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Original Research

Developing a national implementation strategy for enhancing the detection of familial hypercholesterolemia: An Australian experience

Mitchell Sarkies, PhD*; David Sullivan, MBBS; Andrew Black, MBBS; Philip Aylward, MA (Oxon), BM, BCh, PhD; Karam Kostner, MD; Stephen J. Nicholls, MBBS, PhD; Ari Horton, MBBS; Walter P. Abhayaratna, MBBS, PhD; Nadarajah Kangaharan, MBBS; Jing Pang, PhD; Warrick Bishop, MD; Luke Elias, BAppSc (Physio); Shubha Srinivasan, BSc (Med), MBBS, PhD; Kathryn E. Waddell-Smith, MBBS, PhD; Jenny Della-Vedova, BCom; Charlotte Hespe, MBBS (Hons), DCH (Lon); Jane Tiller, PhD; Jeffrey Braithwaite, PhD; Karen Birkenhead, PhD; Gerald F. Watts, DSc, PhD

School of Health Sciences, Faculty of Medicine and Health, University of Sydney, Sydney, New South Wales, Australia (Drs Sarkies, Birkenhead); Implementation Science Academy, Sydney Health Partners, University of Sydney, Sydney, New South Wales, Australia (Dr Sarkies); Australian Institute of Health Innovation, Faculty of Health and Human Sciences, Macquarie University, Sydney, New South Wales, Australia (Drs Sarkies, Braithwaite); Department of Chemical Pathology, Royal Prince Alfred Hospital, NSW Health Pathology, Sydney, New South Wales, Australia (Dr Sullivan); Sydney Medical School, Faculty of Medicine and Health, University of Sydney, Sydney, New South Wales, Australia (Dr Sullivan); Menzies Institute for Medical Research, University of Tasmania, Hobart, Tasmania, Australia (Dr Black); Department of Cardiology, Royal Hobart Hospital, Hobart, Tasmania, Australia (Dr Black); South Australian Health and Medical Research Institute, Flinders University and Medical Centre, Adelaide, South Australia, Australia (Dr Aylward); Department of Cardiology, Mater Hospital, University of Queensland, Brisbane, Queensland, Australia (Dr Kostner); Victorian Heart Institute, Monash University, Melbourne, Victoria, Australia (Drs Nicholls, Horton); Victorian Heart Hospital, Melbourne, Victoria, Australia (Drs Nicholls, Horton); Monash Children's Hospital, Monash Health, Melbourne, Victoria, Australia (Dr Horton); Monash Cardiovascular Research Centre, Melbourne, Victoria, Australia (Dr Horton); Department of Paediatrics, Monash University, Melbourne, Victoria, Australia (Dr Horton); Cardiovascular Medicine, College of Medicine, Biology and Environment, Australian National University, Canberra, Australia (Dr Abhayaratna); Department of Cardiology, Royal Darwin Hospital, Darwin, Northern Territory, Australia (Dr Kangaharan); Menzies School of Health Research, Darwin, Northern Territory, Australia (Dr Kangaharan); Northern Territory Medical School, Flinders University, Adelaide, South Australia, Australia (Dr Kangaharan); School of Medicine, Faculty of Health and Medical Sciences, University of Western Australia, Perth, Western Australia, Australia (Drs Pang, Watts); Department of Cardiology, Calvary Cardiac Centre,

* Corresponding author at: Sydney School of Health Sciences, Faculty of Medicine & Health, The University of Sydney, Level 7 D18 Susan Wakil Health Building, Western Avenue, NSW 2006, Australia.
E-mail address: mitchell.sarkies@sydney.edu.au (M. Sarkies).
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Calvary Health Care, Tasmania, Australia (Dr Bishop); Health consumer, FH Australia, Sydney, New South Wales, Australia (Mr Elias); Institute of Endocrinology and Diabetes, The Children's Hospital at Westmead, Sydney, New South Wales, Australia (Dr Srinivasan); Discipline of Child and Adolescent Health, Faculty of Medicine and Health, University of Sydney, Sydney, New South Wales, Australia (Dr Srinivasan); Department of Cardiovascular Medicine, Flinders Medical Centre, Adelaide, South Australia, Australia (Dr Waddell-Smith); Health consumer, FH Australia, Perth, Western Australia, Australia (Ms Della-Vedova); General Practice and Primary Care Research, School of Medicine, The University of Notre Dame, Sydney, New South Wales, Australia (Dr Hespe); School of Public Health and Preventive Medicine, Monash University, Melbourne, Australia (Dr Tiller); Lipid Disorders Clinic, Cardiometabolic Service, Departments of Cardiology and Internal Medicine, Royal Perth Hospital, Perth, Western Australia, Australia (Dr Watts)

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BACKGROUND: Familial hypercholesterolemia (FH) is a common and treatable inherited condition of cholesterol metabolism that increases cardiovascular health risk. Less than 10% of people with FH have been detected and few receive recommended treatment.

OBJECTIVE: We report on the development of a national implementation strategy for enhancing the detection of FH in Australia.

METHODS: A modified intervention mapping study was conducted, through workshops to identify barriers and facilitators of FH detection, models for detection and their objectives. Practical implementation strategies were selected using a constructivist analysis and summaries of the strategies were disseminated for feedback. A case study showcase was used to monitor strategy implementation.

RESULTS: Potential models for FH detection were identified, including (1) use of pathology lab reports and recall; (2) screening in general practice, pharmacy, and Aboriginal Community Controlled Health Organizations; (3) screening in cardiovascular inpatients, outpatients, and rehabilitation; (4) universal screening; and (5) centralized coordination of primary-tertiary shared care. Implementation strategies were selected for each model to support their operationalization into routine care. A list of general implementation strategies was also produced that could support multiple models for FH detection.

CONCLUSION: We identified multiple FH detection models and strategies that would support implementation across Australia. Centralized coordination of a primary-tertiary shared care model was described as a promising approach for integrating index case detection with family cascade testing. We provide a set of implementation recommendations for policy and practice to enhance the detection of FH in Australia.

Introduction

Premature atherosclerotic cardiovascular disease (ASCVD) is a leading cause of death globally.[1] Familial hypercholesterolemia (FH) substantially increases ASCVD risk if left untreated, but early treatment significantly reduces morbidity and mortality.[2] FH is a treatable inherited condition of cholesterol metabolism that has a prevalence of one in 311 in the general population[3] and one in 20 among those with premature ASCVD.[4] FH is both under-detected and undertreated. Less than 10% of people with FH are aware of their condition[5] and only 20% achieve guideline-recommended low-density lipoprotein (LDL)-cholesterol goals after diagnosis.[6]

The persisting gaps between clinical practice guidelines in the care of FH has led to several global calls to action[7,8] and updated guidance.[9-12] However, implementation of clinical recommendations has often been less than optimal.[13,14] Several barriers to implementing detection of FH have been identified.[15] These include the costs of testing and limited capacity and capability among the health workforce to provide genetic counselling.[16] There are also privacy and potential discrimination concerns related to insurance coverage.[16] A lack of accessible and user-friendly information about FH and the perception that relatives will not understand the information limit genetic cascade testing,[17] a cost-effective method of detecting new cases in families.[18]

Implementing new models of care is a complex process that has become a major priority for health systems.[19] In Australia, the cost of genetic testing for FH is reimbursed by the Medicare Benefits Schedule, creating an opportunity to improve FH detection in the community. Action is needed to improve the organization and delivery of FH care.[20] Implementation science focuses on the design of strategies to change clinical practice and addresses barriers to improve FH care.[21] The FH Australasia Network harnessed an implementation science approach to develop a national implementation strategy for enhancing the detection of FH in Australia.

Materials and methods

Study design, participants, and setting

An 'intervention mapping' study[22] was undertaken to develop a national implementation strategy to improve the de-

Box 1. Five-step modified intervention mapping approach.

1. Undertake an implementation needs assessment.
2. Identify models for FH detection and implementation objectives.
3. Select practical and general implementation strategies.
4. Produce state and territory summaries of strategies to implement FH detection models.
5. Monitor, evaluate, and sustain implementation outcomes.

tection of FH. Intervention mapping facilitates collaborative planning by individuals of varying backgrounds to develop interventions. It involves a multi-step process for decisions based on a combination of theory and empirical evidence (Box 1).

This approach was selected to ensure solutions were matched to the contextual factors influencing each Australian jurisdictional state ($n = 6$) and territory ($n = 2$). Australia's health system is a shared responsibility between the federal government (overseeing primary care) and state/territory governments (managing public hospitals and public health programs). Perspectives were sought from a purposive sample of key FH stakeholders drawn from each of the 8 jurisdictions, including representation from 10 healthcare stakeholders: patients, general practice, physicians, pediatrics, precision medicine, pathology, nurse practitioner, pharmacy, policy makers, and the pharmaceutical industry (Fig 1). Ethical approval was provided by Macquarie University (Project ID 11,325).

Data collection and analysis

Step 1: Undertake an implementation needs assessment

Four, 2-hour virtual workshops were run at the 2021 Australasian FH Summit to identify the barriers and facilitators of FH detection in Australia (Appendix 1). The workshops were attended by 28 attendees (6-8 each group) and led to the identification of 13 barriers and 10 facilitators at the patient-, provider-, and system-levels. The full publication describing the identification of barriers and facilitators for FH detection in Australia is available in *Heart, Lung and Circulation* at https://doi.org/10.1016/j.hlc.2023.09.012. [15]

Step 2: Identify models for FH detection and implementation objectives, mapped to the consolidated framework for implementation research (CFIR)

Eight face-to-face focus groups were undertaken with 100 key stakeholders across each Australian state and territory between July 2022 and March 2023 to identify models for FH detection, implementation objectives for each model, and specific and general implementation strategies to support them. The focus groups were co-hosted by local clinical key opinion leaders (DS, SN, AH, KK, GW, PA, AB, WB, WA, NK) and an implementation scientist (MS) as a pharmaceutical industry sponsored medical education dinner meeting. Each meeting began with a 15-minute presentation, followed by a facilitated discussion focused on 3 topics: (1) state model for FH index case screening, (2) integrating cascade testing within index case screening, and (3) implementation strategies to support the detection of FH. Each focus group was 2 hours in duration with attendance ranging between 7 and 16 individuals. The discussions were audio recorded and transcribed verbatim.

Step 3: Select practical and general implementation strategies, mapped to the expert recommendations for implementing change taxonomy

A constructivist analysis was undertaken by 4 researchers (MS, KB, CH, NG) using NVivo software.[23] Transcripts and field notes were coded line-by-line using both inductive and deductive logics. This analysis process followed 4 iterative stages: (1) inductive coding of models to improve the detection of FH and their implementation objectives; (2) inductive coding of practical implementation strategies specific to each implementation objective and general implementation strategies that could support multiple objectives; (3) deductive mapping of implementation objectives to the CFIR domains;[24] and (4) deductive mapping of the practical and general implementation strategies to the Expert Recommendations for Implementing Change (ERIC) taxonomy.[25] Refer to Appendix 2 and Appendix 3 for deductive mapping to the CFIR domains and ERIC taxonomy.

Step 4: Produce state and territory-based summaries of strategies to implement detection models for FH

Following each focus group, the implementation science facilitator (MS) prepared a draft summary of the strategies to improve the detection of FH in each state and territory. This summary was reviewed and modified by the local clinical key opinion leader before being distributed to the attendees. The summaries contained the models for FH index case screening, approaches to integrate family cascade genetic testing with diagnostic index case screening, and the implementation strategies to support the detection of FH that were discussed in each group. A list of potential short-, medium-, and long-term actions were provided for each group to consider.

Step 5: Monitor, evaluate, and sustain implementation outcomes

State and territory updates on efforts to improve FH detection were showcased at the 2023 FH Summit. The 2023 FH Summit was hosted face-to-face by the FH Australasia Network in Sydney, Australia, during September 2023. Two local clinical key opinion leaders from each state and territory (1 adult and pediatric physician) outlined local case studies to improve FH detection following the focus groups. Several implementation research projects have commenced across Australia, focused on either index case detection or facilitating cascade testing of relatives.[26-30]

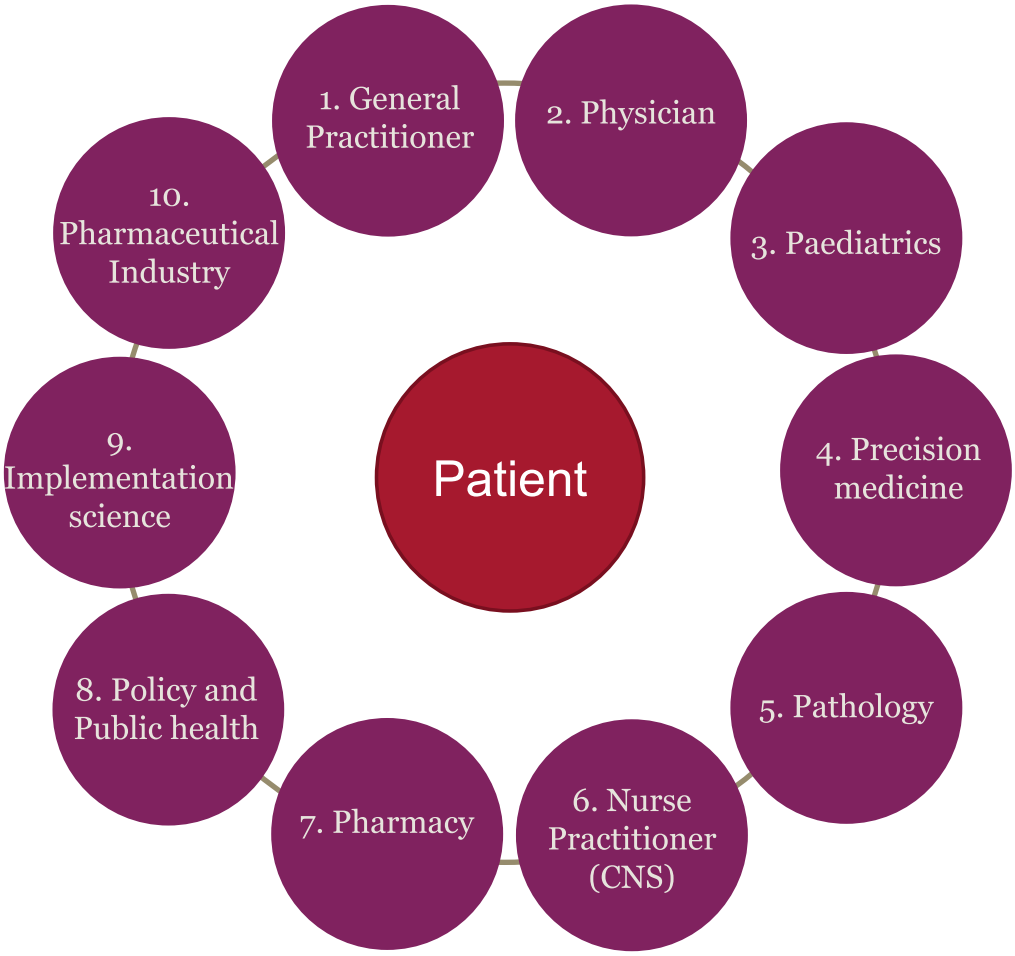

Figure 1. Familial hypercholesterolemia (FH) Australasia Network 10-P approach.

Results

The national implementation strategy for enhancing the detection of FH is presented in Figure 2. Four models for index case detection and 1 model for family cascade testing were identified. A total of 16 implementation objectives were selected for the FH detection models and practical implementation strategies were matched to each objective. Several general implementation strategies were also outlined to support multiple models for FH detection.

Index case detection

Pathology lab reports and recall

Public and private pathology providers can supply requesters with FH alerts and interpretive comments on laboratory reports for plasma lipid profiles.[31,32] While many providers have already implemented these alerts, this was not universal. Inconsistency was reported in the language used for interpretive comments between providers. It was stated that pathology providers hold significant datasets on previous plasma lipid profiles that could be utilized for FH detection. Pathology providers could generate a list of people who potentially have FH based on a predetermined LDL-cholesterol threshold, whereby the requestors could be contacted and notified of this risk. Secondary testing for additional diagnostic investigations was also raised as an option for identifying potential FH cases, particularly in younger patient cohorts where the benefits of early treatment are the most profound. However, limitations would need to be overcome to secondary testing including financial reimbursement. Implementation objectives and strategies for pathology lab reports and recall are presented in Table 1.

General practice, pharmacy, and aboriginal community controlled health organizations

Primary care was identified as an important setting to detect FH, given the frequency that the population visits their general practitioner or other primary care provider. There was uncertainty regarding the best model for detecting FH in primary care,[33] with a combination of both point of care tools and asynchronous approaches suggested. Several FH case finding tools have been developed for general practice software, such as the Canning Tool[34] and TARB-Ex,[35] which enable data extraction and case identification using established clinical criteria. Practice management software alerts based on algorithms that consider LDL-cholesterol in combination with other criteria was suggested to prompt general practitioners (GPs) to consider the risk of FH at

Figure 2. Implementation strategy for familial hypercholesterolemia (FH) detection.
ACCHO, Aboriginal Community Controlled Health Organization; EMR, electronic medical record.

Table 1. Implementation objectives and strategies for the use of pathology lab reports and recall.

Implementation objective	Practical implementation strategies
1. Pathology report alerts: Establish alert threshold for public and private pathology reports and include interpretive comments for requester to consider FH screening	Standardize alert wording across public and private pathology labs Use simplified alert wording that retains clinical autonomy Provide diagnostic and management pathway recommendations Offer multiple patient pathways by partnering with private providers
2. Pathology database screening: Generate list of patients with high cholesterol and contact requester to suggest review and FH screening, particularly for young patients	Pathology provider phone call with requesting clinician to recommend FH testing Standardized pathology letter to requesting clinician recommending testing
3. Secondary testing: Screen for FH when undertaking tests for other conditions, for example, screen total cholesterol when undertaking electrolyte liver profile for development delay or add LDL-cholesterol when requesting iron levels	Streamline process for undertaking a full lipid profile in addition to commonly performed tests Streamline process for undertaking secondary tests for additional diagnostic investigations in people with high total cholesterol Explore feasibility of pathology providers automatically providing a full lipid panel when only total cholesterol level has been requested

Abbreviations: FH, familial hypercholesterolemia; LDL, low density lipoprotein.

point of care. Community pharmacy was also considered an opportune setting to screen people for FH, particularly when filling scripts for lipid lowering medications.[36] Implementation objectives and strategies for general practices, pharmacies, and Aboriginal Community Controlled Health Organizations are presented in Table 2.

Cardiovascular inpatients, outpatients, and rehabilitation

The identification of people with FH in cardiovascular inpatients,[37,38] outpatients and rehabilitation settings was considered potentially high yielding, given the higher prevalence of FH in the presence of ASCVD.[3,39] It was thought that proactive inpatient case finding needed to be delegated to

Table 2. Implementation objectives and strategies for general practice, pharmacy, and Aboriginal Community Controlled Health Organization.

Implementation objective	Practical implementation strategies
1. FH case finding tool: Use FH screening and data extraction tool to identify and recall patients with high cholesterol	Update general practice cholesterol screening guidelines to identify younger cohorts potentially at risk of FH Create GP education programs, linked to FH identification and quality improvement Practice Incentive Program Use a pre-appointment waiting room patient screening tool Incorporate FH screening and data extraction tool into local FH HealthPathways
2. Practice management software alert: Embed pathology alert for high LDL-cholesterol in practice software	Use lower LDL-cholesterol thresholds for identification and testing reimbursement in pediatrics Provide clear FH care pathway with action to be taken in response to alert flag in practice software Create patient alerts by sending an SMS text message reminder to discuss pathology result with their GP
3. Community pharmacy: Routine pharmacist FH screening procedures	Recall SMS text message to encourage cholesterol testing for at risk patients Advise younger patients filling statin scripts to get tested and notify their family members of potential risk Complete DLCNS in hospital and community settings Partner with private health insurance providers for funding

Abbreviations: FH, familial hypercholesterolemia; LDL, low density lipoprotein; GP, general practitioner; SMS, short message service; DLCNS, Dutch Lipid Clinic Network Score.

a specific staff member and incorporated into their routine patient review process. Point of care alerts in the electronic medical record require nuanced algorithms to ensure specificity of those patients flagged and would work best by triggering a standard order set and templates for diagnostic criteria. Data contained in the electronic medical record, and other databases and registries was thought to be useful for identifying FH cases. It was suggested that screening these data sources could form research projects for students or clinical staff, which could be linked to quality assurance where patients are notified of risk and recalled to clinics. In the outpatient and cardiac rehabilitation setting, nursing and allied health staff could be trained to undertake a Dutch Lipid Clinic Network Score as part of routine assessments. Implementation objectives and strategies for cardiovascular inpatients, outpatients and rehabilitation are presented in Table 3.

Universal screening

Universal screening was viewed as a promising development. FH was seen to meet all the World Health Organization (WHO) criteria for screening[40] and that universal screening of children could enable the early detection of homozygous FH.[12] Two potential models for undertaking universal screening for FH were identified: (1) newborn and childhood screening and (2) general population screening. It was agreed that the most appropriate pathway for newborn screening of FH would be to nominate for inclusion in the national genetic newborn bloodspot screening program. This process would require a Medical Services Advisory Committee application according to the Newborn Screening National Policy Framework. It was thought that universal screening of children should be paired with cascade testing of family members of affected individuals, particularly child-parent testing. Determining the process for centralized coordination was considered imperative prior to the implementation any newborn screening process paired with cascade testing of relatives, including the consenting of confirmed FH cases into the National FH Registry.[41] Care pathways need to be developed in partnership with primary care providers and consumer advocates. There were also existing research programs testing the screening of FH in the general population.[26] It was suggested that funding could be sought to scale up these projects, if deemed successful. Suggestions for screening of FH across the lifecycle included offering LDL-cholesterol testing with genetic testing for positive cases or genetic testing in coordination with the National Immunization Program, offering genetic testing to all hospital inpatients, or when undertaking preconception screening. Implementation objectives and strategies for universal screening are presented in Table 4.

Family cascade testing

Centralized coordination of primary-tertiary shared care

Centralized coordination of family cascade testing in partnership with primary care providers was viewed as a potentially sustainable approach. GPs have access to an Australian Medicare Benefits Schedule (MBS) Item that reimburses the cost of genetic testing of patients who have first- or second-degree relatives with FH. Hospital lipid clinics commonly undertake confirmatory diagnosis of FH through genetic testing which generates an index case within a family dyad. The cascade testing of relatives can then be undertaken by the relative's GP with support and coordination from hospital lipid clinics who diagnosed the original index case. The establishment of public centers for central coordination of cascade testing was also described. It was thought that this

Table 3. Implementation objectives and strategies for cardiovascular inpatients, outpatients, and rehabilitation.

Implementation objective	Practical implementation strategies
1. Proactive inpatient case finding: Delegate responsibility for screening for FH during inpatient admission to a specific staff member, particularly for young patients	Speak with family during inpatient admission Local hospital education and awareness campaign for staff and patients Protocolize collecting inpatient LDL-cholesterol using standardized hospital order set on admission for coronary events Add DLCNS to cardiac nurse review and improve FH coding in past history
2. EMR alerts: Automatically screen and alert potential FH patients in EMR, particularly young patients	Use algorithm to increase specificity of FH alerts Trigger a standard order set for cholesterol testing, DLCNS or other diagnostic processes using a pathway and EMR template for ward rounds Add DLCNS to discharge summary checklist Order genetic test directly, bypassing referral to outpatient clinic Utilize existing single digital patient record where available
3. EMR, database and registries: Identify and recall potential FH cases post-event using existing databases and registries and then cascade screen relatives	Adopt approaches used for other conditions to contact potential index cases and their relatives Calculate untreated LDL-cholesterol using algorithm for those on cholesterol lowering therapy Undertake a small collaborative project involving junior medical staff, trainees and/or research students Provide patients with information on what to do after diagnosis Ensure capture of FH in ICD-10 codes
4. Nursing and allied health screening: Staff to screen for people with high LDL-cholesterol and trigger family cascade testing process	Train nursing and allied health staff to take detailed family history and screen for FH with DLCNS Add DLCNS into standard cardiac rehab assessment Standardize requesting cholesterol levels in cardiac outpatient clinics Add FH to standard patient education in cardiac rehab Ensure Heart Foundation cardiac rehab patient brochure includes information on familial risk

Abbreviations: FH, familial hypercholesterolemia; EMR, electronic medical record; LDL, low density lipoprotein; DLCNS, Dutch Lipid Clinic Network Score; ICD, International Classification of Diseases.

Table 4. Implementation objectives and strategies for universal screening.

Implementation objective	Practical implementation strategies
1. Genetic newborn screening: Nominate for inclusion in the genetic newborn bloodspot screening program according to the Newborn Screening National Policy Framework	Determine process for a centralized database, consent, handling of incidental findings, non-paternity, notification of confirmed cases and their relatives, and other issues prior to implementation Develop care pathways for FH in partnership with primary care providers Ensure process for consenting confirmed FH cases into the National FH Registry Include consumer voice in nomination to advocate for policy change
2. Childhood screening: Screening before puberty, preferably between 1 and 2 y	Offer the option of LDL-cholesterol testing first, followed by genetic testing for positive cases, or genetic testing first, aligned with National Immunization Program schedule
3. General adult population screening: Screening at different stages of the life cycle	Seek funding to scale up screening of adult populations using an opt-in postal saliva genetic testing kit Offer to include genetic testing for FH during pre-conception screening Offer genetic testing for all inpatient hospital admissions

Abbreviations: FH, familial hypercholesterolemia; LDL, low density lipoprotein.

could be implemented by nursing or allied health professionals through family kindred clinic appointments or home visits for testing of relatives. The identification of potential public-private partnerships and utilization of existing private services for genetic counselling through pathology providers was thought to be important to access ancillary support for the family cascade testing process. Implementation objectives and strategies for centralized coordination of primary-tertiary shared care are presented in Table 5.

General implementation strategies

General implementation strategies were described in addition to the specific implementation objectives and strategies for each potential FH detection model. These general strategies were thought to underpin a coordinated, state- and territory-based approach to improving the detection of FH nationally. Using evaluative strategies to monitor and evaluate efforts to improve FH care was consistently raised, with specific reference to using the National FH Registry[41] for this

Table 5. Implementation objectives and strategies for centralized coordination of primary-tertiary shared care.

Implementation objective	Practical implementation strategies
1. Public-private partnership: To provide ancillary support for the family cascade testing process	Access genetic counselling services available from existing private pathology providers Establish dedicated role to coordinate the process in nurse, allied health, or genetic counsellor led clinic, funded through public-private partnership Seek private health insurance funded program by demonstrating exemplar model that would save resources
2. Coordinating center: Establish public center for central coordination of cascade testing	Provide a family notification letter to the index case to provide to their relatives Undertake family kindred clinic appointments or home visits for FH testing of family members, facilitated through family reunions Undertake direct notification of relatives with permission from index case, in addition to indirect notification of relatives via the index case Establish dedicated role to coordinate the process in nurse, allied health, or genetic counsellor led public clinic through activity-based funding Employ GPs or other specialists to fund clinic through MBS Item bulk billing and order FH tests Ensure relatives diagnosed with FH are included in the National FH Registry for ongoing management and quality improvement
3. Central coordination in primary care: Provide index case with information for relatives, obtain contact details for relatives and their GP, contact relatives and their GP to provide information on cascade testing pathway	Provide detailed instructions and tools for relatives to undertake cascade testing with their GP Text message reminders to patients from GP software Obtain consent for National FH Registry Identify options for no cost support to GPs when providing genetic counselling, including telehealth and virtual access to the hospital lipid clinic for advice Provide stratified risk reduction pathways for positive cases

Abbreviations: FH, familial hypercholesterolemia; GP, general practitioner; MBS, Medicare Benefits Schedule.

purpose. Interactive assistance was thought to be imperative, particularly to support GPs undertaking family cascade testing. Each state and territory were working with a different set of local circumstances, infrastructure, and funding for FH services that would require adaptation and tailoring of FH detection models; for example, by providing rural outreach services and access to virtual and telehealth services where population distribution is spread over large geographic areas. Developing stakeholder relationships was said to underpin all efforts to improve FH detection, focusing on partnerships between the key groups and organizations, and improving communication between hospitals and general practice. Training and educating stakeholders is needed to support all FH detection models. Supporting clinicians at point of care by providing access to advice and information about FH testing was raised, with special mention about the importance of ongoing liaison between the hospitals and general practice. Engaging consumers was mentioned at both the public health messaging level and supporting individual patients and families to make informed decisions about their care. Several financial strategies were suggested, including accessing new funding options (eg, new MBS Items, research grants, and philanthropy) for detecting FH and making use of existing funding options (eg, existing MBS Items and hospital activity-based funding). Some states and territories were interested in changing the existing infrastructure to support FH detection, particularly those with limited or no access to public hospital lipid clinics. General implementation strategies to support models for FH detection are presented in Table 6.

Discussion

We developed a compendium of implementation objectives and practical implementation strategies for each identified model for FH detection in Australia. Several models for index case detection were described across healthcare settings and a centralized coordination of a primary-tertiary shared care model was described as the key approach for integrating index case detection with family cascade testing of first- and second-degree relatives. Each model for FH detection was paired with potential implementation strategies that could be used depending on the local contextual circumstances in each state and territory. Several general implementation strategies were outlined to support multiple models for FH detection. Implementation recommendations for policy and practice are provided in Table 7.

There was no agreement on a single model for FH detection across Australia. This aligns with professional consensus that an integrated approach to universal, cascade, and opportunistic FH screening programs must be compatible with each jurisdiction's local needs.[42] By way of example, there is 1 state-wide public pathology provider in New South Wales making the standardization of alert wording a relatively simple exercise in the public setting, whereas there are multiple private and public pathology providers elsewhere requiring a more concerted effort to develop consensus on standardized alert wording for FH across labs. Our comprehensive approach ensured that the national implementation strategy for enhancing the detection of FH in Australia aligns with

Table 6. General implementation strategies to support models for familial hypercholesterolemia detection.

Use evaluative strategies:

- Conduct audit and feedback using datasets such as the National FH Registry

Provide interactive assistance:

- Hospital lipid clinic outreach to support GPs with family cascade testing

Adapt and tailor to context:

- Adapt approaches and messaging from other clinical conditions, such as bowel cancer screening
- Offer virtual and telehealth access to FH services, and rural outreach to provide face-to-face services

Develop stakeholder interrelationships:

- Engage with key stakeholders to develop a national call to action on FH
- Partner with academic institutions and involve junior staff, students and multidisciplinary teams in quality improvement research
- Identify special interest GPs as key opinion leaders and champions for FH
- Create and promote new and existing referral and treatment pathways for FH

Train and educate stakeholders:

- Embed FH education into health professional qualifications
- Distribute FH point of care resources for clinicians and patients, such as HealthPathways
- Develop accredited continual professional development education linked to quality improvement program for FH
- Update clinician resources to include information on FH, such as *Therapeutic Guidelines* and CARPA Standard Treatment Manual
- Publish article to promote FH for *Australian Family Practitioner* and *Medicine Today* journals
- Record Royal Australasian College of Physicians (RACP) – *Pomegranate Health* podcast episode on FH
- Include FH in cardiovascular events and showcases
- Disseminate key messages via social media pages frequented by GPs

Support clinicians:

- Establish systems for automatic relay of information between hospital lipid clinics and general practice and vice versa
- Establish system for requesters to know where pathology testing is being conducted

Engage consumers:

- Establish annual FH awareness week
- Align public awareness campaigns with health priorities and target different population groups using empowering messaging
- Establish FH patient advocacy group and support government lobbying by making use of patient stories
- Provide patients with written resources using accessible language, such as communication aids
- Develop and use existing patient self-management tools
- Support patient mediated reminder prompting of clinicians regarding their cholesterol levels

Utilize financial strategies:

- Produce cost effectiveness business case to encourage political, funder, organization or philanthropic support for programs
- Advocate for FH research funding and utilize as a mechanism for pilot projects and ongoing activity
- Utilize existing MBS Items to incentivize detecting and managing FH
- Apply for MBS Items for GPs, pediatric FH genetic testing, genetic counselling and nursing services

Change infrastructure:

- Promote family cascade testing as an essential component of care using behavioral nudges
- Incorporate pathology, family history, and index case in the medical record
- Establish public hospital lipid clinics where services aren't available

Abbreviations: FH, familial hypercholesterolemia; GP, general practitioner; MBS, Medicare Benefits Schedule; CARPA, Central Australian Rural Practitioners Association.

Table 7. Implementation recommendations for policy and practice.

1.	Public and private pathology providers should include laboratory report interpretative comments highlighting FH
	a. Simple, standardized wording should be used to retain requester autonomy and provide multiple diagnostic and management pathway recommendations
2.	General practice and Aboriginal Community Controlled Health Organizations should implement FH screening of health records as routine quality assurance, which may include:
	a. Electronic data extraction and screening tools
	b. Pre-appointment waiting room patient screening tools
	c. Practice software alerts for LDL-cholesterol over 5.0 mmol/L
	d. Patient SMS text message reminders to discuss cholesterol and FH with their GP
3.	Hospital coronary care units should test LDL-cholesterol and screen for FH using the DLCNS for acute coronary syndrome patients with LDL-cholesterol levels over 5.0 mmol/L
	a. Untreated LDL-cholesterol levels can be calculated for those on cholesterol lowering therapy
	b. Electronic medical record alerts can be considered to trigger DLCNS screening
	c. Cardiac rehabilitation programs can add the DLCNS to routine clinical assessment
4.	Universal screening programs for early detection, diagnosis, and management of FH can be implemented using 1 or a combination of approaches, if considered cost effective:
	a. Nominate for inclusion in the national genetic newborn bloodspot screening program
	b. Opt-in genetic testing of the population using postal saliva testing kits
	c. LDL-cholesterol testing (with genetic testing for positive cases) of children using age- and sex (males or females)-specific criteria or genetic testing coinciding with the National Immunization Program Schedule
5.	Family cascade genetic testing should be coordinated by a centralized service
	a. Direct or indirect notification of first- and second-degree relatives should be facilitated by the centralized service using family letters, kindred clinic appointments, or home visits for family member testing
	b. Relatives can undertake testing via their GP with coordination from a centralized service using MBS Item 73353
	c. Genetic counselling services can be accessed from private pathology providers through public-private partnerships
6.	General implementation strategies can support models for FH detection, which may include:
	a. Audit and feedback of FH detection and management using the National FH Registry
	b. Establish FH patient advocacy group and partner with key stakeholders to run targeted public awareness campaigns and undertake quality improvement research
	c. Distribute existing educational resources, establish point of care resources for clinicians and patients (eg, HealthPathways), and develop accredited continual professional development education linked to quality improvement programs
	d. Add FH to *Therapeutic Guidelines* used by GPs and update CARPA Standard Treatment Manual to include FH information
	e. Utilize existing financial reimbursement and explore opportunities for philanthropic and commissioning funding to establish new or expand existing lipid clinics and FH services

Abbreviations: FH, familial hypercholesterolemia; LDL, low density lipoprotein; SMS, short message service; DLCNS, Dutch Lipid Clinic Network Score; MBS, Medicare Benefits Schedule; GP, general practitioner; CARPA, Central Australian Rural Practitioners Association.

the National Health Genomics Policy Framework Implementation Plan and Australia's National Roundtable on Cholesterol.[43]

A more cohesive approach was identified for integrating the detection of people with FH and cascade testing of their relatives. A sustainable source of financing for cascade testing was identified through the listing of genetic testing for FH on the Australian Medicare Benefits Schedule in 2020 and ability for GPs to make reimbursable pathology requests for genetic testing of patients who have first- or second-degree relatives with FH. Many people visit their GP frequently and undertake regular LDL-cholesterol testing, enabling the integration of cascade genetic testing for FH into routine workflows and systems of care for other conditions. Previous qualitative research indicates that GPs are comfortable undertaking genetic testing if appropriate supports are available.[44] The central coordination of family cascade testing by hospital lipid clinics was seen as essential, given this was the setting where many FH patients are diagnosed and undertake genetic testing to enable the cascade testing process to commence.

The lack of comprehensive FH screening programs in Australia and reliance on smaller scale institution- and jurisdiction-specific initiatives necessitated a comprehensive approach to identify both the potential models for FH detection and implementation strategies to support them. There were limitations to our approach. Each jurisdiction focused on different models for FH detection, which limited the ability to generalize. Not all jurisdictions will be able to implement each FH detection model and implementation strategy described in our study. Prioritization of implementation efforts within each state and territory is necessary based on experience from previous pilot projects, local processes, systems, and infrastructure available.

Conclusion

We report on the development of a national implementation strategy for enhancing the detection of FH in Australia. Updated Australian clinical guidance and expansion of the Medicare Benefits Schedule reimbursement for genetic testing set the scene for state and territory ambitions to improve the detection of FH. Multiple screening models and implementation strategies to support them were identified across each state and territory and a set of nationally cohesive implementation recommendations for policy and practice is provided. Implementation of these recommendations for different screening models provides an opportunity for future research to evaluate the effectiveness of implementation strategies beyond observational investigations.

CRediT authorship contribution statement

Mitchell Sarkies: Writing – review & editing, Writing – original draft, Visualization, Supervision, Resources, Project administration, Methodology, Investigation, Funding acquisition, Formal analysis, Data curation, Conceptualization. **David Sullivan:** Writing – review & editing, Supervision, Methodology, Investigation, Funding acquisition, Conceptualization. **Andrew Black:** Writing – review & editing, Supervision, Resources, Project administration, Investigation, Funding acquisition. **Philip Aylward:** Writing – review & editing, Supervision, Resources, Project administration, Investigation, Funding acquisition. **Karam Kostner:** Writing – review & editing, Supervision, Resources, Project administration, Investigation, Funding acquisition. **Stephen J. Nicholls:** Writing – review & editing, Supervision, Resources, Project administration, Investigation, Funding acquisition. **Ari Horton:** Writing – review & editing, Supervision, Resources, Project administration, Investigation, Funding acquisition. **Walter P. Abhayaratna:** Writing – review & editing, Supervision, Resources, Project administration, Investigation, Funding acquisition. **Nadarajah Kangaharan:** Writing – review & editing, Supervision, Resources, Project administration, Investigation, Funding acquisition. **Jing Pang:** Writing – review & editing, Supervision, Resources, Project administration, Investigation, Funding acquisition. **Warrick Bishop:** Writing – review & editing, Supervision, Resources, Project administration, Investigation, Funding acquisition. **Luke Elias:** Writing – review & editing, Supervision, Resources, Project administration, Methodology, Investigation, Funding acquisition, Conceptualization. **Shubha Srinivasan:** Writing – review & editing, Supervision, Resources, Project administration, Investigation, Funding acquisition. **Kathryn E. Waddell-Smith:** Writing – review & editing, Supervision, Resources, Project administration, Investigation, Funding acquisition. **Jenny Della-Vedova:** Writing – review & editing, Supervision, Resources, Project administration, Investigation, Funding acquisition. **Charlotte Hespe:** Writing – review & editing, Supervision, Resources, Project administration, Investigation, Funding acquisition. **Jane Tiller:** Writing – review & editing, Investigation. **Jeffrey Braithwaite:** Writing – review & editing, Supervision, Resources, Methodology, Funding acquisition, Conceptualization. **Karen Birkenhead:** Writing – review & editing, Formal analysis, Data curation. **Gerald F. Watts:** Writing – review & editing, Supervision, Resources, Project administration, Investigation, Funding acquisition.

Ethical approval

Macquarie University Human Research Ethics Committee approval was granted for this research (REF: 11,325).

Use of AI and AI-assisted technologies statement

No AI or AI-assisted technologies were used in the preparation of this work.

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Declaration of competing interests

Mitchell Sarkies – All support for the present manuscript: Heart Foundation Vanguard Grant, NSW Health Translational Research Grant Scheme (H22/4011) grant, and Amgen Australia; Leadership or fiduciary role in other board, society, committee or advocacy group, paid or unpaid: FH Australia Professional Advisory Body membership. David Sullivan – Grants or contracts from Regeneron, Amgen, Arrowhead, Ionis, and Novartis; Consulting fees: Amgen and Novartis; Payment or honoraria for lectures, presentations, speakers bureaus, manuscript writing or educational events: Amgen and Novartis; Participation on a Data Safety Monitoring Board or Advisory Board: LIPID Study, FIELD Study, and Nomad Study, NHMRC Clinical Trials Centre; Leadership or fiduciary role in other board, society, committee or advocacy group, paid or unpaid: Clinical Committee Chairman, Australian Atherosclerosis Society Executive; Receipt of equipment, materials, drugs, medical writing, gifts or other services: Sanofi Alirocumab samples. Andrew Black –

None declared. Philip Aylward – Grants or contracts: CSL and Janssen; Payment or honoraria for lectures, presentations, speakers bureaus, manuscript writing or educational events: Novartis, Boehringer Ingelheim, Astra Zeneca, Amgen, and CSL; Leadership or fiduciary role in other board, society, committee or advocacy group, paid or unpaid: Novartis, Boehringer Ingelheim, Astra Zeneca, Amgen, and CSL. Karam Kostner – None declared. Stephen J Nicholls – Grants or contracts: Amgen, Anthera Pharmaceuticals, AstraZeneca, Cerenis Therapeutics, CSL Behring, Eli Lilly and Company, Esperion Therapeutics, Infraredx, New Amsterdam Pharma, Novartis, Resverlogix, and Sanofi Regeneron; Consulting fees: Akcea Therapeutics, Amgen, AstraZeneca, Boehringer Ingelheim, CSL Behring, Eli Lilly and Company, Esperion Therapeutics, Kowa Pharmaceuticals, Merck, Novo Nordisk, Pfizer, Sanofi Regeneron, Daichii Sankyo, Vaxxinity, Cyclarity, CSL Sequirus and Takeda. Ari Horton – None declared. Walter P. Abhayaratna – None declared. Nadarajah Kangaharan – None declared. Jing Pang – Grants or contracts: National Health and Medical Research Council, Royal Perth Hospital Research Foundation, Department of Health WA. Warrick Bishop – Payment or honoraria for lectures, presentations, speakers bureaus, manuscript writing or educational events: Amgen, Sanofi, and Novartis; Payment for expert testimony: Lexmedicus; Support for attending meetings and/or travel: Novartis; Leadership or fiduciary role in other board, society, committee or advocacy group, paid or unpaid: CSANZ Prevention Taskforce. Luke Elias – Support for attending meetings and/or travel: Novartis Australia. Shuba Srinivasan – None declared. Kathryn E Waddell-Smith – Payment or honoraria for lectures, presentations, speakers bureaus, manuscript writing or educational events: Pfizer Honoraria for speaking; Participation on a Data Safety Monitoring Board or Advisory Board: CSL Payment for participating in advisory board. Jenny Della-Vedova – All support for the present manuscript: WA Department of Health funding for genetic testing to FH Australia; Payment or honoraria for lectures, presentations, speakers bureaus, manuscript writing or educational events: Murdoch University. Charlotte Hespe – None declared. Jane Tiller – Grants or contracts: National Health and Medical Research Council; Payment or honoraria for lectures, presentations, speakers bureaus, manuscript writing or educational events: Pfizer and Rare Cancers Australia; Support for attending meetings and/or travel – Pfizer, Rare Cancers Australia, Pathology Technology Australia, Zero Childhood Cancer, and St Aloysius College; Leadership or fiduciary role in other board, society, committee or advocacy group, paid or unpaid: Rare Voices Australia. Jeffrey Braithwaite – None declared. Karen Birkenhead - Grants or contract: Post Doctoral Research position is funded by a NSW Health Translational Research Grant (TRGS) Scheme. Gerald F Watts – Grants or contracts: Amgen, Arrowhead, Novartis; Consulting fees: Amgen, Novartis, Arrowhead, Novo Nordisk, CSL Sequirus, and Esperion; Payment or honoraria for lectures, presentations, speakers bureaus, manuscript writing or educational events: Amgen, CSL Sequirus, Arrowhead, Novo Nordisk, Novartis; Support for attending meetings and/or travel – Novartis, Arrowhead, and Amgen.

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Data statement

Individual focus group data is not available to others due to the potential risk for re-identification of participants and the clinical sensitivity of some discussions that took place. This is in line with our human research ethics committee approval for the study, which requires the protection of privacy for participants.

Supplementary materials

Supplementary material associated with this article can be found, in the online version, at doi:10.1016/j.jacl.2025.03.004.

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