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Developing general practitioner and consumer supports for genomics in Australian primary care: a mixed-methods protocol

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ABSTRACT

Introduction This project will establish a nationally consistent and ethically defensible approach to embed genomic testing in Australian primary care. Many non-genetic health professionals (eg, general practitioners (GPs) and other specialists) have limited experience with such testing. Current tests—both subsidised and consumer-paid—target a range of genes and conditions, making appropriate selection challenging. A structured implementation approach is therefore crucial. We will develop, test, refine and evaluate internationally relevant tools to support GPs and consumers in using genomics effectively.

Methods and analysis Aims of the project are to (1) develop, implement and evaluate key supports for GPs offering tests through three interventions: primary health point-of-care resources, a practical guide to dealing with ethical issues affecting clinicians and established recommendations for a national approach for genetic counsellors to support GPs providing genetic testing; (2) develop and evaluate consumer resources and plan the implementation strategies; (3) evaluate real-world utilisation and equity of access to genetic testing in primary care using linked Medicare and population data. This project will focus on two genomic applications recently made available in Australia on the universal insurance scheme (Medicare): a reproductive genetic carrier screen (an example of the role of genetics in reproductive testing) and genetic testing for familial hypercholesterolaemia (an example of a condition-specific test). Both tests can be complex for GPs to understand and explain to consumers and have potential implications beyond the purpose of the test (eg, personal health implications for carriers and results are also relevant to genetic relatives). Developing a robust clinical pathway and process for these tests will prepare GPs for future more complex applications of clinical genomics. The study will take place from January 2024 to December 2026.

Ethics and dissemination Ethical approval for this work has been received from the Macquarie University Human Research Ethics Committee (Ref: 520241849560183) and the Royal Children's Hospital Research Ethics and Governance (HREC/112451). Findings will be disseminated via publications, conferences and engagement with primary care networks and policymakers.

STRENGTHS AND LIMITATIONS OF THIS STUDY

⇒ This work builds on the knowledge and understanding from the nationwide Mackenzie's Mission study of reproductive genetic carrier screening and builds on existing strategies and knowledge exchange networks.
⇒ Leveraging existing platforms such as HealthPathways and clinical decision support tools increases the feasibility and uptake of resources in routine practice.
⇒ The use of established implementation science frameworks (eg, the Behaviour Change Wheel) ensures systematic evaluation and refinement of interventions.
⇒ Limitations include difficulties generalising evaluation results from our set of participants, especially against the backdrop of a dynamic, diverse and fast-changing primary care sector.

INTRODUCTION

Clinical genomics, the testing of all or a large proportion of an individual's genetic makeup, has great promise for more effective patient care.[1] There are many possible applications of clinical genomic testing, such as diagnosing genetic conditions (including rare and ultra-rare conditions),[2] providing information relevant to reproductive choices,[3]

guiding treatment choices in cancer[4] and tailoring individual medication choices.[5] While the scientific and technological challenges of genomic testing are being met through an extensive research agenda studying clinical effectiveness, utility and economic benefits,[6 7] the *practice* of genomics—especially by non-genetic specialists—remains a challenging and underexplored area.[8]

Until recently in Australia, the practice of genomics was almost entirely led by the nation's state-based public genetics services, provided by genetic specialists such as clinical geneticists and genetic counsellors.[9] Most of these publicly funded services are based in metropolitan or regional settings, creating access problems for rural and remote populations, although telemedicine is increasingly used to ease this issue. There is evidence that these services are experiencing heavy demand with increasing wait times for services, and there are concerns around the capacity of the genetic specialist workforce.[10–12] There is a strong case for the provision of certain genomic tests to move into the primary health setting with support from specialist services.[13]

Through the work of the Australian Genomics research collaboration[2] and state-based genomic programmes, non-genetic specialists in secondary and primary health settings are increasingly interested in genomic applications. They showed that these health professionals can be supported to undertake genomic testing appropriate to their patient cohorts.[14] In primary care, uptake of publicly funded genomics has largely, and only recently, been centred on reproductive genetic carrier screening (RGCS). RGCS is offered to people thinking of starting a family to provide information about carrier status for certain single-gene conditions.[3] Some valuable resources for the offer of tests like RGCS in primary care have been developed but in a piecemeal way. This has led to inconsistent language and criteria used in genetic test directories developed by different pathology services or peak professional bodies.[15 16] In this fast-changing area, information can also rapidly become out of date and risk confusion for practitioners and patients alike.

The majority of genetic and genomic tests available for a general practitioner (GP) (family physician or primary care provider) to order are usually consumer-funded (commercial). A key exception is the three-gene RGCS for cystic fibrosis, spinal muscular atrophy and fragile X syndrome, which is supported by Australia's universal health insurance scheme (Medicare). A second example of a funded test is for people who are diagnosed as having familial hypercholesterolaemia (FH). In this instance, Medicare rebates are available to GPs for cascade testing (ie, testing of first-degree relatives). Other genetic tests that are available in primary care settings, notably RGCS, which uses a larger screening panel, must be paid for by consumers. The growing number of these commercially available tests, offering a bewildering array of gene panels, applications and quality criteria, adds to the complexity for GPs and consumers. Additionally, some tests are actively marketed to GPs, while others are not.

Implementation science

Implementation science uses evidence-based interventions to identify and address barriers to the adoption of new practices (here, the use of genomic testing in primary healthcare), helping support their feasibility, acceptability and appropriateness for all stakeholders.[17] Implementation science interventions also maximise the reach and penetration of a new practice by disseminating a well-designed package of care that is easy to access and use. Implementation science draws on the expertise and experience of multiple stakeholders to build a clear picture of barriers, then uses theoretically informed frameworks to address each one (eg, Consolidated Framework for Implementation Research).[18] Our project design applies implementation science methodology, evidence and concepts to support the use of genomics in primary health.

Genomics in primary healthcare

Although there are over 31 000 GPs working across Australia, configured into 31 primary health networks (PHNs: figure 1),[19 20] many GPs report having limited experience in providing genetic or genomic tests.[21] Government funding for some tests allows for more equitable access to testing, where previously these consumer-funded tests were predominantly taken up by people in higher sociodemographic areas.[21] There is a need to move away from ad hoc management by individual practitioners using varied consumer-funded commercial tests (especially for RGCS) to nationally consistent, equitable care pathways that will ensure high-quality, ethically defensible care is provided for consumers and their families.

Previous work

One of the most significant genomics applications trialled in general practice in the last 5 years was the Australian Reproductive Genetic Carrier Screening Project, also known as Mackenzie's Mission.[22 23] In that research project, GPs and other relevant healthcare providers were supported to offer RGCS to reproductive couples pre-pregnancy or early pregnancy. The comprehensive gene list of approximately 1300 genes (associated with ~750 conditions) provided reproductive couples with information about their combined chance of having children with a serious or life-threatening condition.[23 24] The Mackenzie's Mission project established the clinical utility, cost-effectiveness, feasibility, appropriateness and ethical acceptability of the programme offered predominantly through primary health services.[22] Health professionals (n=990)—mostly GPs—recruited 9107 reproductive couples who underwent screening, including 2378 from culturally, ethnically or linguistically diverse (CEALD) backgrounds or who were Aboriginal or Torres Strait Islander.[23] Detailed testing pathways, laboratory protocols and a national variant review panel were developed to ensure high-quality testing and consistent curation of results across states.[22] The Mackenzie's Mission programme developed a comprehensive support platform for primary health practitioners offering the

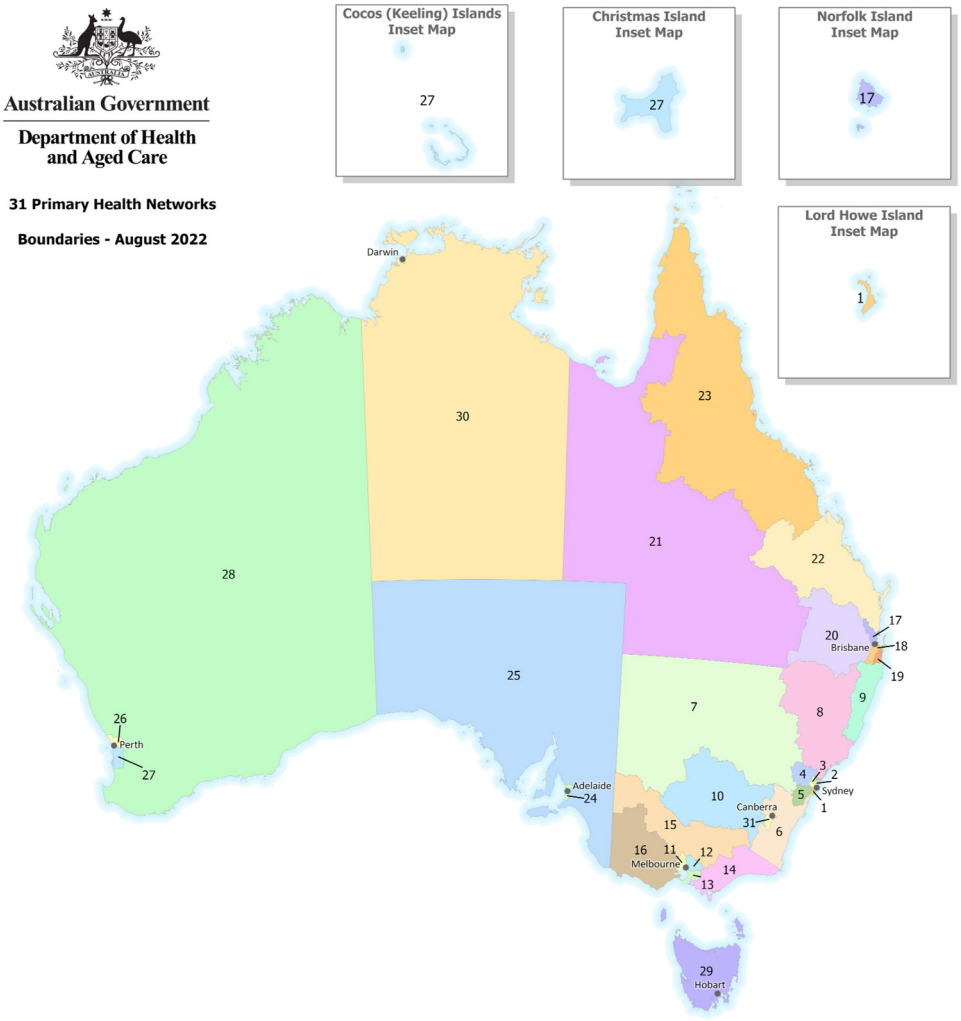

Figure 1 :31 Primary health networks around Australia. Image obtained from the Australian Government, Department of Health and Aged Care (August 2022 version).[20]

test, including flexible education[25] and support from genetic counsellors.[22]

The Mackenzie's Mission investigator team (which included investigators in the present project) produced a large body of work around the barriers, enablers and acceptability of RGCS. Barriers for GPs offering genomic screening and testing included uncertainty about which tests are appropriate, time taken to explain genetic concepts and manage expectations (especially to those patients with limited health literacy or from a CEALD background), ability to facilitate communication between family members, ability to manage a clinically actionable result[26–28] and the risk of ethical tensions arising (eg, concern over allowing enough time in the consultation for patients to make meaningful decisions about the test, in line with their values),[29] which could lead to health professionals developing moral distress.[30] Enablers identified by Australian GPs included clear, frequently updated guidelines on available genomic tests (including decision support algorithms), consumer-facing resources to adequately explain genomic concepts and limitations of the test, support for returning results and access to genetic specialist advice to discuss questions and issues that arise.[26 28 29 31]

The completion of the Mackenzie's Mission project left a gap in the practice of genomics in Australia. A very real risk was identified of GPs losing their skills and their intention to continue offering screening, as research funding for the test was no longer available. The only alternatives at the time were consumer-funded commercial tests. In addition, barriers that had been identified had not fully been addressed, and opportunities to spread the learnings of the participating GPs to their colleagues were potentially being lost.

The embedding genomics in primary care research programme

The embedding genomics in primary care research programme is addressing the gap in support for genomics in primary care. It has the overarching aim of delivering a nationally consistent, ethically defensible approach to supporting and embedding the use of genomics in primary healthcare. Leveraging data, tools, systems and partnerships our team developed in the Australian

Genomics and Mackenzie's Mission projects, we will build, evaluate and integrate a robust suite of tools to support GPs and consumers.

Two distinct types of gene-based application will form the focus of the project: (1) reproductive carrier screening, focusing on the Medicare-funded three-condition RGCS for cystic fibrosis, fragile X syndrome and spinal muscular atrophy (referred to hereafter as the 'three-condition RGCS') (as an example of reproductive screening for X-linked and autosomal recessive conditions) and (2) condition-specific testing, using FH testing (as an example of a dominant trait, condition-specific test), which can be complex to understand and explain to consumers and consistently has implications beyond the individual having the test, to their families. Both applications are recognised as the vanguard for complex genomic interventions that will be managed in the primary health sector. This project will tailor implementation strategies to facilitate high-quality integration of these tests, enabling them to be readily adaptable and scalable as additional tests become available. The project will build a structured, sustainable and easily updatable suite of tools to support the offering of these and future genomic applications in primary care.

METHODS AND ANALYSIS

The study uses a mixed-methods design to develop, implement, disseminate and evaluate a suite of high-quality resources. The work will be run in three parallel and interconnected work packages over 2024–2026. Work Package 1 focuses on supports for GPs, Work Package 2 on supports for consumers, and Work Package 3 examines real-world utilisation patterns and equity of access to genetic testing in primary health settings. This protocol paper outlines work for all the work packages.

Ethics and dissemination plan

Ethical approval for this programme of work has been received from the Macquarie University Human Research Ethics Committee (Ref: 520241849560183) and the Royal Children's Hospital Research Ethics and Governance (HREC/112451). Data management plans were approved to store raw data from focus groups and interviews on the university's and Murdoch Children's Research Institute's secure Teams platforms. Consent will be obtained from all participants for their de-identified data to be used for this project. Dissemination will be via international and national conferences and via peer-reviewed articles.

The Person Level Integrated Data Asset (PLIDA) operates under a privacy-by-design approach and the Five Safes framework. Data linkage is performed by the Australian Bureau of Statistics (ABS), and linkage identifiers are stored separately from analytical information and analysts do not have access to identifying details alongside analytical variables. Researchers are given access to anonymised data sets (restricted to the requested modules) within a secure cloud environment (DataLab). Prior to access, all researchers must complete DataLab Safe Researcher training. All actions taken by researchers are controlled and monitored by ABS. All outputs are subject to ABS disclosure control and must be submitted for ABS DataLab output clearance before they can be accessed outside the DataLab. Data access to PLIDA data is granted only for approved projects, which are reviewed by both ABS and relevant data custodians.

Patient and public involvement

Consumers were an integral part of designing this protocol and were part of the core research and design team from the beginning. Partners on this project included key representatives from consumer support agencies such as SMA Australia, Cystic Fibrosis Australia, Rare Voices Australia, Fragile X Association of Australia and Genetic Support Network of Victoria. Consumers are valued participants across all the work packages.

Overview of work packages

Work Package 1 will develop, tailor and implement key supports for GPs, focusing on five aims (described in more detail below) that will operationalise evidence-based enablers of genomic practice in primary care: (Aim 1.1) establish a Genomics in General Practice Advisory Group to guide, review and provide feedback on the work; (Aim 1.2) investigate and enhance a set of primary health genetic point-of-care resources; (Aim 1.3) collect qualitative data to explore perspectives of GPs and genetic testing laboratories; (Aim 1.4) develop and implement a guide written by bioethics researchers, *Navigating the ethical complexity of genetic testing in general practice*; and (Aim 1.5) establish a national approach for supporting practitioners providing genetic testing. An implementation and evaluation plan for these varied resources will be designed.

The aim of Work Package 2 is to develop and evaluate RGCS consumer resources and plan their implementation strategies, focusing on four aims (described in more detail below): (Aim 2.1) establish a consumer advisory group to guide decision-making; (Aim 2.2) develop RGCS resources, consisting of educational materials and a decision aid; (Aim 2.3) evaluate the resources with potential consumers; (Aim 2.4) design a fit-for-purpose implementation strategy for embedding RGCS resources into primary care.

Work Package 3 will assess equity in access to the three-condition RGCS following its Medicare listing. This work package will use linked administrative and socio-demographic data to examine patterns of RGCS uptake across population groups and identify socioeconomic, demographic and geographic inequities in utilisation. Findings from this work package will inform strategies to support equitable access to genomic screening in primary healthcare.

Work Package 1: development, implementation, dissemination and evaluation of supports for GPs

Backed up by evidence from our body of work with GPs offering RGCS in the Mackenzie's Mission project,[25 26 28 29 31]

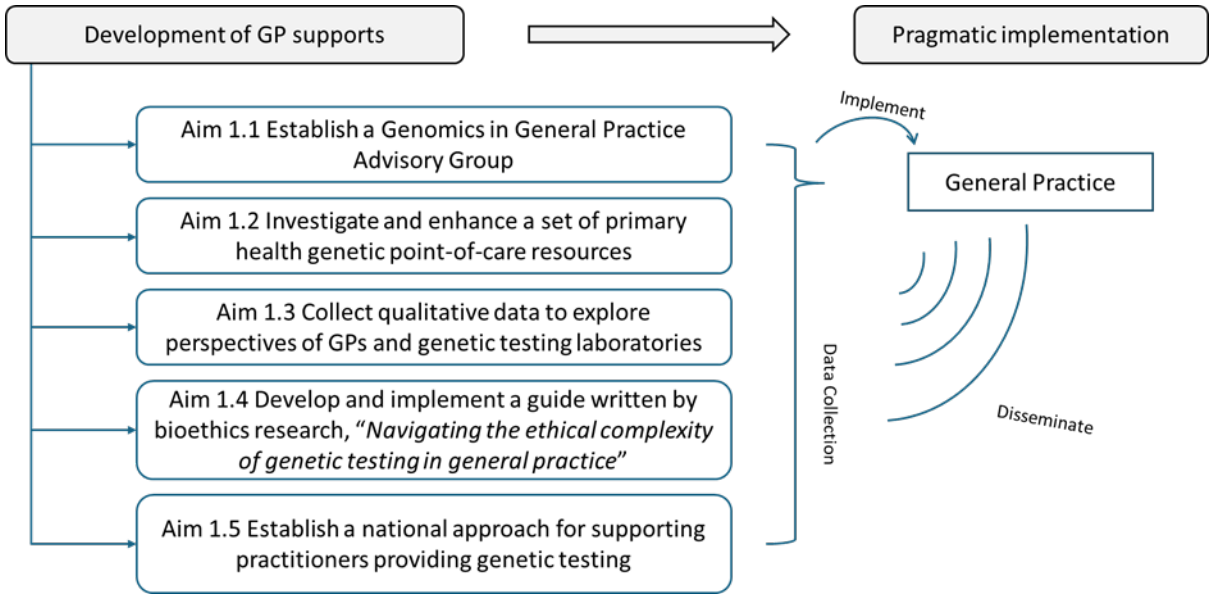

Figure 2 Work Package 1 aims.

this work package addresses known barriers to implementation around knowledge and skills and incorporates identified enablers.[22 26] To bring this work package up to date, we will take into consideration the current context for GPs offering genomic testing to identify other appropriate support strategies. All resources will be overseen by our interdisciplinary research team using co-design principles with a diverse panel of GPs, consumers and other stakeholders to maximise dissemination and adoption of resources (see figure 2).

Aim 1.1: establish a genomics in general practice advisory group (GGPAG)

Common challenges in implementation science include how to make new resources align with existing workflows and be acceptable to users, easily accessible and fit for purpose. A virtual panel comprising GPs and implementation scientists will be convened, representative of different states and regions of Australia (metro, regional and rural)—the GGPAG. A mix of doctors with experience in offering testing and those with limited experience will be sought. We will draw on our existing extensive partner networks to recruit appropriate participants for this group. While membership will be voluntary, members will be remunerated for their time. The GGPAG will work with the research team throughout the project to advise and provide feedback on all draft resources developed in this project.

The purpose of the GGPAG is to ensure pragmatic issues are identified and resolved before implementation. This will maximise the extent to which Work Package 1 supports GPs: (1) are known to GPs, (2) are easily accessible (eg, embedding links on lab order forms, pop-up reminders on general practice software) and (3) are feasible to use (eg, aligned with routine GP workflows).

Aim 1.2: develop and implement a set of primary health genetic point-of-care resources

The needs of GPs identified in Mackenzie's Mission, along with resource gaps identified through GGPAG discussions, peer-reviewed and grey literature reviews and consultations with the embedding genomics in primary care expert network, emphasise a key missing element of support for genomics-related decision-making in general practice: primary care-framed expert summaries easily accessible during consultations. GPs require concise overviews of:

- Relevant genetic tests and clinical guidelines.
- Reasons people may choose to have or not have genomic tests.
- Guidance on the interpretation of results.
- Associated management and referral pathways.

Since GPs use a variety of practice-based software and clinical support tools, several different approaches will be taken to the development and provision of these resources.

A well-established general practice resource, HealthPathways,[32] will be leveraged to disseminate the primary care provider-framed expert summaries. HealthPathways is a suite of region-specific, evidence-based information pathways for assessment, management and referral, disseminated through Australian PHNs, local health districts and healthcare providers.[32] Each PHN oversees the continuous development, review and updating of HealthPathways content. Additionally, HealthPathways programmes across Australia share expertise and resources to enhance pathway development and maintain nationally framed best practice. By collaborating with HealthPathways' clinical content editors, we will develop relevant pathways for genetic screening and testing or work with clinical editors to revise existing pathways in line with current genomics guidelines.

We expect that the evaluation of HealthPathways in a future follow-up project will involve both quantitative measures, such as usage tracking, and qualitative feedback from surveys and interviews to assess its impact on healthcare delivery, adoption and practical application. This multifaceted approach will ensure the resource effectively supports genomics decision-making in general practice.

Clinical support tool providers will be engaged in discussions to explore additional options for genomic support for GPs. We will consider preconsultation tools such as *Better Consult*, which use patient-reported questionnaires to gather information on patients' needs, symptoms and relevant family and clinical history, supporting proactive diagnosis of inheritable conditions. We will also review AI-based tools such as *TrakGene*, which use specialised genetic software to generate accurate family trees that can be integrated with patients' medical histories to produce genetic risk information. Additionally, we will explore population data tools such as *Primary Sense* and *POLAR*, which are designed to identify trends and disparities across GP practice populations and integrate with electronic health records to support clinical decision-making. These tools may help GP practices to identify patients who meet criteria for genetic testing. We will also appraise the utility for GPs of fact sheets, such as those produced by HealthShare, that are designed to embed in commonly used practice software platforms for ease of access.

The research team will examine how these varied initiatives can be tailored to facilitate genomic testing in general practice. Inclusion will depend on feasibility and alignment with project outcomes. Discussions with providers, in consultation with project partners and GGPAG, will ensure consensus on the relevance to GPs, considering each tool's capability and the grant's parameters. To manage potential vendor influence, tool selection will follow a structured process: (1) systematic review of available tools against pre-defined criteria (evidence base, usability, accessibility and cost); (2) independent evaluation by the GP advisory group; (3) declaration and management of any vendor relationships or conflicts of interest and (4) prioritisation of open-access or publicly funded resources where feasible. All team members will declare any relationships with tool developers or commercial entities.

If we proceed to engaging a provider and developing a primary care-facing clinical support tool, implementation and evaluation will occur initially at a single test site and will assess, in particular, how the tool supports the uptake of the two exemplar genetic tests. Usage of the tool will be monitored through the audit functionality of each proprietorial resource and feedback from GPs at the test clinic will be sought through a short 5-minute survey and qualitative interviews. Interview feedback will be analysed using an implementation science framework, the Behaviour Change Wheel,[33] employing double independent coding. Results will inform refinements to be incorporated into the final version of the tool. Wider dissemination of the selected tool would be facilitated by communication through our GP team's network.

Aim 1.3: explore perspectives of GPs and genetic testing laboratories

Recent innovations in laboratory service models for genetic testing have significant implications for GPs, with laboratories publishing detailed physician-facing summaries of genetic testing results[16] and, in some cases, providing genetic counselling services before or after testing. There is no Australian research on GPs' experiences of interfaces with genetics laboratories and how this impacts genomics decision-making in primary care. Additionally, there is a gap in Australian data on GPs' perspectives regarding these resources and how they influence GPs' roles in supporting patients and coordinating care with specialists over time. However, qualitative data suggest that genetic counselling support offered by laboratories improves GP confidence in offering RGCS.[34] Understanding the perspectives of GPs and testing laboratories will contextualise the new landscape of genetic testing services and address the gap in literature concerning the practical feasibility and integration of laboratory-driven resources and genetic counselling services within Australian primary care.

To address Aim 1.3, we will interview Australian GPs with experience or interest in genetic testing and representatives from Australian laboratories offering reproductive carrier screening and/or FH testing services, particularly those involved in GP support for genomics testing. We will seek the perspectives of approximately 15–20 GPs and 5–10 laboratory representatives, with sampling continuing until no new themes are emerging. Recruitment will involve targeted email invitations through professional networks and medical associations, with stratified sampling to ensure diversity across practice settings. Data will be collected via an online video call platform that will be recorded and transcribed. Thematic analysis, using NVivo software, will be both inductive and deductive, and regular team discussions will ensure consistency in coding. Findings will be presented with illustrative quotes and disseminated through professional journals, conferences, and steering committee meetings. The results may also inform the development and refinement of point-of-care tools for GPs.

Aim 1.4: develop, implement and evaluate 'navigating the ethical complexity of genetic testing in general practice' guide

Building on our extant genetic specialist experience and the research we undertook in Mackenzie's Mission,[26 29 35] and other research on ethics and familial communication of genetic information,[36–39] this resource will address ethical issues either flagged by GPs as problematic or identified as ethically complex in wider bioethics literature. Recognising the risk of moral distress from unresolved ethical issues, especially those that can lead to moral injury[40] in health professionals, this guide will give GPs confidence in offering genetic testing to all consumers

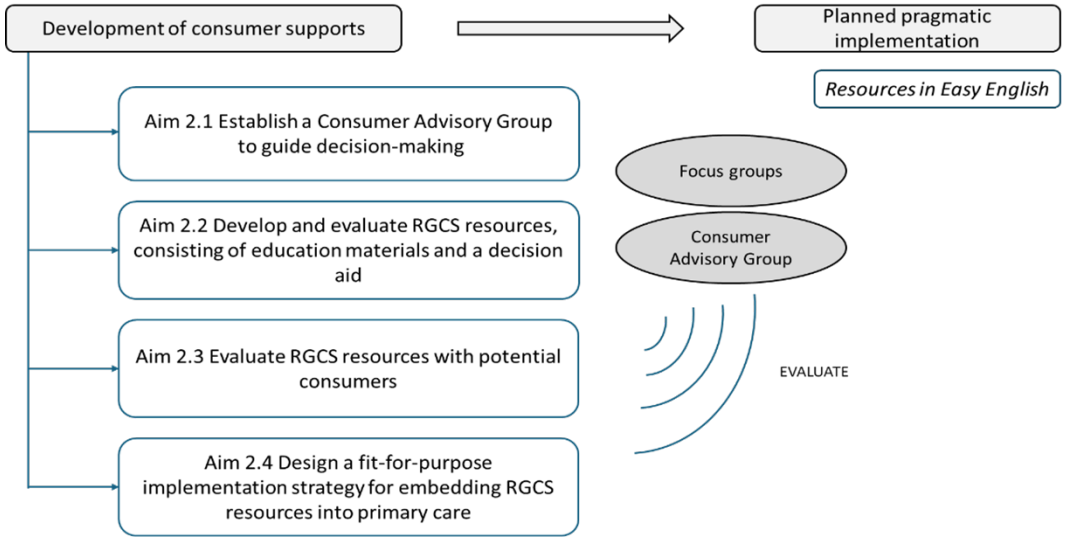

Figure 3 Work Package 2 aims.

and responding to queries or concerns. It will include ethical issues in genetic testing, suggested approaches to offering particular tests, such as carrier screening or diagnosis of conditions such as familial hypercholesterolaemia, managing potential familial non-disclosure of genetic information (relevant to conditions like familial hypercholesterolaemia) and approaches to answer or support consumer concerns. Led by our experienced bioethicists, this resource will also be reviewed by the GGPAG, disseminated, implemented, and evaluated by survey and interview after 12 months. We expect that a similar evaluation strategy to the point of care tools (Aim 1.2) evaluation will be employed in a future project, that is, a 5-minute survey containing an invitation to an interview, widely disseminated throughout the PHN partners.

Aim 1.5: establish a national approach for supporting practitioners providing genetic testing

One of the significant enablers identified by GPs taking part in Mackenzie's Mission was the one-to-one support given by study genetic counsellors.[26 41] This increased GPs' confidence in offering RGCS, answering questions such as the logistics of the test, the appropriate offering of screening, responding to consumer questions and support for when results were returned. This model has also been used successfully by Victorian Clinical Genetics Services in delivering reproductive genetic testing services, including non-invasive prenatal testing and RGCS.[3] This model uses genetic counsellors to support healthcare providers offering RGCS. We will draw on input from the GGPAG as well as interviews with GPs to understand the advice needs of GPs regarding genetic testing and establish recommendations for a national approach to support GPs providing genetic testing.

Work Package 2: development and evaluation of consumer resources and design of the implementation strategies

This work package aims to generate evidence-based resources (eg, information sources and a decision aid) to empower the public to discuss RGCS with their GP, obstetrician or other healthcare provider. These resources will support people considering RGCS to make informed decisions. A mixed-methods approach, including focus groups[42] and interviews with potential consumers and co-design with the advisory groups and partners, will be adopted (figure 3).

Consumer supports for reproductive genetic carrier screening

Aim 2.1: establish a consumer advisory group to guide decisions

We will engage a consumer advisory group for this project. This will include relevant community representatives (eg, support organisations for people with screened conditions, patient advocates, people with lived experience of RGCS and researchers with an interest in this topic). Members of the research team have established relationships with many of these stakeholders through clinical and research-related activities. CAG members will be asked to recommend other relevant individuals for invitation. Members may leave the group at any stage, and if other relevant stakeholders/groups are identified during the project timeline, they may also be invited. The CAG will agree on terms of reference, and members will be offered compensation for their involvement, including time to prepare for meetings (reviewing documents) and attend meetings, in the manner preferred by individuals. Members will be paid rates consistent with the specific rates for remuneration listed in the Australian Genomics community member honorarium and reimbursement policy. The CAG will be provided with timely communication and clear information regarding each stage of resource development and evaluation.

Aim 2.2: develop the consumer resource

The Mackenzie's Mission project provided plain English information on genetics and how carrier screening tests work, with materials also translated into Arabic and simplified Chinese. It also included a decision aid[25] designed to facilitate couples' informed decision-making. The existing Mackenzie's Mission decision aid,[25] initially developed through a Delphi study involving expert reviews and iterative content adjustments, was designed for use in a controlled research setting. Although its validity was supported through think-aloud interviews and qualitative feedback, it has not yet been evaluated beyond the research project or tested in public or primary healthcare contexts.

In this study, we will adapt the MM informational resource and decision aid for broader public use, particularly in primary healthcare settings. The adaptation will draw on:

1. Concepts about consumer test acceptance, decision-making, educational and information needs and experiential knowledge identified in a scoping review. Here we will collate and analyse what is known globally about consumer preferences about RGCS, identify gaps and make recommendations for the development and fit-for-purpose service implementation.
2. Expert advice from the CAG and the GGPAG formed in Work Package 1. Here, we will seek guidance on recommendations for resource design and implementation, prototypes and study design.
3. Outcomes from qualitative interviews and focus groups exploring the views and preferences of potential consumers regarding the content and delivery of RGCS resources.

Recruitment will involve advertisements through university and MCRI bulletins and social media platforms, aiming for diverse participation across different regions and demographics. We will use maximum variation sampling and participants will be aged 18–50 years, living in Australia and may have undergone, be considering or consider RGCS in the future, with English proficiency or interpretation support. The sample size is guided by the concept of information power. Information power considers factors such as the study aim, sample specificity, use of theory, quality of dialogue and analysis strategy at various stages of the research. Data collection will continue until no new information relevant to the research question emerges from the focus groups or interviews. Based on these principles, we anticipate this will occur after approximately 30–40 participants. For a qualitative study, this represents a small to medium sample size, justified by the considerations outlined above and informed by the research team's experience conducting and reviewing similar studies. The sample size will be reassessed throughout the study as data are collected and analysed, in line with information power guidelines.

The qualitative interviews and focus groups will be analysed using inductive content analysis to identify themes and categories emerging from the data.

Aim 2.3: evaluate the consumer resource

We expect that the adapted resource will be developed as a website using the URL carrierscreening.org.au. This may include lab-agnostic information in the form of videos, audio recordings, fact sheets and a decision aid to support individuals and couples to make informed decisions about RGCS.

We will evaluate the resource on its website format using think-aloud interviews with potential consumers. These interviews will be conducted using a hypothetical RGCS model ('imagine you were getting screened, would this') and will focus on the following content areas below, as well as any other topics identified as important in Aims 2.1–2.2 above:

- Provides sufficient and digestible information.
- Addresses key factors involved in decision-making.
- Incorporates perspectives beyond biomedical considerations (eg, religiosity and cultural factors).
- Meets practical and implementation needs (eg, involving partners; fitting within GP-guided pathways).
- Demonstrates inclusive design and functionality (widely accessible, understandable and translatable).
- Is future-proofed to accommodate emerging developments in RGCS (eg, direct-to-consumer delivery, larger gene panels).

Participants will be recruited through the same methods as Aim 2.1–2.2 above. The sample size is based on previous research that developed the decision aid for the MM study.[25]

Data will be analysed using inductive content analysis.

Aim 2.4: design implementation strategies for the consumer resource

The implementation setting for resources in primary care is highly complex. Consumers may undertake RGCS through sequential testing (with the female partner screened initially) or through simultaneous testing of both partners for a couple-level risk estimate. Test panels vary widely, from targeted assays to extensive panels analysing hundreds to thousands of genes. RGCS can be accessed in primary care, via specialist clinicians, or through direct-to-consumer services. We will draw on outcomes from the review, interviews, CAG, GGPAG and partners to suggest appropriate timing and placements for different lab-agnostic resources and when and how these can integrate with the experience of RGCS being supported in primary healthcare. To ensure that the resource reaches a diverse audience, we plan (pending funding) to conduct awareness-raising campaigns. Based on further stakeholder consultation, we will determine whether a national campaign is sufficient or if targeted campaigns and outreach interventions, specifically designed to engage and educate priority populations, are required. These interventions may need to be co-designed in partnership with communities to ensure cultural relevance, acceptability and effectiveness.

Work Package 3: assessing equity in access to three-condition RGCS in primary health settings

Work Package 3 will examine equity in access to three-condition RGCS following their Medicare listing, using linked, person-level administrative and sociodemographic data. This work package will focus on identifying socioeconomic, demographic and geographic gradients in RGCS uptake and assessing whether publicly funded screening is equitably accessed across the Australian population. The analysis will build on prior evidence demonstrating socioeconomic disparities in reproductive carrier screening and will provide contemporary, population-level evidence using real-world data. This work package will use a large, linked Australian government administration data asset known as PLIDA. It has linked Medicare Benefits Schedule (MBS) data on healthcare service utilisation, Pharmaceutical Benefits Scheme data on medication use, and individual-level socioeconomic characteristics derived from census and national survey sources.[43] By leveraging this linked data asset, the analysis will move beyond area-level descriptions to a more granular assessment of equity in RGCS utilisation across population subgroups.

Aim 3.1: assess socioeconomic and demographic inequities in RGCS uptake

We will investigate the association of socioeconomic and demographic factors with the uptake of reproductive carrier screening in Australia. This will build on Robson *et al*,[21] which reported a pronounced socioeconomic gradient, showing that women in the most advantaged postcodes were significantly more likely to access reproductive genetic carrier screening compared with those in disadvantaged areas. Using much more detailed PLIDA data, our RGCS uptake will be examined across individual- and area-level indicators of socioeconomic position, including education, household income, employment status and geographic disadvantage. It will also be evaluated across time to determine whether inequity has changed.

For the purposes of this analysis, uptake of RGCS will be defined as the presence of a relevant MBS claim for three-condition RGCS at the individual level. The primary unit of analysis will be the individual, with uptake expressed relative to the reproductive-age population in the relevant geographic area and time period, using population denominators derived from ABS population estimates. Where appropriate, analyses will also be presented as rates per population (eg, tests per 1000 reproductive-age individuals).

Given the limitations of administrative data, testing pathways (including sequential vs simultaneous testing of partners) cannot be consistently distinguished and will not be modelled separately. Instead, all valid MBS claims for RGCS will be treated as indicators of uptake, consistent with the aim of assessing population-level utilisation and equity.

Multivariable regression models will be used to assess associations between RGCS utilisation and socioeconomic characteristics, adjusting for relevant demographic and geographic factors. Primary analyses will use mixed-effects logistic regression models at the individual level or area-level count models (such as Poisson or negative binomial regression of tests per reproductive-age population), depending on data structure and availability. Models will account for clustering within geographic areas, such as postcodes or PHNs, through random effects or robust variance estimation.[44 45]

Core covariates will include age, sex, rurality or remoteness, state or territory and the distribution of reproductive-age women. Additional covariates will include education, household income, indicators of culturally and linguistically diverse populations and Aboriginal and Torres Strait Islander status. Analyses will assess gradients in RGCS uptake across socioeconomic measures, with primary contrasts specified a priori (eg, comparisons between the most and least disadvantaged socioeconomic groups).

The findings from this work package will provide rigorous, policy-relevant, detailed evidence on whether Medicare-funded RGCS is being accessed equitably across the population and will identify groups with persistently lower uptake and what individual and geographical characteristics are associated with this low uptake. These insights have the potential to inform the development of targeted policy to improve equitable access to genomic screening in primary healthcare, using three-condition RGCS as an exemplar for future genomic testing programmes.

DISCUSSION

This project aims to deliver a nationally consistent, ethically defensible approach to support and embed the use of genomics in primary healthcare in Australia. Leveraging data, tools, systems and partnerships our team developed via Australian Genomics and the Australian Reproductive Carrier Screening project (Mackenzie's Mission), we will build, evaluate and integrate a robust suite of tools to support both GPs and consumers using genomic applications, ensuring that the benefits of genomics for human health are realised.

The two tests that are the focus of this work represent two key types of genomic testing that will be used in primary care in the future and build a foundation on which new, more complex or comprehensive programmes can be managed. By embedding a robust understanding and clear process for the three gene RGCS for X-linked and autosomal recessive conditions, more complicated expanded carrier screening tests for couples that involve hundreds of genes can be understood and managed. Likewise, cascade testing for the autosomal dominant condition FH equips GPs for the process of cascade testing for other conditions.

Ethical considerations are an important part of this project. Key to the adoption of new genomic tests in

primary care is the assurance that they are ethically defensible and appropriately offered. The *Navigating the ethical complexity of genetic testing in general practice* resource will identify and articulate some of the complex ethical issues that can arise in the use of genomics in primary care settings. For example, a test result can have significant consequences, including relevance to reproductive choices and impacts on other family members, and the issues can vary according to individual family dynamics. Understanding the ethical complexity as relevant to the primary care context will mitigate the risk of moral distress to GPs.

This project is strategically designed to take a national approach to the use of genomics in primary care. While each jurisdiction in the federation of Australia governs and funds its own genetic services, Australian Genomics and Mackenzie's Mission have amply demonstrated the power of a consolidated and collaborative approach in genomic research and practice.[2 8] Our research team, partners and participants are from across Australia so we can standardise best practice and benefit from economies of scale: for example, one RGCS decision aid for all of Australia, not eight individually developed ones for each state or territory. Standardising methods across regions and aligning with government interests provides a comprehensive view that also aids in policy formulation. The national integration captures broader impacts, ensures uniformity, considers budget implications and aligns with accepted practices. This approach enables a more robust evaluation of the intervention, considering the diversity and complexity of Australia's healthcare system.

A national approach is also appropriate for assessing access to genomic testing in primary healthcare from an equity perspective. The evaluation component will provide key insights into patterns of utilisation of a representative genetic test, while the equity analysis will determine whether Medicare listing has led to a more balanced uptake across socioeconomic groups, ensuring that these benefits are shared equitably.

The research team has been carefully selected to bring together expertise from different disciplines. We have a strong collaboration of consumers, GPs, genetic counsellors and clinical geneticists, laboratory scientists, bioethicists, health economists and implementation scientists. We also bring key partner organisations to the table, sharing their unique expertise and extensive member networks: MQ Health General Practice Clinic, Human Genetics Society of Australasia, Australian Genomics, Royal College of Pathologists Australia and consumer advocacy agencies Rare Voices Australia, Fragile X Association of Australia, Cystic Fibrosis Australia and the Genetic Support Network of Victoria.

Our project illustrates the opportunities that exist after a research intervention trial has finished to continue the momentum and embed new practices into usual care. Many projects assume that publication of findings at the end of the funded work will automatically result in adoption. Such passive, 'paper implementations'[46] rarely succeed, as they do not take into consideration the attitudes of users, contextual constraints, current workflows or historical issues that can easily derail new processes. Our intentional implementation work, underpinned by implementation science methods, will diagnose individual and system-level barriers to change, checking for key outcomes of acceptability, appropriateness, accessibility and feasibility of all the resources developed.

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