BMJ Open

Development of a person-centred digital platform for the long-term support of people living with an adult-onset genetic disease predisposition: a mixed-methods study protocol

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To cite: Best S, Al Mahmud A, Tyagi S, *et al*. Development of a person-centred digital platform for the long-term support of people living with an adult-onset genetic disease predisposition: a mixed-methods study protocol. *BMJ Open* 2023;**13**:e071492. doi:10.1136/bmjopen-2022-071492

► Prepublication history and additional supplemental material for this paper are available online. To view these files, please visit the journal online (http://dx.doi.org/10.1136/bmjopen-2022-071492).

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Received 10 January 2023
Accepted 14 July 2023

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ABSTRACT

Introduction Individuals at an inherited high-risk of developing adult-onset disease, such as breast cancer, are rare in the population. These individuals require lifelong clinical, psychological and reproductive assistance. After a positive germline test result, clinical genetic services provide support and care coordination. However, ongoing systematic clinical follow-up programmes are uncommon. Digital health solutions offer efficient and sustainable ways to deliver affordable and equitable care. This paper outlines the codesign and development of a digital health platform to facilitate long-term clinical and psychological care, and foster self-efficacy in individuals with a genetic disease predisposition.

Methods and analysis We adopt a mixed-methods approach for data gathering and analysis. Data collection is in two phases. In phase 1, 300 individuals with a high-risk genetic predisposition to adult disease will undertake an online survey to assess their use of digital health applications (apps). In phase 2, we will conduct focus groups with 40 individuals with a genetic predisposition to cardiac or cancer syndromes, and 30 clinicians from diverse specialities involved in their care. These focus groups will inform the platform's content, functionality and user interface design, as well as identify the barriers and enablers to the adoption and retention of the platform by all endusers. The focus groups will be audiorecorded and transcribed, and thematic and content data analysis will be undertaken by adopting the Unified Theory of Acceptance and Use of Technology. Descriptive statistics will be calculated from the survey data. Phase 3 will identify the core skillsets for a novel digital health coordinator role. Outcomes from phases 1 and 2 will inform development of the digital platform, which will be user-tested and optimised in phase 4.

Ethics and dissemination This study was approved by the Peter MacCallum Human Research Ethics Committee (HREC/88892/PMCC). Results will be disseminated in academic forums, peer-reviewed publications and used to optimise clinical care.

STRENGTHS AND LIMITATIONS OF THIS STUDY

⇒ Development of a novel digital platform to embed easy access to clinical genetic expertise into the long-term risk management and care for people living with a high-risk genetic disease predisposition.
⇒ Use of a codesign approach integrating behavioural theory to optimise uptake and use of the digital platform by patients, and to empower self-efficacy and improve health outcomes.
⇒ Explore, with clinical stakeholders, potential strategies by which the digital tool could be integrated into current clinical practice with maximum efficiency.
⇒ The use of online tools, emails and surveys may bias recruitment towards individuals who have high digital access and literacy.
⇒ Exclusion of non-English speakers from the study is a major limitation, reducing applicability and useability of the digital platform by culturally and linguistically diverse groups. This will be actively addressed in future work.

INTRODUCTION

Over the last 21 years,[1] technological advances and efficiencies in DNA sequencing have driven an unprecedented expansion in a genome-first approach to precision medicine. No longer limited to single gene analysis to identify high-risk pathogenic variants (PVs) which explain a family disease history, genomic testing has established DNA variation as an important contributor to rare as well as common disease diagnosis, prediction, prognostication and treatment decisions.[2] These findings underpin the establishment of large longitudinal cohort research studies, assessing clinical outcomes in the presence of genome sequencing, biomedical assessment, survey and electronic medical record

data. The most notable of these is the National Institutes of Health's 'All of Us' research initiative, which aims to recruit 1 million Americans.[3] This study will return high-risk PVs in 59 genes related to diseases for which there are proven early treatment or preventative measures as determined by American College of Medical Genetics.[4] They anticipate returning 30 000 high-risk genetic PV results. Similarly, the 100 000 genomes project led by Genomic England is returning high-risk genetic secondary findings to study participants.[5] Smaller cohort and population testing studies reporting results back to participants are increasing worldwide.[6]

With escalation in genomic testing and return of 'actionable' findings occurring across large healthy populations, it is important to consider that the identification of a disease-associated PV is only a surrogate outcome. Key to achieving the improved health outcomes promised by genomic technology is the vital next step, namely supporting equitable, lifelong access to optimal care for all individuals with a known high-risk disease predisposition. To date, developing the prerequisite frameworks to support this critical element of patient care has received less attention.

The lived experience of individuals with high-risk PVs mirrors those living with a chronic disease. They report a lifelong need for psychological, reproductive and family communication support alongside multidisciplinary clinical care programmes,[7–10] which can change and evolve over their lifetime. There are the additional implications that risk management strategies can impact physical and psychological quality of life, and genetic conditions are highly transmissible to future generations. Many genetic conditions are associated with young-onset disease, and the premature death of parents and close relatives who would otherwise provide vital emotional and practical support.

In many countries such as Australia, specialist clinical genetic services (CGSs) provide psychological support, coordination of care and a lifelong point of contact for individuals at a high genetic disease risk. The largest group of patients are often those with common familial cancer or cardiac syndromes. Formal clinical follow-up programmes which aim to support and ensure equitable access to optimal person-centred clinical care are rare.[11] There are no established clinical frameworks comparable, for example, with national breast cancer screening programmes, that support these clinically vulnerable individuals in accessing care. Patients are often left to navigate the complexity of a healthcare system unaided. This challenge will only escalate as genetic testing becomes increasingly decentralised across multiple non-genetic specialities. Many patients may never encounter a CGS to support care coordination or family communication. Even within CGSs, where funding is finite, the increasing number of individuals with PVs means clinical follow-up programmes are becoming too much of a time, financial and human-resource burden to be maintained in their current formats.

There are limited real-world studies of clinical follow-up programmes[12–14] yet, the few that do exist demonstrate that the coordinated management of high-risk breast, ovarian or colorectal cancer risk is both clinically efficient and cost-effective.[15 16] However, the uptake of risk-appropriate management programmes internationally is suboptimal.[17–19] Barriers to people accessing and following recommended guidelines over the long term are relatively unexplored,[20] although older age and higher physical functioning are linked to adherence.[21] Nevertheless, patients are often ill informed and not following the most up-to-date management guidelines.[22] Consumer advocacy and support groups in the high-risk breast cancer setting[23] endeavour to fill this void but women still call for 'unambiguous, clear and unified information from health professionals' to counteract conflicting advice[24] and limited knowledge[25] from non-genetic specialists.

Fulfilling the promise of genomic medicine demands a move beyond just the completion of genomic tests to the provision of ongoing support for patients and their families to enable them to live with the ongoing impact of their test results. Digital health technologies have the potential to offer efficient and sustainable ways to deliver affordable and equitable care,[26] which can help build ongoing relationships between health professionals and their patients. With regards to genomic medicine, a digital technology that promotes and supports easy communication between health professionals and patients provides a platform for ongoing delivery of care. Such a platform could provide patients with access to reliable curated information from trusted sources and long-term triangulated support from both CGS and their non-genetic care providers, complementing in-person care. For health professionals, it enables them to provide continuity of care, build trust and relationships and monitor patient engagement over a long-term period, in some cases a lifetime, to ensure patients have access to the most current health information and recommendations, and clinical trials. Furthermore, with advances in technologies such as artificial intelligence (AI), digital health platform powered by AI can help identify specific subgroups of patients at key clinically relevant life stages that require clinical care and support CGS teams in delivering optimal care.

Digital health technologies in CGS provision and chronic disease management

With the proliferation of mobile health applications (mHealth apps), potential opportunities to empower patients to self-manage their chronic diseases grow.[27] Such applications (apps) already support management of diabetes,[28] cardiovascular disease[29] and chronic obstructive pulmonary disease.[30] These solutions typically focus on improving ease of communication between patients and multiple healthcare providers, enabling disease management and reducing medication errors through monitoring at home, and supporting self-efficacy.[31] A systematic review of digital health platforms used in long-term cancer care highlighted their role in improving

availability of information, and self-efficacy. However, clinical effectiveness data were absent due to limited rigorous assessment of long-term outcomes.[32 33]

Digital technology is also reshaping genetic counselling practice.[34] It can assist in identifying at-risk patients, providing counselling services and establishing communication with a diverse group of people.[35] For instance, researchers have developed a web-based app called GUÍA to enhance communication with patients and families concerning genomic results and clinical information.[36] Several mobile phone apps and chatbots have been used to enhance and support genetic counselling.[34 37] However, one limitation of note of the platforms developed to date is that they do not provide ongoing personalised bi-directional communication to foster closer, long-term relationships with CGS.[38]

Despite the increasing use of digital health platforms across health services, evidence regarding uptake and particularly retention of these platforms in long-term care is limited. In 2003, Venkatesh *et al* developed the Unified Theory of Acceptance and Use of Technology (UTAUT) based on three determinants of behavioural intention—performance expectancy, effort expectancy and social influence, and facilitating conditions.[39] This theory underlines the importance of developing digital platforms in a systematic and rigorous fashion in consultation with all stakeholders to assess and target stakeholder expectations with minimal effort, and to understand parameters that impact social influence.

The need for such collaboration in developing digital solutions for patient-centred care has been emphasised in recent literature.[40 41] A variety of potential digital tools exist including web-based portals, chatbots and decisions aids. A recent review by Lee *et al*[42] with the Genetics Navigator team identified 70 unique digital tools from the genetic space. Many were for use in oncology (n=34) and were primarily aimed at consumer education (n=59) and/or decision-making about pursuing genetic testing and/or types of results to receive (n=32). While the Genetics Navigator team found that digital tools can improve workflow and reduce the time patients need to spend with genetic services, they also report a shortage of digital tools that support the entire patient trajectory. Our preliminary review of the literature reveals that there is no off-the-shelf digital health platform that can provide ongoing management of care and support to patients with a high-risk genetic predisposition through fostering long-term bi-directional communication with the care providers and genetic specialists.

Aims

This paper provides an outline of the study protocol of a 2-year project whose aim is to *codesign and develop a user-centric digital health platform with functionality elicited from patients, CGS staff and treating specialists to align with their needs and enablers, ensuring an acceptable, interactive and highly usable platform.*

We hypothesise that increasing ease and equity of access to effective care via the use of a digital health platform will empower patient self-efficacy to make evidence-informed decisions aligned with their values, access support to navigate the healthcare system and optimise clinical care and outcomes, both psychological and clinical, for individuals living with high-risk genetic predisposition, and their families.

The remit of this project aligns with the first life cycle phase of the Design and Evaluation of Digital Health Interventions (DEDHI) framework.[43] This framework assigns stage-specific design and evaluation goals over a digital health tool development and evaluation lifecycle. The first-stage evaluation criteria include ease of use, adherence, personalisation, safety, privacy and security, while in tandem assessing implementation barriers, useability and impact on work force. The research team comprises a transdisciplinary mix of specialists in clinical genetics and genetic counselling, computer science and software engineering with a specialist interest in AI, human-computer interaction, human-centred design and implementation science.

Methodology principles

Development of the platform will weight all stakeholders' (patients and clinicians) experiential needs and preferences equally by adopting a codesign, user-centric methodological approach.[44] Preferences and requirements from all key stakeholders will be elicited in the predesign phase, and reassessed through an iterative 'design and evaluate' consultatory process throughout the design and development phases. This approach goes beyond specialist stakeholders developing and then patients *post hoc* assessing the product's 'acceptability', 'feasibility' and 'usability'.[45]

The design process will be guided by UTAUT to match stakeholder expectations and digital literacy to the final platform design. In addition, we will follow the Experience-Based Co-Design approach, which will assist in identifying the gaps in current services and ensuring the prioritised features from patients and clinicians' perspectives are incorporated into the platform.[46]

Integrating digital health technologies into a clinical service has potential to impact current workforce practices.[47] Demarcation of roles and responsibilities across the multidisciplinary CGS team will be explored early through the development of CGS-based digital health coordinator role. This is in keeping with implementation of evaluation processes indicated in phase 1 of DEDHI.[43]

METHODS

The study will use a mixed-methods approach, and data will be gathered in multiple, overlapping phases. An overview of the approach is shown in figure 1.

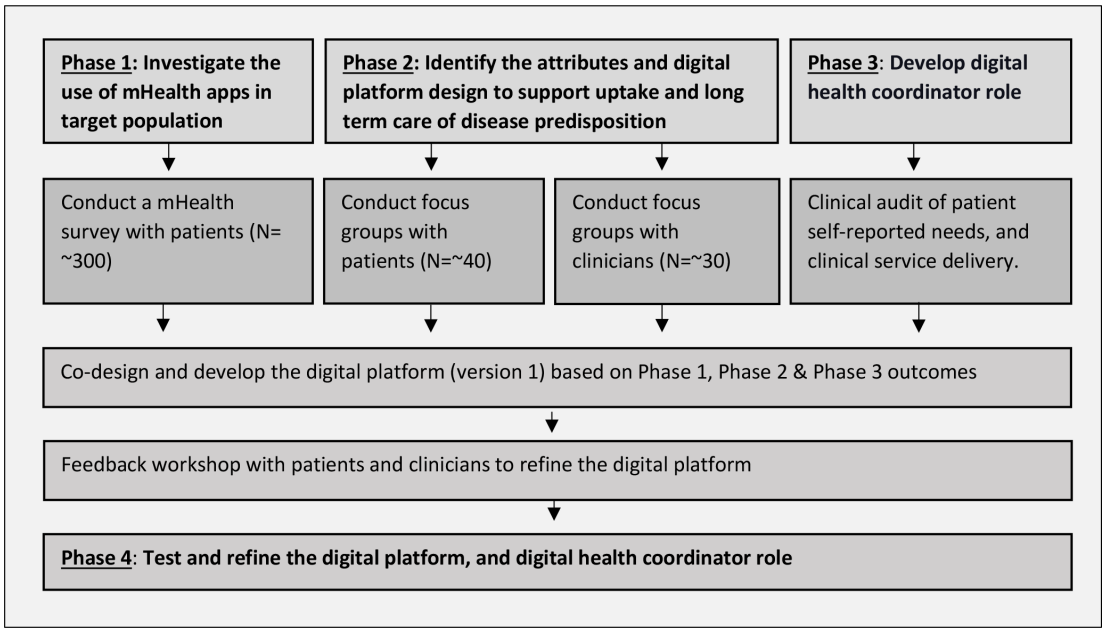

Figure 1 Overview of the platform codesign and development process. mHealth apps, mobile health applications.

Participants and clinical context

The primary stakeholders of the user-centric digital health platform - CGS-associated clinicians and patients with a high-risk genetic predisposition - will be recruited through (1) the Parkville Familial Cancer Centre (PFCC), Peter MacCallum Cancer Centre and The Royal Melbourne Hospital, and (2) the Department of Genomic Medicine, The Royal Melbourne Hospital, Victoria, Australia, respectively. These two integrated adult clinical genomic services are situated in tertiary public-funded hospitals in an urban setting. Both departments accept clinical genetic referrals from across the State of Victoria in Australia and offer multidisciplinary specialist genetic risk assessment and high-risk management clinics. The PFCC also provides an electronic survey-based clinical follow-up programme for people with a high-risk cancer predisposing PV—'The ongoing care programme'.

Stakeholder patient group: Individuals over age 18 years with a high-risk genetic predisposition to either a cancer or cardiac syndrome who have attended either the PFCC or Department of Genomic Medicine. The initial design phase will focus on healthy individuals living with a high genetic risk of disease, but affected individuals may participate in the later prototype assessment stages.

Stakeholder CGS-associated clinician group: Participants will be recruited from the group of multidisciplinary clinicians who specialise in the care and management of patients attending the two recruitment sites. These will include specialists in clinical genetics, genetic counselling, cardiology, oncology, gynaecology and breast surgeons across all stages of career development.

Phase 1

Determine the current landscape of digital health app use by patients (0–6 months).

In phase 1, we will explore how our target patient group uses mobile apps for managing their health conditions. Though it is evident that mHealth apps can assist individuals in managing chronic diseases, it is not known whether or how people with genetic disorders would use mHealth apps for ongoing care. Understanding the use of previous and current use of mHealth apps is vital to the development of a new user-centric digital health platform to support that population and to identify barriers which may impact equitable implementation and use of the platform.[48]

Electronic survey: Current and previous experience of digital health/well-being smartphone apps and types of apps used by patients will be assessed through an online survey developed by the research team drawing from Zhang *et al*[49] and presented in Qualtrics software (online supplemental appendix A).

Three hundred patients from the two recruitment sites will be contacted by email inviting them to fill in and return the epidemiology and mHealth survey. These patients will be identified by categorising eligible patients into 10-year age categories and randomly selecting 300 from across these categories. If the survey is not completed, a single reminder email and survey link will be sent one week later. The inclusion criteria are as follows:

1. Individuals with a germline high-risk PV, which predisposes them to disease requiring risk mitigation strategies.
2. Age 18 years or over.

The exclusion criteria would be individuals unable to provide consent for any reason and non-English speakers.

Data analysis: Relevant descriptive statistics will be reported using SPSS software. The survey data will be used to describe the cohort demographics and previous and current apps use and experience.

Phase 2

Determine the potential benefits, uses, functions and attributes of a digital health platform from the perspectives of (1) individuals at high-risk of a genetic disease and (2) clinicians who manage the care of these individuals (0–9 months).

Focus groups provide an interactive process which encourages participants to share and clarify their thoughts, as well as to express similarities and differences.[50] Focus group discussions enable participants to offer concepts that may not have been considered by the researchers.[51] This is important here as there is limited literature on this topic. Focus groups will be run by an implementation scientist with support from specialists in clinical genetics and human-centred design. Discussion will be guided by a semi-structured focus group protocol. The inclusion and exclusion criteria are identical to phase 1.

Patient stakeholder group: A series of online focus groups of 4–6 individuals or personal interviews, as preferred by participants, will be undertaken with up to 40 individuals in total from the stakeholder patient group. Initial focus group discussions will centre on digital literacy and preferences to inform the development of a prototype of the digital health platform. Later, focus groups will share a simple basic prototype with participants to allow for user feedback and refinement before initial platform development.

Focus groups with individuals at risk of cancer or cardiac disease will be held separately.

Up to 400 individuals (assuming 10% response rate), purposively selected based on age, gender, geographic location of home address (urban/rural) and length of time since they had their genetic test. Purposeful selection across these parameters will be undertaken to ensure a wide range of age-related and geographic barriers, enablers, opinions and ideas are collected. The focus groups will be run online due to remaining COVID-19 restrictions in Victoria and provides the opportunity to recruit from a wide geographical population.

Clinician stakeholder group: In parallel, a series of focus groups will be undertaken with the stakeholder clinician group. Participants will be drawn from the CGS-associated clinician group described previously. Four to six focus groups with between 4 and 6 clinicians will be held, with up to 30 clinicians invited in total. Clinicians will be purposively sampled to ensure representation from the diverse set of clinical specialities: breast surgeons, medical oncologists, gynaecologists, clinical genetic physicians and counsellors, specialist management clinic coordinators, cardiologists and nurses.

The discussion framework for both sets of focus groups will be semi-structured based on digital intervention design and UTAUT[52] (online supplemental appendix B: Consumer and clinician focus group guides (predevelopment and user experience of prototype)).

Data analysis: Content analysis will be undertaken informed by the UTAUT to identify domains of interest for platform development. This will entail development of a content coding grid to align the UTAUT constructs in the context of codesign and development. A selection of transcripts will be considered independently by at least two researchers to achieve consensus and to ensure scientific rigour, being repeatedly read and reread and sections of data cross-compared and then coded. This coding method enables the identification of recurrent themes.

Phase 3

Map the core skillset, resources and support requirements of a digital health coordinator role within the GCS (0–9 months).

A clinical review of patient self-reported needs in the medium and long term over the last 2 years identified through the PFCC's 'ongoing care programme' will be undertaken. Needs will be categorised by type, such as organisational (difficulty navigating healthcare system), informational (management recommendations), family communication, psychological and clinical. An audit of the clinical service processes required to resolve or provide support for each need category, including skillset required at all stages, will be determined. An additional audit will be made of department-instigated communication with patient groups.

Data analysis: The audit data will be assessed by the research team to identify potential areas where the digital tool could provide a first-line adjunct to care, how this would most appropriately be instituted, and the skillsets and support required to achieve this. Through this interactive process, the competencies and level of autonomy required by the digital health coordinator role can be assessed against the position accountability and influence.[53] These data in association with the known skillsets and salaries of specialists within the CGS can be used to determine the most appropriate combination of skillsets required by this position.

Phase 4

Prototype development, user testing, evaluation and refinement (9–24 months)

Once the first version of the digital prototype is co-developed, its 'acceptability', 'feasibility' and 'useability'[45] will be beta-tested in a small group of patients and clinicians. To test the prototype, we will recruit 10 patients and 10 clinicians (5 from the previous focus groups and 5 new participants). The prototype will be installed on a laptop/desktop computer and mobile phone, and participants will be given 10 min of training to interact with the prototype. Later, participants will be asked to complete 5–10 tasks, and they will be asked to comment on their experience with the prototype. During the interaction with the prototype, the participant will be asked to think aloud.[54] The session will be audiorecorded and transcribed later for further analysis. Through the think-aloud process, we aim to find out the users' interactions and experiences with the features and functions of the prototype to improve overall user experience and ease of use.

Data analysis

Data from the testing session will be analysed thematically based on the Theoretical Domain Framework (TDF) to identify the barriers and enablers using the tool.[55 56] Participants' concerns with digital tool interface will be mapped against possible recommendations to improve them. Finally, the recommendations will be prioritised for the next version of the tool.

Patient and public involvement

This project was driven by the ongoing needs of patients diagnosed with a high cancer or cardiac risk who contact the CGS requesting information, support or clinical advice. The design and functionality of this user-centric digital health platform will be co-designed and modified based on patient-generated needs directly elicited iteratively through focus groups or personal interviews. Further personalisation will occur after the platform has been developed. Dissemination and implementation plans are being developed in tandem with the design, but we will work closely with consumers and consumer advocates in achieving those aims once the platform is developed.

DISCUSSION

Digital health solutions have the potential to aid the lifelong, personalised care of people living with a high-risk genetic predisposition by improving both health outcomes and their experience of accessing and traversing the health system. In Australia, this would include being informed of services available in the public sector so that the use of the private healthcare with its associated costs was an informed choice. The COVID-19 pandemic has accelerated increasing acceptance from clinicians, patients and importantly healthcare funders for digital technology such as telehealth in the provision of clinical care.[57] What has not been established to date is how this specific population would choose to interact with a digital health platform, and ultimately how it would influence their behaviour. This study aims to start addressing this gap by co-designing and developing a bi-directional user-centric digital health platform.

Such a platform would allow patients to send information, appointments or contact requests at a time and place of their choosing to the CGS and know they will be actioned. The patient would then receive a response to their request within the platform within a transparently prespecified time. Alternatively, it would enable the CGS to notify specific subgroups of patients of changes to clinical risk management guidelines, or new clinical trials for which they have been identified as being eligible.

Capturing a granular understanding of patients' current use of smartphone apps and preferences for a mHealth app that meets their needs will improve uptake and retention of use. Concurrently, the clinician perspective is vital to the adoption of the user-centric digital health platform: without clinician input into content and useability the app is unlikely to succeed.

Aligning with both the UTAUT and later the TDF, we aim to determine how users (both clinicians and patients) would prefer to interact with the platform (UTAUT) and how that interaction will impact their health behaviour (TDF). This approach will support future evaluation and modification of the platform over its lifecycle, and improve the acceptability and useability of the platform to both patients and clinical services.

This initial stage also provides an opportunity to craft the novel role of a digital health coordinator creating the required competencies informed by live data. This role will ensure the platform functionality remains relevant as genomic testing and management guidelines evolve, and provides a pivotal link between patient, clinician, data and data analysis.

Another area that has gained much attention in recent years and will be incorporated into this digital health platform once developed is the use of AI to assess and improve clinical outcomes. AI algorithms provide promising outcomes in several clinical diagnostics, including genetic testing.[37] A smart user-centric digital health platform powered by AI algorithms can help to undertake gene-specific analysis by interrogating clinical outcome data conditioned on risk management strategies, socioeconomic or geographic location. AI algorithms will be able to identify specific subgroups of patients at key clinically relevant life stages—such as women with *BRCA1* PVs at an age when risk-reducing ovarian cancer management is recommended or who are at risk of not adhering to management recommendations, and proactively alert the CGS genetic counsellors to those patients. A directional communication via the digital health platform could then be used to notify this subgroup of patients, ask if they require support, information or help in navigating the healthcare system and collect the responses to these questions or requests for clinic follow-up. Such technology will help to assess, in tandem with data collection, the performance of the current clinical risk assessments provided by the CGSs, and the effectiveness of risk mitigation strategies. Further, a digital health coordinator role would support the genetic counsellors in ensuring the accuracy and timing of care delivery.

Future work will build on the platform design and data captured by undertaking an adapted hybrid 1 trial[58] where the implementation of the platform will be simultaneously tested alongside its uptake. At this stage, there will be a focus on increasing the diversity of platform users including people with culturally and linguistically diverse backgrounds, and people with varying levels of digital literacy, and in assessing the impact that other factors, known to exacerbate the digital divide,[59] may have on optimal uptake and use of this tool across the target population.

A mixed-methods approach will be used to investigate implementation and clinical outcome measures. Concurrent testing of implementation outcomes, costs, reach and sustainability alongside impact on patient behaviour will be examined to provide detailed analysis of the complex factors associated with sustained adoption of digital health

interventions. The 12-point Template for Intervention Description and Replication checklist will be used to guide the development of implementation strategies and recording of any platform refinements required.[60]

To the best of our knowledge, this study is the first real-world example of a digital tool to support the long-term care of people with a genetic predisposition disease. However, it does have limitations. As we use a range of online data collection tools—digital survey, online focus groups—to design a digital tool, this approach will bias our sample and platform development towards people who have a good level of digital literacy. An online approach does provide more potential to recruit a geographically disparate population but is limited to those with access to the internet. While not addressed in the initial platform development stage, we plan to actively seek out a more diverse population during a future hybrid 1 stage of the study. Equally, a lack of English precludes people from participating in the early phases of this project outlined in this protocol. We plan to proactively recruit people with culturally and linguistically diverse backgrounds during the future phases of the study.[58] At this point, the platform will still be amenable to modifications, and both content and useability can be addressed. Participants in this study will be recruited from a specialist Familial Cancer Centre in Victoria, Australia. This will limit the pool of participants to those actively engaged in their healthcare which may not be reflective of all Australians.

Digital tools herald great promise as a complement, and at times, a substitute for in-person care, but the primary goal of this approach should always remain on person-centred care. It is essential that such tools are co-designed with the endusers and vigorously evaluated to optimise potential impact.

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Contributors AHT, SB, PPJ, AAM, ST, ARMF and NW designed this study and in conjunction with JCWW, AL, NS, RK and AW were instrumental in obtaining HREC approval. All authors were responsible for drafting, editing and finalising this manuscript. The manuscript has been approved by all authors.

Funding This work was supported jointly by Peter MacCallum Cancer Centre, Swinburne University of Technology and Digital Health CRC Limited (DHCRC). Grant award number: DHCRC -0131. DHCRC is funded under the Commonwealth's Cooperative Research Centres Program.

Competing interests None declared.

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

Patient consent for publication Not applicable.

Provenance and peer review Not commissioned; externally peer reviewed.

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