH. Additionally, this project will lead to improvements in patient outcomes through earlier and higher rates of FH detection in the community, facilitating commencement of lipid-lowering therapy, and addressing lifestyle factors to reduce the risk of coronary artery disease.[35 36] Our shared care model will enable patients to receive care closer to home, within their communities, and avoid potential long waiting lists. Our shared care model will also support the development of meaningful and equal partnerships between hospital clinics and general practice. As a tier 1 genomic application, FH has strong evidence supporting the use of genetic testing.[5] In Australia, cascade screening for FH and treatment with lipid-lowering therapy is a cost-effective means of preventing coronary heart disease in families at risk of FH,[37] which underpinned the MBS Items for genetic testing. By frequently engaging with policymakers

throughout the planning and development of this project we worked to ensure our model aligns with NSW government strategies and initiatives on genomic medicine.[38 39] Acknowledging the dynamic nature of healthcare systems, we recognise that the long-term success of our model hinges on continuous engagement with general practice to understand and address any issues that may impede long-term success. These actions will support and lead the way for introducing genomic medicine into general practice and other disciplines. We anticipate this primary-tertiary shared care model will demonstrate an exemplar approach for integrating genetic medicine into standard care and provide a roadmap for implementation across other genetic conditions.

Author affiliations
[1]School of Health Sciences, Faculty of Medicine and Health, The University of Sydney, Sydney, New South Wales, Australia
[2]Sydney Health Partners, Implementation Science Academy, Sydney, New South Wales, Australia
[3]Department of Chemical Pathology, NSW Health Pathology, Royal Prince Alfred Hospital, Sydney, New South Wales, Australia
[4]Central Clinical School, Faculty of Medicine and Health, The University of Sydney, Sydney, New South Wales, Australia
[5]Institute of Precision Medicine and Bioinformatics, Sydney Local Health District, Sydney, New South Wales, Australia
[6]Institute of Endocrinology and Diabetes, The Children's Hospital at Westmead, Westmead, New South Wales, Australia
[7]Discipline of Child and Adolescent Health, Faculty of Medicine and Health, The University of Sydney, Sydney, New South Wales, Australia
[8]Flinders Health and Medical Research Institute, Flinders University, Adelaide, South Australia, Australia
[9]FH Australasia Support Group, Sydney, New South Wales, Australia
[10]School of Medicine, The University of Notre Dame Australia, Sydney, New South Wales, Australia
[11]Core Pathology and Clinical Chemistry, NSW Health Pathology, Westmead Hospital, Sydney, New South Wales, Australia
[12]Department of Cardiology, Royal Prince Alfred Hospital, Sydney, New South Wales, Australia
[13]Department of Medical Genomics, Royal Prince Alfred Hospital, Sydney, New South Wales, Australia

X Karen Birkenhead @KLBirkenhead and Charlotte Mary Hespe @runningdoc14

Acknowledgements The authors acknowledge the contributions of Sophie Green and Marie Brigette Cusack who assisted with this project, as well as the project partners NSW Health Pathology, Sydney Local Health District, Central and Eastern Sydney Primary Health Network, NSW Agency for Clinical Innovation, NSW Ministry of Health and the FH Australasia Network and Australian Atherosclerosis Society. The authors also acknowledge research and implementation partners Dr Brendan Goodger, Dr Teresa Anderson, Bridie Carr, Kerrie Martin, Dr Olivia Hibbitt, Professor Gerald Watts, Dr Jing Pang and Dr Ross McLennan.

Contributors The study was conceived by MNS, CT, DS, CS, SS, AP, LE, CMH, GF, SL, MC, ER and RT and all authors contributed to the study design. KB and MNS wrote the initial draft of the manuscript. All authors contributed to and approved the final version of the manuscript.

Funding This work was supported by an NSW Health Translational Research Grant Scheme (H22/4011) and a Heart Foundation Vanguard Grant (105721).

Disclaimer The funding arrangement ensured the funders have not and will not have any role in study design, collection, management, analysis and interpretation of data, drafting of manuscripts and decision to submit for publication.

Competing interests DS has received grants from Regeneron, Amgen, Arrowhead, Ionis and Novartis via Sydney Local Health District; consulting fees and speaker fees from Amgen and Novartis; and drug samples for a treatment adherence programme from Sanofi. CMH has received Amgen, Sanofi and MRFF grants looking at FH identification in the general practice setting, and assisted in the Amgen-funded independent education design for the GP. MNS is supported by an NHMRC Investigator Grant and received honoraria related to consulting, research and/or speaker activities from Amgen.

Patient and public involvement Patients and/or the public were involved in the design, or conduct, or reporting, or dissemination plans of this research. Refer to the Methods section for further details.

Patient consent for publication Not applicable.

Provenance and peer review Not commissioned; externally peer reviewed.

ORCID iDs
Karen Birkenhead http://orcid.org/0000-0002-8311-7794
Charlotte Mary Hespe http://orcid.org/0000-0002-4582-7728
Mitchell N Sarkies http://orcid.org/0000-0001-7318-3598

REFERENCES

1 Pang J, Sullivan DR, Brett T, *et al*. Familial Hypercholesterolaemia in 2020: a leading tier 1 Genomic application. *Heart Lung Circ* 2020;29:619–33.
2 Watts GF, Gidding SS, Mata P, *et al*. Familial Hypercholesterolaemia: evolving knowledge for designing adaptive models of care. *Nat Rev Cardiol* 2020;17:360–77.
3 Nordestgaard BG, Chapman MJ, Humphries SE, *et al*. Familial Hypercholesterolaemia is Underdiagnosed and Undertreated in the general population: guidance for Clinicians to prevent coronary heart disease: consensus statement of the European Atherosclerosis society. *Eur Heart J* 2013;34:3478–90a.
4 Stone NJ, Robinson JG, Lichtenstein AH, *et al*. ACC/AHA guideline on the treatment of blood cholesterol to reduce Atherosclerotic cardiovascular risk in adults. *Circulation* 2014;129:S1–45.
5 Watts GF, Sullivan DR, Hare DL, *et al*. Integrated guidance for enhancing the care of familial Hypercholesterolaemia in Australia. *Heart Lung Circ* 2021;30:324–49.
6 Hu P, Dharmayat KI, Stevens CAT, *et al*. Prevalence of familial hypercholesterolemia among the general population and patients with Atherosclerotic cardiovascular disease: a systematic review and meta-analysis. *Circulation* 2020;141:1742–59.
7 Beheshti SO, Madsen CM, Varbo A, *et al*. Worldwide prevalence of familial hypercholesterolemia: meta-analyses of 11 million subjects. *J Am Coll Cardiol* 2020;75:2553–66.
8 Watts GF, Gidding SS, Hegele RA, *et al*. International Atherosclerosis society guidance for implementing best practice in the care of familial Hypercholesterolaemia. *Nat Rev Cardiol* 2023;20:845–69.
9 Bell DA, Pang J, Burrows S, *et al*. Effectiveness of genetic Cascade screening for familial Hypercholesterolaemia using a centrally Co-Ordinated clinical service: an Australian experience. *Atherosclerosis* 2015;239:93–100.
10 Marquina C, Lacaze P, Tiller J, *et al*. Population Genomic screening of young adults for familial Hypercholesterolaemia: a cost-effectiveness analysis. *Eur Heart J* 2022;43:3243–54.
11 Watts GF, Sullivan DR, Hare DL, *et al*. Essentials of a new clinical practice guidance on familial Hypercholesterolaemia for physicians. *Intern Med J* 2021;51:769–79.
12 Ademi Z, Norman R, Pang J, *et al*. Health economic evaluation of screening and treating children with familial hypercholesterolemia early in life: many happy returns on investment. *Atherosclerosis* 2020;304:1–8.
13 Sarkies MN, Testa L, Best S, *et al*. Barriers to and Facilitators of implementing guidelines for detecting familial Hypercholesterolaemia in Australia. *Heart Lung Circ* 2023;32:1347–53.

14 Kullo IJ. Familial hypercholesterolemia. *Circulation* 2020;142:1999–2001.
15 Sarkies MN, Francis-Auton E, Long JC, *et al*. Making implementation science more real. *BMC Med Res Methodol* 2022;22:178.
16 The Royal Australian College of General Practitioners. General practice: health of the nation. 2022. Available: www.racgp.org.au/general-practice-health-of-the-nation-2022
17 Sarkies M, Jones LK, Pang J, *et al*. How can implementation science improve the care of familial Hypercholesterolaemia *Curr Atheroscler Rep* 2023;25:133–43.
18 Sarkies MN, Jones LK, Gidding SS, *et al*. Improving clinical practice guidelines with implementation science. *Nat Rev Cardiol* 2022;19:3–4.
19 Eccles MP, Mittman BS. Welcome to implementation science. *Implementation Sci* 2006;1:1.
20 Jones LK, Williams MS, Ladd IG, *et al*. Collaborative approach to reach everyone with familial hypercholesterolemia: CARE-FH protocol. *J Pers Med* 2022;12:606.
21 Sarkies MN, White J, Morris ME, *et al*. Implementation of evidence-based weekend service recommendations for Allied health managers: a cluster randomised controlled trial protocol. *Implement Sci* 2018;13:60.
22 Moullin JC, Dickson KS, Stadnick NA, *et al*. Systematic review of the exploration, preparation, implementation, Sustainment (EPIS) framework. *Implement Sci* 2019;14:1.
23 Curran GM, Bauer M, Mittman B, *et al*. Effectiveness-implementation hybrid designs: combining elements of clinical effectiveness and implementation research to enhance public health impact. *Med Care* 2012;50:217–26.
24 Hoffmann TC, Glasziou PP, Boutron I, *et al*. Better reporting of interventions: template for intervention description and replication (Tidier) checklist and guide. *BMJ* 2014;348:bmj.g1687.
25 Weiner BJ, Lewis CC, Stanick C, *et al*. Psychometric assessment of three newly developed implementation outcome measures. *Implementation Sci* 2017;12:108.
26 Proctor E, Silmere H, Raghavan R, *et al*. Outcomes for implementation research: conceptual distinctions, measurement challenges, and research agenda. *Adm Policy Ment Health* 2011;38:65–76.
27 Proctor EK, Bunger AC, Lengnick-Hall R, *et al*. Ten years of implementation outcomes research: a Scoping review. *Implement Sci* 2023;18:31.
28 Sarkies MN, Bowles K-A, Skinner EH, *et al*. Data collection methods in health services research. *Appl Clin Inform* 2015;06:96–109.
29 Sarkies MN, Bowles K-A, Skinner EH, *et al*. Do daily ward interviews improve measurement of hospital quality and safety indicators? A prospective observational study. *J Eval Clin Pract* 2016;22:792–8.
30 Thomas A, Menon A, Boruff J, *et al*. Applications of social Constructivist learning theories in knowledge translation for Healthcare professionals: a Scoping review. *Implement Sci* 2014;9:54.
31 Powell BJ, Beidas RS, Lewis CC, *et al*. Methods to improve the selection and Tailoring of implementation strategies. *J Behav Health Serv Res* 2017;44:177–94.
32 Qureshi N, Da Silva MLR, Abdul-Hamid H, *et al*. Strategies for screening for familial Hypercholesterolaemia in primary care and other community settings. *Cochrane Database Syst Rev* 2021;10:CD012985.
33 Leonardi-Bee J, Boateng C, Faria R, *et al*. Effectiveness of Cascade testing strategies in relatives for familial hypercholesterolemia: A systematic review and meta-analysis. *Atherosclerosis* 2021;338:7–14.
34 Wiegman A, Gidding SS, Watts GF, *et al*. Familial Hypercholesterolaemia in children and adolescents: gaining decades of life by optimizing detection and treatment. *Eur Heart J* 2015;36:2425–37.
35 Tada H, Kawashiri MA, Nohara A, *et al*. Impact of clinical signs and genetic diagnosis of familial Hypercholesterolaemia on the prevalence of coronary artery disease in patients with severe Hypercholesterolaemia. *Eur Heart J* 2017;38:1573–9.
36 Koff E, Lyons N. Implementing value-based health care at scale: the NSW experience. *Med J Aust* 2020;212:104–6.
37 Ademi Z, Watts GF, Pang J, *et al*. Cascade screening based on genetic testing is cost-effective: evidence for the implementation of models of care for familial hypercholesterolemia. *J Clin Lipidol* 2014;8:390–400.
38 NSW Ministry of Health. NSW Genomic Strategy. Sydney: NSW Government, 2017.
39 NSW Ministry of Health. NSW Health Genomics Strategy Implementation Plan 2021–25. Sydney: NSW Government, 2021.

Appendix 1. TIDieR Checklist

Intervention described according to the TIDieR guidelines

Item		Control Group	Implementation Strategy Group
1	**BRIEF NAME:** Provide the name or a phase that best describes the intervention	Historical usual care model group	Primary-tertiary shared care model group
2	**WHY:** Describe any rationale, theory, or goal of the elements essential to the intervention.	The usual care model involves previous patients referred to the Vascular Health Clinic by general practitioners (GPs), cardiologists, endocrinologists, or other medical professionals with suspected or clinically diagnosed FH. This will serve as a pragmatic reference standard for the implementation research.	The primary-tertiary shared care model involves patients referred to the Vascular Health Clinic by GPs, cardiologists, endocrinologists, or other medical professionals with suspected or clinically diagnosed FH and their relatives who are transitioned through the new model of care. The co designed shared care model will lead to increased genetic testing for FH by providing appropriate support, guidance and clear communication between general practice and tertiary hospital clinics.
3	**WHAT (**Materials): Describe any physical or informational materials used in the intervention, including those provided to participants or used in intervention delivery or in training of intervention providers. Provide information on where the materials can be accessed (e.g., online appendix, URL).	A confirmed index case in the usual care control group was offered a family letter to pass onto their relatives that suggested they contact genetic services at the clinic, or seek a referral to a local genetics service, if interested in learning more about cascade testing. The family letter included contact information for the Vascular Health Clinic.	A confirmed index case in the primary tertiary shared care model is offered a revised family letter to give to their relatives. The revised family letter includes contact information for the Vascular Health Clinic and a link to an online expression of interest form to find out more about cascade testing. In addition, the shared care model includes a cascade testing package which is provided to relatives and the relatives nominated GP. The cascade testing package includes the following: 1) a one-page cascade screening guide for the GP, 2) a prefilled pathology form, 3) a genetic testing consent form, 4) a patient FH factsheet, 5) links to GP resources and contact details for the Vascular Health Clinic for additional support. The revised family letter and cascade testing package can be found in the online supplemental Appendices 2 and 3. The shared care model also includes a post results package which is sent to the GP after results are returned. For a positive result, the package includes the following: 1) results letter for GP, 2) results letter for patient, 3) family letter, and 4) a FH registry consent form. A positive post result package can be found in the online supplemental material Appendix 4. GPs may contact the Vascular Health Clinic throughout the process for assistance via telephone or email.

Appendix 1. TIDieR Checklist

Intervention described according to the TIDieR guidelines *(continued)*

Item		Control Group	Implementation Strategy Group
4	**WHAT** (Procedures): Describe each of the procedures, activities, and/or processes used in the intervention, including any enabling or support activities.	In the usual care group, patients with suspected or clinically diagnosed FH (i.e., index case) are provided a clinic appointment with a lipid specialist and genetic counsellor to discuss FH and genetic testing. Pre-test genetic counselling is provided to those who consent to genetic testing, which includes a discussion about the possible results and potential benefits of testing. On an individual patient basis (e.g., patients interested in genetic testing) the genetic counsellor spoke about risk to family members, discussed cascade testing, and provided a family letter, when relevant. Once results were returned to the clinic, an appointment was arranged with the genetic counsellor in person or by telephone, depending in the patient's preference, for post-test genetic counselling. Post-test counselling includes reminding patients to notify their at-risk relatives if a positive genetic variant was found. This may also include offering a family letter that encourages relatives to arrange an appointment with the genetic counsellor at the clinic, or with their local genetic services. If the relative did seek a referral to the Vascular health Clinic they would be put on a waitlist. They would then have an appointment with the genetic counsellor to discuss cascade testing. Following testing a letter would be written back to their referring doctor with the result and advice for patient and family members.	In the primary tertiary care group, patients with suspected or clinically diagnosed FH (i.e., index case) are provided a clinic appointment with a lipid specialist and genetic counsellor to discuss FH and genetic testing. Pre-test genetic counselling is provided to those who consent to genetic testing, which includes a discussion about the possible results and potential benefits of testing. The genetic counsellor will talk about risk to family members, discuss cascade testing and provide the revised family letter during the pre-test counselling appointment on an individual patient basis. Once results are returned to the clinic an appointment is arranged with the genetic counsellor, in person or by telephone, who provides post-test genetic counselling. This includes providing the revised family letter if they were not provided at the pre-test counselling or reminding patients to notify their at-risk relative (s) if a positive genetic variant was found. The relative completes an expression of interest form and is then contacted by the Vascular Health Clinic to discuss cascade testing. They are emailed, or mailed, the cascade testing package, which is also sent to the relatives nominated GP. The package includes all information needed by the GP to conduct cascade testing of first degree relatives. The relative undergoes genetic testing through their GP by providing either a saliva or blood sample. The sample is returned to hospital pathology. Results of the test are sent from NSW Pathology, or other testing laboratory, to the Vascular Health Clinic and the relative's GP. The GP is then sent the post results package. GPs may contact the Vascular Health Clinic throughout the process for assistance via telephone or email.
5	**WHO PROVIDED:** For each category of intervention provider (e.g., psychologist, nursing assistant), describe their expertise, background and any specific training given.	The usual care model primarily includes clinic staff comprised of a lipid specialist, genetic counsellors, clinic nurse, and other healthcare providers, such as general practitioners, on an ad hoc basis.	The primary-tertiary shared care model includes staff in tertiary and primary care comprised of a lipid specialist, genetic counsellor, clinic nurse, pathologists, and general practitioners guided by a well-defined shared care pathway based on equal partnership between primary and tertiary care.

Appendix 1. TIDieR Checklist

Intervention described according to the TIDieR guidelines *(continued)*

Item		Control Group	Implementation Strategy Group
6	**HOW:** Describe the modes of delivery (e.g., face-to-face or by some other mechanism, such as internet or telephone) of the intervention and whether it was provided individually or in a group.	In the usual care model, patients seeking index testing or cascade testing would have a face-to-face clinic appointment, or have a telephone consultation to arrange testing, depending on the patient's preference (and whether they needed to come in for an appointment to see a specialist doctor). Depending on the patient's preference, and situation, the results would be discussed by phone or in clinic.	The primary-tertiary shared care model includes an initial individual face-to-face clinic appointment with the index case, a telephone appointment with relatives, and individual face-to-face visits in general practice between relatives and their GP. Follow up appointments for relatives to discuss results face-to-face with their GP, and further support is available via telephone and internet (i.e., email, internet resources) from the clinic.
7	**WHERE:** Describe the type(s) of location(s) where the intervention occurred, including any necessary infrastructure or relevant features.	The usual care model is conducted in a tertiary care setting (i.e., Vascular Health Clinic).	The intervention is conducted in the primary and tertiary care settings through general practice and the Vascular Health Clinic, respectively.
8	**WHEN and HOW MUCH:** Describe the number of times the intervention was delivered and over what period of time including the number of sessions, their schedule, and their duration, intensity or dose.	Between 2022 - 2023 of the usual care model at the Vascular Health Clinic, diagnosing one index patient took approximately three to four months (from genetic testing to return of results). If a relative was then interested in a referral for cascade testing, they were placed on an average 8 month wait list for the Vascular Health Clinic. The process then took approximately 8 to 12 weeks (from cascade genetic test to return of results). Following testing, a letter would be written back to their referring doctor with the result and advice for patient and family members.	An individual patient through the primary-tertiary shared care model begins with diagnosing one index patient which takes approximately 3 to 4 months (from genetic testing to return of results). This is followed by cascade testing for at risk relatives with the process depending on GP appointment availability (anticipated less than the vascular health clinic wait time of 8 months) which include two reminders to index case if an EOI is not received by their relative. Blood or saliva sample provided for genetic testing (approximately 8 to 12 weeks) and follow up with GP and the Vascular Health Clinic, as needed, to discuss results.
9	**TAILORING:** If the intervention was planned to be personalised, titrated or adapted, then describe what, why, when, and how.	The usual care model provides index cases with a family letter, and additional resources on FH which are provided during the initial clinic visit and can be accessed online https://www.athero.org.au/fh/patients/cholesterol-and-cardiovascular-disease/. Patients and their relatives are also able to contact the clinic as needed.	In addition to the revised family letter and additional resources on FH provided as per usual care, the primary-tertiary shared care model will be adapted and refined over the 12-month period based on initial feedback. The model with be tailored and may include additional information in the cascade testing package and further supports such as: indirect vs. direct contact of relatives; online health information portal for GPs (i.e., HealthPathways); a patient information booklet; a factsheet on FH with the NSW Centre for Genetics Education; continuous professional development (CPD) opportunities for GPs; a direct GP contact to the Vascular Health Clinic; utilising NSW Health Pathology or private pathology providers; genetic vs. phenotypic testing.

Appendix 1. TIDierR Checklist

Intervention described according to the TIDieR guidelines *(continued)*

Item		Control Group	Implementation Strategy Group
10	**MODIFICATIONS:** If the intervention was modified during the course of the study, describe the changes (what, why, when, and how).	Not applicable for protocol.	Not applicable for protocol.
11	**HOW WELL** (Planned): If intervention adherence or fidelity was assessed, describe how and by whom, and if any strategies were used to maintain or improve fidelity, describe them.	Adherence or fidelity will not be assessed in the usual care control group as an implementation strategy was not in place during the previous 12-month usual care period.	Whether or not the model is implemented as prescribed as measured by the number of people detected with FH will be explored in the 12-month period of the primary-tertiary shared care model.
12	**HOW WELL** (Actual): If intervention adherence or fidelity was assessed, describe the extent to which the intervention was delivered as planned.	Not applicable for protocol.	Not applicable for protocol.

Appendix 2. Family letter provided to index cases to pass onto relatives

Familial Hypercholesterolaemia Clinical Support Service
Royal Prince Alfred Hospital

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

Genetic File:

___/___/___

Dear family member,

A relative of yours has been diagnosed with a genetic condition called familial hypercholesterolaemia (FH). This condition causes high cholesterol and increases the risk of getting heart disease at a young age.

FH runs in families so there is a risk that you also have FH. Fortunately, therapy is safe, easy, and effective at lowering the risk of heart disease.

Your local GP can help to check if you have FH. This can be done through:
1) LDL-cholesterol check (blood sample), or
2) Genetic testing (blood or saliva sample)

We urge **all relatives** of a person with FH to have an LDL-cholesterol check. Genetic testing for FH is voluntary and Medicare rebated for at-risk relatives. Your GP can arrange testing for the specific FH genetic variant in the _____ **gene,** identified in your family. They will need the reference **laboratory ID number MD -**_____.

Please provide your contact details by scanning the QR code below. Someone from the RPA Hospital Vascular Health Clinic will be in touch to provide information to you and your GP to help with arranging this testing:

QR
CODE

Or, click on the link: http://redcap.link

You are also welcome to talk with one of our genetic counsellors on (xx) xxx xxxx or email [clinic email]

Yours sincerely,
The FH Clinical Support Service

Useful information on FH:
- Australian Atherosclerosis Society what is FH - https://www.athero.org.au/fh/patients/what-is-fh/
- World Heart Federation what is FH video - https://youtu.be/4YwdFSN3xpA

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

Appendix 3 Cascade Testing Package

Familial Hypercholesterolaemia Clinical Support Service
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.

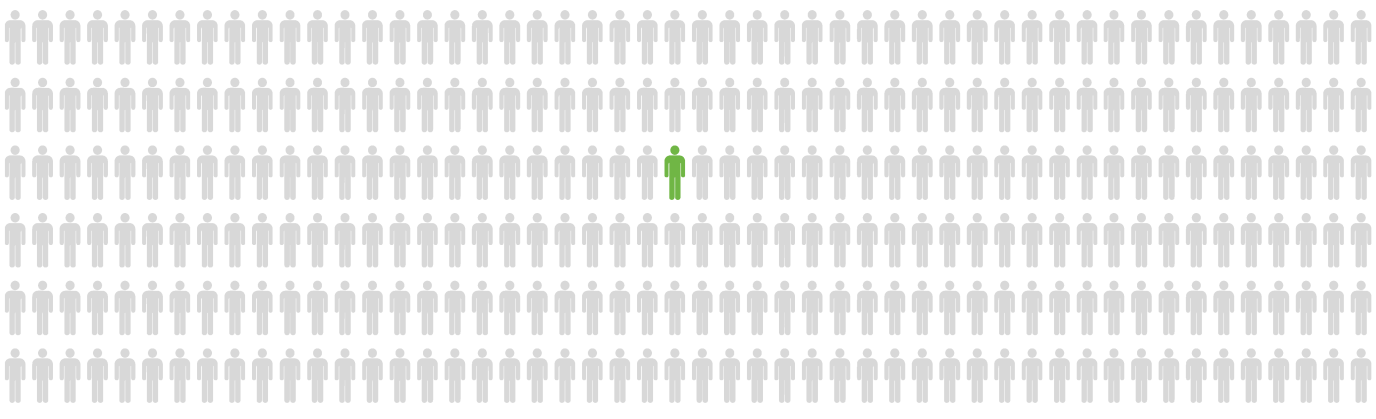
Over time 'bad' cholesterol can build-up in the arteries causing blockages. Blockages in the arteries of the heart (heart disease) cause heart attacks.

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.



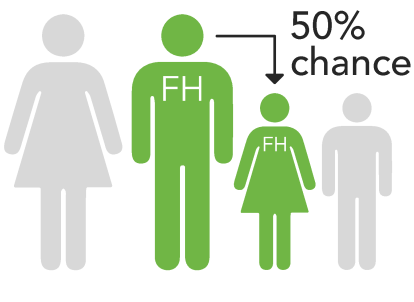
In Australia FH affects 1 in 300 people

90%

adults remain undiagnosed

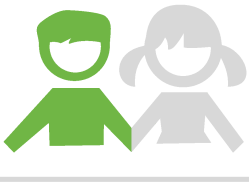
98%

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

fH Australasia Network

Appendix 4. Positive Results Package

fH Australasia Network
everyone deserves a better chance

Familial Hypercholesterolaemia Clinical Support Service
Royal Prince Alfred Hospital

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

Dear Dr [Name],

RE: [patient name] DOB:

As you are aware, results of the familial hypercholesterolaemia (FH) genetic testing that you ordered for [patient name] recently became available.

Their result shows that they **have inherited the gene change in their family.** This result confirms that they have the condition FH, so it is important they monitor and manage their cholesterol levels with your help.

We have enclosed a pre-written letter that can be given to your patient regarding the outcome of their genetic testing. We encourage you to also provide them with a copy of their genetic test report.

Management guidelines for individuals with FH are available at HealthPathways:
https://sydney.communityhealthpathways.org/

Additional information can be found in the FH Cascade Screening information (enclosed).

We have also enclosed a letter for your patient to pass onto any of their relatives who may now need genetic testing for the FH gene change found in their family. Please note that first-degree relatives (parents, siblings, children) have a 50% of also having FH and second-degree relatives (grandparents, grandchildren, half siblings, aunt/uncle, niece/nephew) have a 25% chance of also having FH.

National FH Registry
Your patient is invited to contribute their health information to a national database of patients receiving medical treatment for FH throughout Australia. Further participant information for the patient is enclosed. If your patient is interested in participating in this Registry they will only need to complete the consent form and return it to [clinic email address]

If you or your patient have any additional questions, you are welcome to contact one of our genetic counsellors on [phone number] or email [address].

Yours sincerely,

The FH Clinical Support Service

Enc: Patient FH Results Letter
Family Letter
FH Registry Patient Information and Consent Form

Appendix 4. Positive Results Package

everyone deserves a better chance

Familial Hypercholesterolaemia Clinical Support Service
Royal Prince Alfred Hospital

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

Dear [Patient],

You underwent genetic testing for a gene change identified in your family that causes Familial Hypercholesterolaemia (FH). **Your result showed that you have inherited this gene change (XXXX).** This result confirms that you have the condition FH. **Please ensure you arrange an appointment with your doctor to discuss these results and your ongoing management.**

Additional information about FH can be found at the FH Australasia Network:
https://www.athero.org.au/fh/patients/what-is-fh/

We have enclosed a letter for you to pass onto any relatives who may now need genetic testing for the FH gene change found in your family. Please note that first-degree relatives (parents, siblings, children) have a 50% of also having FH and second-degree relatives (grandparents, grandchildren, half siblings, aunt/uncle, niece/nephew) have a 25% chance of also having FH.

National FH Registry
You are also invited to contribute your health information to a national database of patients receiving medical treatment for Familial Hypercholesterolemia (FH) throughout Australia. Further participant information is enclosed. If you are interested in participating in this registry you will need to complete the consent form and return it to [clinic email address]

If you have any additional questions, you are welcome to contact one of our genetic counsellors on [phone number] or email [address]

Yours sincerely,

The FH Clinical Support Service

Enc: FH Registry Patient Information and Consent Form
Family information letter
Genetic Test Result

Familial Hypercholesterolaemia Clinical Support Service, Royal Prince Alfred Hospital
P 02 9515 5062 **F** 02 9515 5490 **E** SLHD-FH-Genetics@health.nsw.gov.au

Appendix 4. Positive Results Package

Familial Hypercholesterolaemia Clinical Support Service
Royal Prince Alfred Hospital

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

Genetic File:

___/___/___

Dear family member,

A relative of yours has been diagnosed with a genetic condition called familial hypercholesterolaemia (FH). This condition causes high cholesterol and increases the risk of getting heart disease at a young age.

FH runs in families so there is a risk that you also have FH. Fortunately, therapy is safe, easy, and effective at lowering the risk of heart disease.

Your local GP can help to check if you have FH. This can be done through:
1) LDL-cholesterol check (blood sample), or
2) Genetic testing (blood or saliva sample)

We urge **all relatives** of a person with FH to have an LDL-cholesterol check. Genetic testing for FH is voluntary and Medicare rebated for at-risk relatives. Your GP can arrange testing for the specific FH genetic variant in the _____ **gene,** identified in your family. They will need the reference **laboratory ID number MD -**_____.

Please provide your contact details by scanning the QR code below. Someone from the RPA Hospital Vascular Health Clinic will be in touch to provide information to you and your GP to help with arranging this testing:

QR CODE

Or, click on the link: http://redcap.link

You are also welcome to talk with one of our genetic counsellors on (xx) xxx xxxx or email [clinic email]

Yours sincerely,
The FH Clinical Support Service

Useful information on FH:
- Australian Atherosclerosis Society what is FH - https://www.athero.org.au/fh/patients/what-is-fh/
- World Heart Federation what is FH video - https://youtu.be/4YwdFSN3xpA

Appendix 4. Positive Results Package

National Familial Hypercholesterolemia Registry

INFORMATION FOR PATIENTS

Introduction

You are invited to contribute your health information to a national database of patients receiving medical treatment for Familial Hypercholesterolemia (FH) throughout Australia. It is hoped that research on this database will help researchers to better understand the nature of Familial Hypercholesterolemia and assist in developing future clinical research. Over 1500 people are currently enrolled in the FH Registry.

The National FH Registry is overseen and supported financially by the FH Australasia Network (Australian Atherosclerosis Society).

Contributing to the Registry

If you agree to participate in this Registry, you will not be required to do anything other than sign the Patient Consent Form. Relevant information will then be obtained from your medical record and stored in the database. The Registry will record demographic information including name, address, date of birth, email and contact telephone numbers as well as treating doctor's name, address, telephone number and email. Clinical information such as family history of FH, history of cardiovascular disease and cardiovascular disease risk factors, treatment and lipid concentrations will be provided by your treating doctor and recorded. If genetic testing has been done, the affiliated laboratory services will be asked to provide details of the results. This information will be entered by your clinic co-ordinator or treating doctor when you are registered onto the Registry. Your information will be kept in the Registry for 60 years.

In the Registry your health information will be identified with a number to protect your privacy. Your name will be recorded in connection with this number, but information about you will only be linked to your number. The information will always be treated confidentially, and only the database custodian, its staff and authorised researchers will have access to it. All confidential information will be encrypted and stored securely in accordance with the Privacy Act 1988 (Cth) and the Australian Privacy Principles. We will use Amazon Web Services (AWS) cloud infrastructure located in Australia and all patient data collected in the Registry will remain in Australia.

The results of research conducted using the registry may be presented at a conference or in a scientific publication, but individual patients will not be identifiable in such a presentation. Research data may be shared locally and internationally with other research collaborators in future, however ethics approval will be sought prior to sharing.

Benefits

While we intend this database to be used to further medical knowledge and to improve treatment of FH in the future, it may not be of direct benefit to you.

Appendix 4. Positive Results Package

Risks
There is minimal risk in taking part in the Registry. The Registry includes questions that may be considered sensitive and some participants may feel uncomfortable answering. You do not have to share any information you do not want to. Another unlikely risk is potential breaches in the computer system. In the event that there is a breach in the Registry's computer system all participants will be notified.

Costs
Contributing to this Registry will not cost you anything, nor will you be paid.

Voluntary Participation
Contributing to this database is entirely voluntary. You do not have to do so. If you do, you can withdraw your health information at any time without having to give a reason by contacting your treating doctor, or the local Registry co-ordinator at [email], or the National Co-ordinator at fh@athero.org.au. Whatever your decision, please be assured that it will not affect your medical treatment or your relationship with the staff who are caring for you.

Further Information
When you have read this information, the FH Clinical Support Service at {insert hospital name} will discuss it with you further and answer any questions you may have. If you would like to know more at any stage, please feel free to contact the genetic counsellor on {insert phone number}.

For more information, please refer to the Terms of Reference of the FH Registry Charter provided by your treating doctor and/or please refer to the Australian Familial Hypercholesterolemia Registry website (https://fhregistry-international.com/).

This information sheet is for you to keep.

Ethics Approval and Complaints
The establishment of this Registry has been approved by the Ethics Review Committee (RPAH Zone) of the Sydney Local Health District. Any person with concerns or complaints about it should contact the Executive Officer on 02 9515 6766 and quote protocol number X14-0135.

Appendix 4. Positive Results Package

National Familial Hypercholesterolemia Registry

PATIENT CONSENT FORM

I, ... [name]

of

.....…….[address]

have read and understood the Information for Patients on the above named Registry

and have discussed it with …….....

I have been made aware of the procedures involved.

I understand that contributing to the Registry will allow the researchers to have access to my medical record, and I agree to this.

I consent to the future use of any data I provide for research purposes. I understand that before they can use any data I provide, they must seek additional ethics approval.

I consent for other research collaborators to use any data I provide for future research purposes. I understand that before they can use my data, they must seek additional ethics approval.

I freely choose to contribute to the Registry and understand that I can withdraw my health information at any time.

I also understand that the Registry is strictly confidential.

- I would like to be contacted about clinical trials[1] and other studies in which I can participate YES/NO
- I would like to be kept informed of new information and research in FH YES/NO
- I would like to be emailed a copy of the study results YES/NO
    - (if YES) my email address is: ……..

I hereby agree to contribute my health information to this Registry.

NAME: .....

SIGNATURE: ..... DATE: …….....

NAME OF WITNESS: .....

SIGNATURE OF WITNESS: ..... DATE: …….....

[1] **Please note** that if we inform your doctor about the existence of a trial, this does not imply that we endorse it. In order to participate in any trial, you and your child will need to discuss it with your family and your doctor and will be required to fill out a separate informed consent form that relates to that specific trial.

Appendix 4. Positive Results Package

PARTICIPANT REGISTRATION DETAILS

First name: _____

Family name: _____

Date of Birth (dd/mm/yyyy) _____

Address: _____

_____

_____

Postcode _____

Telephone: _____

Mobile phone: _____

Email: _____

If you would like to register directly with the Registry please provide the name of your doctor below giving us permission to contact your doctor directly if we require further information to complete your registration

You have my permission to contact my doctor for my personal details:

Doctors Name: _____

Clinic / Medical Practice Address:

_____

_____

_____

Clinic / Medical Practice Telephone:

_____

Specialist Name: _____

FH Registry MASTER Consent Form, Version 3, 10/06/2020
RPAH SITE SPECIFIC FH Registry Consent Form, Version 3, 10/06/2020