


Original research

Public willingness to participate in population DNA screening in Australia

Jane M Tiller ¹, Andrew Bakshi,¹ Adam R Brotchie,¹ Robert C Green,² Ingrid M Winship,³ Paul Lacaze¹

► Additional supplemental material is published online only. To view, please visit the journal online (<http://dx.doi.org/10.1136/jmg-2022-108921>).

¹School of Public Health and Preventive Medicine, Monash University Faculty of Medicine Nursing and Health Sciences, Melbourne, Victoria, Australia
²Harvard Medical School, Boston, Massachusetts, USA
³Clinical Genetics, Royal Melbourne Hospital, Parkville, Victoria, Australia

Correspondence to

Jane M Tiller, Department of Epidemiology and Preventive Medicine, Monash University, Clayton, VIC 3800, Australia; jane.tiller@monash.edu

Received 7 September 2022
Accepted 1 November 2022

ABSTRACT

Background Population-based DNA screening for medically actionable conditions has the potential to improve public health by enabling early detection, treatment and/or prevention; however, public attitudes and willingness to participate in DNA screening have not been well investigated.

Methods We presented a scenario to members of the Australian public, randomly selected from the electoral roll via the Australian Survey of Societal Attitudes, describing an adult population DNA screening programme currently under development, to detect risk of medically actionable cancers and heart disease. We asked questions regarding willingness to participate and pay, preferred delivery methods and concerns.

Results We received 1060 completed questionnaires (response rate 23%, mean age 58 years). The vast majority (>92%) expressed willingness to undertake DNA screening. When asked about the optimal age of screening, most (56%) favoured early adulthood (aged 18–40 years) rather than at birth or childhood. Many respondents would prefer samples and data be kept for re-screening (36%) or research use (43%); some preferred samples to be destroyed (21%). Issues that decrease likelihood of participation included privacy (75%) and insurance (86%) implications.

Conclusion Our study demonstrates public willingness to participate in population DNA screening in Australia, and identifies barriers to participation, to be addressed in the design of screening programmes. Results are informing the development of a pilot national DNA screening programme.

INTRODUCTION

Population-based genomic testing (or ‘adult DNA screening’) for medically actionable conditions has the potential to improve public health by enabling early detection, treatment and/or prevention.^{1–4} This includes certain familial cancer predisposition syndromes and inherited heart conditions caused by single germline pathogenic DNA variants (table 1). For such conditions, evidence-based guidelines, interventions and risk management options are available for high-risk individuals.^{5–10} However, diagnostic rates and access to reimbursed genetic testing for these conditions remain very low. Population DNA screening for these conditions, especially if offered in early adulthood through a public healthcare system, has the potential to greatly improve detection and diagnosis, compared with

WHAT IS ALREADY KNOWN ON THIS TOPIC

⇒ Population-based DNA screening for medically actionable conditions has the potential to improve public health by enabling early detection, treatment and/or prevention; however, public attitudes and willingness to participate in DNA screening have not been well investigated.

WHAT THIS STUDY ADDS

⇒ Members of the general public are highly interested in participating in DNA screening for medically actionable conditions, however there are several concerns which may decrease their likelihood of participating, such as privacy and insurance concerns.

HOW THIS STUDY MIGHT AFFECT RESEARCH, PRACTICE OR POLICY

⇒ This study provides important information for policy-makers regarding public preferences and barriers to participation, which must be addressed in the design of future screening programmes.

the current status quo of clinical criteria-based genetic testing.

Despite this opportunity, and increasing public interest in genetic testing,¹¹ no countries have yet begun offering preventive, population-level DNA screening for these conditions to adults via a national public healthcare system. Unlike some countries, Australia has a national, government-funded universal public healthcare system. Some large US healthcare networks have commenced offering preventive genomic testing to their members.^{12–14} However, this testing is limited only to certain networks and locations, and not delivered via a national public healthcare system.

In Australia, a national pilot study of adult DNA screening has recently been established,² offering DNA screening to 10 000 young adults aged 18–40 years for three adult-onset medically actionable genomic conditions (table 1). The *DNA Screen* pilot study will assess the delivery of population DNA screening for medically actionable conditions in Australia, and has been designed to inform the future potential development of a national population-based DNA screening programme. This proposed programme would be delivered through



© Author(s) (or their employer(s)) 2022. No commercial re-use. See rights and permissions. Published by BMJ.

To cite: Tiller JM, Bakshi A, Brotchie AR, *et al.* *J Med Genet* Epub ahead of print: [please include Day Month Year]. doi:10.1136/jmg-2022-108921

Table 1 Medically actionable genomic conditions for population DNA screening

Genetic predisposition	Associated high-risk condition	Risk management interventions
Hereditary breast and ovarian cancer	Increased risk of breast cancer in women	High-risk breast surveillance, chemoprevention and risk-reducing mastectomy to reduce breast cancer risk ⁸
	Increased risk of ovarian cancer in women	Risk-reducing salpingo-oophorectomy to significantly lower risk ⁴⁰
	Increased risk of breast and prostate cancer in men	Increased surveillance from a younger age than population screening programmes
Lynch syndrome	Increased risk of colorectal cancer in men and women	Aspirin use and regular colonoscopy, which together can reduce risk ⁹
	Increased risk of gynaecological cancers in women	Risk-reducing hysterectomy to reduce endometrial cancer risk ⁴¹
Familial hypercholesterolaemia	Increased risk for heart disease or stroke due to high cholesterol levels (with genetic causes) from an early age	Use of statins and other cholesterol-lowering agents to reduce risk of myocardial infarction ^{10,42}

the public healthcare system, in accordance with the National Population-Based Screening Framework.¹⁵

Prior research shows Australians value the utility of genomic data¹⁶; however, public attitudes and willingness to participate in DNA screening have not been well investigated. In 2016–17, a survey of the Australian public investigated perspectives about personal genomic testing.¹⁷ However, the findings^{18,19} were not focused on population DNA screening. Similarly, recent international surveys have focused on issues such as data sharing^{20–22}; ethical issues around receipt of genomic screening information²³; prenatal genomic testing and newborn screening^{24–26}; reproductive carrier screening^{27,28} and willingness to pay for commercial genomic sequencing.¹⁶ In the present study, we sought to survey the Australian public's attitudes and willingness to participate in a DNA screening programme for medically actionable conditions, and to use the findings to help guide the development of the *DNA Screen* national pilot study.²

METHODS

We partnered with the Australian Consortium for Social and Political Research, who routinely administer the Australian Survey of Social Attitudes (AuSSA)²⁹—the Australian component of the International Social Survey Project, a collaboration of researchers in 40 countries.³⁰ The AuSSA randomly selects citizens quarterly from the Australian electoral roll (for which all citizens), and posts an explanatory letter, followed a week later by a questionnaire booklet and reply-paid envelope. In Australia, voting in political elections is compulsory, and all citizens ≥ 18 must legally register on the electoral roll and update their name and address details if they change. Our study findings were drawn from responses to surveys sent between May 2021 and February 2022.

We developed survey questions about DNA screening (see online supplemental file S1) through a workshop of clinical, research and policy experts. Questions were based largely around a hypothetical DNA screening programme (box 1), covering respondents' hypothetical willingness to have DNA screening; opinions about sample collection methods, age of testing and use/storage of the data; willingness to pay and potential concerns that may be a barrier to participation. No validated scales existed for the development of questions about this hypothetical scenario. However, potential participant concerns were based on issues raised by clinical and research partners who assisted with survey development. The questions were piloted on ~100 respondents who had previously participated in AuSSA surveys, to enable amendment or clarification of language (which was ultimately not required).

We hypothesised that individuals with and without a personal or family history of cancer, and older versus younger individuals, may have different attitudes towards DNA screening. Therefore,

age and cancer history were collected in order to accommodate subgroup analysis and examine any such differences.

Statistical analysis

Questionnaire data were entered and coded using queXF/queXC software,³⁰ and made available to researchers quarterly. We conducted descriptive analysis of the data, including subgroup analysis of younger (18–40 years) and older (>40 years) respondents and those with and without a self-reported personal/family cancer history. Differences between categories were tested using a χ^2 test of independence. Significant ($p < 0.05$) differences of interest between groups have been highlighted in the results.

RESULTS

Of 5000 invitations sent (4657 eligible), 1060 eligible surveys were returned, for an overall response rate of 23%. Ineligible invitees ($n=343$) were excluded for reasons including

Box 1 Scenario used for genomic testing questions in survey

The following text was used to introduce the hypothetical DNA screening test in the survey:

'Genetic testing can be used to predict the chance of developing certain future health conditions. Some of these include types of cancer (breast, bowel, etc) and some heart conditions. It is now possible to take a DNA test to identify increased risk of developing these conditions in adulthood. If detected early, these conditions can be prevented or treated, which can be life-saving. However, most people currently do not know they are at risk, due to limited access to testing. Around 2% of people in the population have such a gene change, which could be identified by a genetic test. Genetic information is passed down through the generations. Thus, every first-degree relative (parent/child/sibling) of an individual with a gene change also has a 50% chance of having that gene change. Genetic testing can help us to identify whole families who are unaware of their genetic risk.

For this section, imagine you are offered a genetic test that predicts your chance of developing three such future health conditions:

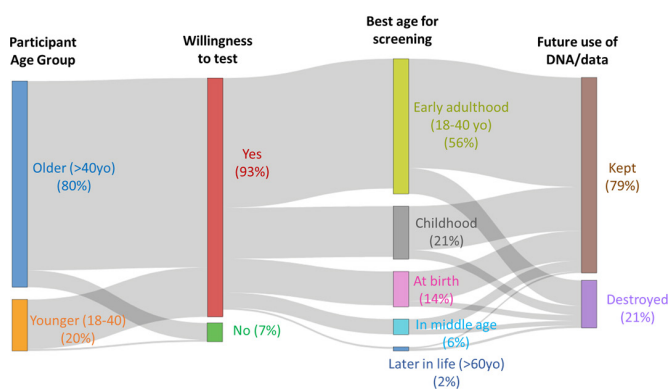
1. breast and ovarian cancer occurring due to a genetic change in the *BRCA1/BRCA2* genes (such the genetic change that Angelina Jolie has, which many people have read about);
2. bowel cancer occurring due to a genetic change;
3. genetically high cholesterol, which stays high regardless of your lifestyle, and can cause life-threatening early heart attacks'.

Table 2 Demographic information

Gender (n=1021)	N (%)
Female	569 (55.7)
Male	452 (44.3)
Age (n=983) (mean 58.3; SD 16.83)	
Over 40 years	787 (80.1)
18–40 years	196 (19.9)
Cancer history (family or personal) (n=978)	
Yes	477 (48.7)
No	502 (51.3)
Highest level of education (n=979)	
Year 10 or equivalent, or less	150 (15.3)
Year 11 or 12 or equivalent	104 (10.6)
Postschool certificate/diploma	328 (33.5)
Undergraduate qualification	290 (29.6)
Postgraduate qualification	107 (10.9)

incorrect address, physical/language difficulties with responding or death. Respondents were 56% female, with mean age 58 years (table 2). Of those who disclosed their age and/or cancer history (optional), 20% (n=196/983) were aged 18–40 years (the ‘younger’ subgroup), and 49% (n=477/978) had a personal or family history of cancer in first-degree relatives (the ‘cancer history’ subgroup). As respondents could skip questions, some respondents did not answer all questions. The number of respondents for each question is noted throughout the results.

Figure 1 shows an overview of our study findings. A full summary of all findings is included at online supplemental file S2. When asked about their interest in having screening as described in box 1 (n=1032), 92% of survey respondents were willing to have DNA screening, and only 8% of respondents said they would not participate. Figure 2 highlights a number of particular participant preferences and views. Figure 2A shows willingness to pay categories (multiple could be selected). Responses to this question indicated that 32% of respondents would be willing to pay between \$A1 and \$A20 for the test, 42% between \$A21 and \$A100; 18% between \$A101 and \$A500; 5% between \$A501 and \$A1000 and 4% >\$A1000. A larger proportion of respondents (54%) would prefer to take the test if paid for by the public health system. Some participants would rather pay for the test, rather have it for ‘free’ (through a public healthcare system). Younger subgroup respondents were even more willing to participate in public health-based screening than the older subgroup—66% of younger respondents (n=106/161) would prefer to take the test if paid for by the public health system,

**Figure 1** Overview of study findings.

compared with 51% of over 40s (n=288/559) (p=0.0002). There was no difference between the willingness to participate of respondents with or without cancer history (p=0.7).

When asked about interest in DNA screening based on the likelihood (risk) of having particular health conditions (n=986), respondents' interest in screening increased with increasing levels of hypothetical risk. For a disease with risk of 1/1000, 47% of respondents were interested in screening. For a disease with risk of 1/20, 65% were interested (increasing further to 76% for those aged 18–40 years (n=142/187), compared with 63% for those over 40s (n=459/732) (p=0.0002).

When respondents were asked about the best age to offer DNA screening (n=723), a minority chose childhood (21%), at birth (14%), middle age (6%) or later in life (2%). The majority (56%) chose early adulthood (aged 18–40 years). This percentage was higher in the younger subgroup (65%; n=123/188); however, even among the older subgroup, the majority (53%; n=390/735) chose early adulthood (p=0.003).

Respondents' preferences regarding DNA sample collection (n=981) and use/storage of genetic data (n=1000) included a majority (55%) preference for providing a sample via a pathology lab, followed by a preference for saliva kits posted to their home (18%). Younger respondents were more likely than older respondents to prefer a saliva kit (25% (n=45/182) vs 16% (n=116/738); p=0.00002). Most respondents preferred genetic data be either kept for future re-screening for more conditions (36%) or future research (43%). Of those who chose storage for future research (n=426), 73% preferred it only be available to universities and non-profit research institutions; the remaining 27% also accepted availability to for-profit researchers such as pharmaceutical companies. Overall, 21% would want their DNA sample and data destroyed after screening, with this percentage higher in younger versus older subgroups (27% (n=51/188) vs 18% (n=137/749); p=0.01).

We asked respondents about trust in different entities to manage their genomic data (figure 2B). Overall, the most respondents indicated a low level of trust in commercial companies (85%; n=757/888), and a high/medium level of trust in Australian universities or research institutions (79%; n=721/909) and the Commonwealth Department of Health (83%; n=829/999). When asked about inclusion of DNA screening results in the Australian electronic health record (*My Health Record*) (n=1002), most respondents preferred results either be included automatically (44%) or with specific consent (40%), while 16% preferred results not be included.

When asked about potential concerns regarding DNA screening that might deter participation (figure 2C), the possibility of life insurance companies using results in underwriting (n=957) was the most common choice—86% of respondents said insurance concerns would definitely (61%) or may (25%) decrease their likelihood of participating. Privacy concerns (eg, what would happen with the data) (n=971) were the next most common choice. Respondents stated that privacy concerns would definitely (31%) or may (44%) decrease likelihood of participating. Other potential concerns listed were more likely to not affect participation—including not wanting to know about risks of developing disease in the future (71%; n=671/947), uncertainty about what steps they would be willing to take to prevent developing disease (69%; n=654/944) and concern that they might pass/have passed genetic risk onto their children (78%; n=739/951). An overwhelming majority (89%) would be more comfortable taking this test if a law was in place to protect their information (n=1014). This was not associated with age or cancer history.

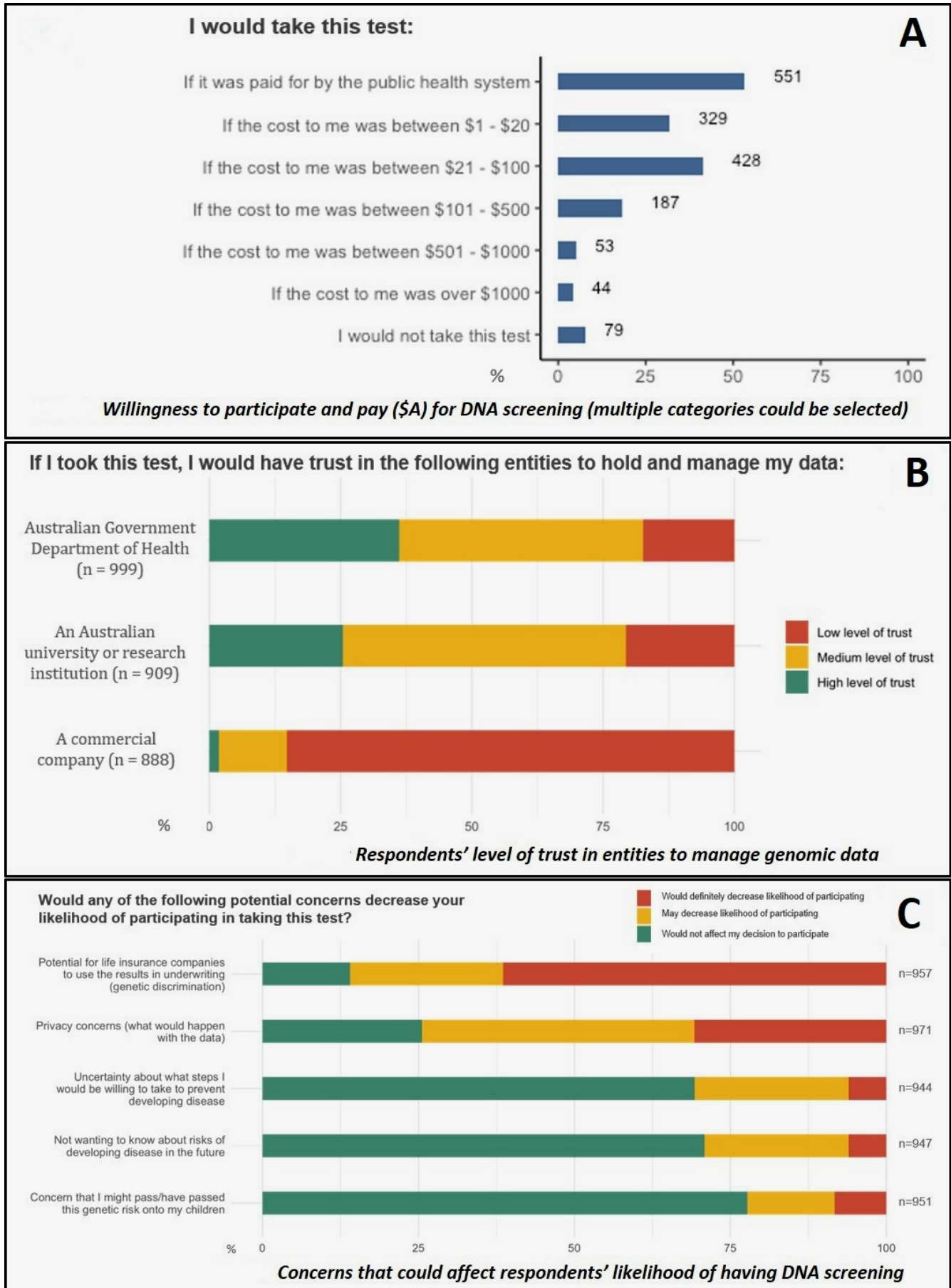


Figure 2 Participant preferences and views.

Finally, respondents were asked general questions about their views and understanding of genetic information. Respondents most commonly associated the phrase ‘human genetic information’ (n=1035), with ‘DNA’ (79%), followed by ‘inherited health problems’ (54%) and ‘medical research’ (45%). When asked whether genetic information is the same as other medical information (like blood pressure) or different, 63% of respondents (n=600/947) said genetic information is different to other medical information. This was not associated with age or cancer history.

DISCUSSION

This is the first study, to our knowledge, to specifically gather the attitudes of the Australian public towards a potential national DNA-based population screening programme. Our study provides evidence of strong public acceptability and interest in adult DNA screening, as a new strategy for public health prevention. Our findings provide important evidence regarding the Australian public’s attitudes and preferences, to inform future research and policy. Of the 1060 survey respondents, only 8% indicated they would not participate in DNA screening, indicating strong willingness to participate. The findings of the study have helped inform the development of the *DNA Screen* national pilot study,² which is assessing delivery models and acting as a proof-of-concept for a potential future DNA screening programme in Australia.

The public’s willingness to participate will be critical to the success of any future DNA-based population screening programme, necessitating the exploration of public views, preferences and concerns.²³ Our survey was designed to explore public sentiment on pertinent issues related to population DNA screening, that could directly impact the future success of a national DNA-based screening programme. The responses provide an encouraging indication that the Australian public’s views accord with several of our prior expectations and hypotheses. This includes the optimal age to offer DNA screening (18–40 years), a preference for screening through the public healthcare system (which is universal and government-funded in Australia, unlike in some other countries), use and storage of samples and data and concerns around privacy and insurance issues.

We found that 92% of our survey respondents were willing to have DNA screening, confirming our intention to provide greater access to DNA testing for medically actionable conditions in Australia. Some respondents reported willingness to pay at various amounts, which could appear to support a ‘co-pay’ programme. However, this raises equity issues and would not be consistent with the Australian Government’s Population-based Screening Framework, ‘underpinned by the principles of access and equity, which are fundamental elements of all population screening programmes’ (Clinical Principal Committee Standing Committee on Screening,¹⁵ p. 4). Other currently implemented Australian population-based screening programmes, such as the National Bowel Cancer Screening Programme and the National Breast Cancer Screening Programme, are funded by the federal government and delivered as part of the national healthcare system, at no charge to the end user, in order to optimise uptake and equity of access. International research shows that out-of-pocket costs create inequities and barriers to accessing clinical genomic testing,³¹ and early Australian examples in other genomic contexts have led to calls for public funding to reduce inequity.^{32 33}

The high proportion of personal or family history of cancer (almost 50% of respondents) indicates some potential ascertainment bias of respondents with cancer experiences being interested in this survey. However, contrary to our hypothesis, no differences were found in willingness to test or attitudes towards DNA screening based on cancer history. This indicates a high interest in DNA screening even among Australians without personal experiences of cancer. The preference for sample collection at a pathology lab over home saliva kits was also surprising. Although the preference was stronger in the older subgroup, it still prevailed in the younger subgroup. Without further explanation by respondents, we are uncertain whether this reflects personal preference or a perceived difference in accuracy of DNA testing derived from blood versus saliva, or a familiarity with pathology collection centres versus at-home saliva collection. The recent familiarisation of the public with rapid antigen testing for COVID-19 might affect future opinions on saliva testing since the survey was administered.

‘Genetic exceptionalism’ refers to the idea that genomic information is different from other types of medical information, and therefore deserves special treatment.^{34–36} Although genetic exceptionalism is often criticised, a recent international study²⁰ showed strong correlation between genetic exceptionalist views in the general public and a perception of the clinical and scientific value of genomic information, as well as willingness to share/donate genomic data for research. Most respondents in our study felt genomic information is different from other types of medical information.

We also found a strong willingness for use of respondents’ genomic data for research, although with a small increase in preference for data destruction in younger subgroups. The majority preference for data to only be made available to universities and not-for-profit researchers, rather than for-profit researchers, is consistent with levels of trust reported—most respondents reported a medium-high level of trust in Australian universities/research institutions, but a low level of trust in commercial companies. These preferences may vary from country to country. In Australia, the *DNA Screen* study will offer participants options regarding future use of their data, including an option for destruction of sample and data following screening.

Despite positive attitudes towards DNA screening, respondents identified concerns that would affect participation, including data privacy and genetic discrimination. Although privacy concerns can be addressed through study design, concerns regarding life insurance discrimination are complex and require regulation. Use of genetic data by life insurers is a key ethical concern and research topic internationally.^{37 38} For population DNA screening to be feasible in Australia, the issue of insurance discrimination must be addressed. Our findings show that legal protection of genomic data would increase Australians’ willingness to participate in DNA screening, which in turn could help deliver the benefits of genomic medicine.

Strengths of the study include a large sample size and partnership with an established research consortium, and recruitment through an unbiased recruitment strategy. Limitations include the hypothetical nature of the population DNA screening programme presented to the respondents, meaning reported behaviours and preferences may differ from actions actually taken in reality. The study also only includes Australian participants, limiting the generalisation of our findings to other countries. We did not undertake demographic, cultural or linguistic subgroup analysis, meaning our findings may not reflect possible differences in attitudes and preferences driven by cultural or demographic variation. However, the respondent demographics

(see online supplemental file S2) show our sample had similar numbers of individuals born overseas (25.9%) and identifying as Aboriginal or Torres Strait Islander (3.3%) as 2021 Australian census data (29.3% and 3.2%, respectively), demonstrating a representative sample on these measures.³⁹ Regarding willingness to test, it is unclear why some respondents would be willing to pay for genetic testing but would not take testing if paid for by the public health system. It is possible that the question was confusing for respondents, or that respondents were sceptical about testing being 'free'. Future research could focus on understanding the reasons for these choices.

Our study has the potential for responder bias, although this may have been mitigated through the variety of topics covered on the overall AuSSA survey beyond our specific questions about genetics. Respondent age was skewed towards an older demographic (mean age 58 years), which may reflect the paper-based survey methodology used. Fortunately, the large number of respondents enabled subgroup analysis to elucidate age-based differences.

Our study demonstrates strong public willingness in Australia for participation in a national DNA-based population screening programme, to detect risk of medically actionable conditions, such as cancer and heart disease. It also identifies barriers to participation in DNA-based screening, which must be considered in the design of future programmes. The findings of this survey are already informing the design and development of the *DNA Screen* national pilot study, which has the potential to shape the future of DNA screening research and policy in Australia and other countries. Future research should focus on evaluation of existing screening programmes and identification of key barriers and enablers to the implementation of population-level DNA screening programmes.

Twitter Jane M Tiller @JaneMTiller

Acknowledgements The authors wish to acknowledge the Australian Consortium for Social and Political Research Incorporated (ACSPRI), for its support and expertise in administering the Australian Survey of Social Attitudes (AuSSA).

Contributors Conceptualisation: JMT, PL. Formal analysis: AB, JMT. Methodology: JMT, PL, IMW. Supervision: PL. Writing—original draft: JMT, PL. Writing—review and editing: JMT, AB, ARB, IMW, RCG, PL. Guarantor: PL

Funding This study was funded by National Heart Foundation Future Leader Fellowship (102604).

Competing interests RG has received compensation for advising the following companies: AIA, Allelica, Fabric, Genome Web, Genomic Life, Grail, Humanity, OptumLabs, Verily, VinBigData; and is co-founder of Genome Medical and Nurture Genomics. PL, JMT, IW and AB are named investigators on and ARB is project manager of the DNA Screen study, which is funded by an Australian Government Medical Research Futures Fund Genomics Health Futures Mission grant no. 2021/MRF2009024.

Patient consent for publication Not applicable.

Ethics approval This project was granted approval by the University of Tasmania Social Sciences Human Research Ethics Committee on 24 May 2021, ID number 23546, and was performed in accordance with the ethical standards as laid down in the 1964 Declaration of Helsinki. Participants gave informed consent to participate in the study before taking part.

Provenance and peer review Not commissioned; externally peer reviewed.

Data availability statement Data are available on reasonable request. Data collected through the AuSSA is made publicly available via the Australian Data Archive (subject to some embargoes on sponsored questions). A summary of data collected through the current study is included as a supplementary file to this manuscript.

Supplemental material This content has been supplied by the author(s). It has not been vetted by BMJ Publishing Group Limited (BMJ) and may not have been peer-reviewed. Any opinions or recommendations discussed are solely those of the author(s) and are not endorsed by BMJ. BMJ disclaims all liability and responsibility arising from any reliance placed on the content. Where the content

includes any translated material, BMJ does not warrant the accuracy and reliability of the translations (including but not limited to local regulations, clinical guidelines, terminology, drug names and drug dosages), and is not responsible for any error and/or omissions arising from translation and adaptation or otherwise.

ORCID iD

Jane M Tiller <http://orcid.org/0000-0003-3906-6632>

REFERENCES

- Lacaze P, Tiller J, Winship I. Healthcare system-funded preventive genomic screening: challenges for Australia and other single-payer systems. *Public Health Genomics* 2019;22:140–4.
- Lacaze PA, Tiller J, Winship I, DNA Screen Investigator Group. Population DNA screening for medically actionable disease risk in adults. *Med J Aust* 2022;216:278–80.
- Khoury MJ, Dotson WD. From genes to public health: are we ready for DNA-based population screening? *Genet Med* 2021;23:996–8.
- Murray MF, Evans JP, Khoury MJ. DNA-based population screening: potential suitability and important knowledge gaps. *JAMA* 2020;323:307–8.
- Riedl CC, Luft N, Bernhart C, Weber M, Bernathova M, Tea M-KM, Rudas M, Singer CF, Helbich TH. Triple-modality screening trial for familial breast cancer underlines the importance of magnetic resonance imaging and questions the role of mammography and ultrasound regardless of patient mutation status, age, and breast density. *J Clin Oncol* 2015;33:1128–35.
- Ludwig KK, Neuner J, Butler A, Geurts JL, Kong AL. Risk reduction and survival benefit of prophylactic surgery in BRCA mutation carriers, a systematic review. *Am J Surg* 2016;212:660–9.
- Møller P, Seppälä T, Bernstein I, Holinski-Feder E, Sala P, Evans DG, Lindblom A, Macrae F, Blanco I, Sijmons R, Jeffries J, Vasen H, Burn J, Nakken S, Hovig E, Rodland EA, Tharmaratnam K, de Vos Tot Nederveen Cappel WH, Hill J, Wijnen J, Green K, Lalloo F, Sunde L, Mints M, Bertario L, Pineda M, Navarro M, Morak M, Renkonen-Sinisalo L, Frayling IM, Plazzer J-P, Pylvanainen K, Sampson JR, Capella G, Mecklin J-P, Möslin G, Mallorca Group. Cancer incidence and survival in Lynch syndrome patients receiving colonoscopic and gynaecological surveillance: first report from the prospective Lynch syndrome database. *Gut* 2017;66:464–72.
- Rebeck TR, Friebel T, Lynch HT, Neuhausen SL, van 't Veer L, Garber JE, Evans GR, Narod SA, Isaacs C, Matloff E, Daly MB, Olopade OI, Weber BL. Bilateral prophylactic mastectomy reduces breast cancer risk in BRCA1 and BRCA2 mutation carriers: the PROSE Study Group. *J Clin Oncol* 2004;22:1055–62.
- Burn J, Gerdes A-M, Macrae F, Mecklin J-P, Moeslein G, Olschwang S, Eccles D, Evans DG, Maher ER, Bertario L, Bisgaard M-L, Dunlop MG, Ho JWC, Hodgson SV, Lindblom A, Lubinski J, Morrison PJ, Murday V, Ramesar R, Side L, Scott RJ, Thomas HJW, Vasen HF, Barker G, Crawford G, Elliott F, Movahedi M, Pylvanainen K, Wijnen JT, Fodde R, Lynch HT, Mathers JC, Bishop DT, CAPP2 Investigators. Long-term effect of aspirin on cancer risk in carriers of hereditary colorectal cancer: an analysis from the CAPP2 randomised controlled trial. *Lancet* 2011;378:2081–7.
- Watts GF, Sullivan DR, Hare DL, Kostner KM, Horton AE, Bell DA, Brett T, Trent RJ, Poplawski NK, Martin AC, Srinivasan S, Justo RN, Chow CK, Pang J, FH Australasia Network Consensus Working Group. Integrated guidance for enhancing the care of familial hypercholesterolaemia in Australia. *Heart Lung Circ* 2021;30:324–49.
- Ramos E, Weissman SM. The dawn of consumer-directed testing. *Am J Med Genet C Semin Med Genet* 2018;178:89–97.
- Buchanan AH, Lester Kirchner H, Schwartz MLB, Kelly MA, Schmidlen T, Jones LK, Hallquist MLG, Rocha H, Betts M, Schwiter R, Butry L, Lazeri AL, Frisbie LR, Rahm AK, Hao J, Willard HF, Martin CL, Ledbetter DH, Williams MS, Sturm AC. Clinical outcomes of a genomic screening program for actionable genetic conditions. *Genet Med* 2020;22:1874–82.
- Grzymalski JJ, Elhanan G, Morales Rosado JA, Smith E, Schlauch KA, Read R, Rowan C, Slotnick N, Dabe S, Metcalf WJ, Lipp B, Reed H, Sharma L, Levin E, Kao J, Rashkin M, Bows J, Dunaway K, Slonim A, Washington N, Ferber M, Bolze A, Lu JT. Population genetic screening efficiently identifies carriers of autosomal dominant diseases. *Nat Med* 2020;26:1235–9.
- Abul-Husn NS, Manickam K, Jones LK, Wright EA, Hartzel DN, Gonzaga-Jauregui C, O'Dushlaine C, Leader JB, Lester Kirchner H, Lindbuchler D'Andra M, Barr ML, Giovanni MA, Ritchie MD, Overton JD, Reid JG, Metpally RPR, Wardeh AH, Borecki IB, Yancopoulos GD, Baras A, Shuldiner AR, Gottesman O, Ledbetter DH, Carey DJ, Dewey FE, Murray MF. Genetic identification of familial hypercholesterolemia within a single U.S. health care system. *Science* 2016;354.
- Clinical Principal Committee Standing Committee on Screening. *Population-based screening framework*. Canberra: Australian Government Department of Health, 2020.
- Goranitis I, Best S, Christodoulou J, Stark Z, Boughtwood T. The personal utility and uptake of genomic sequencing in pediatric and adult conditions: eliciting societal preferences with three discrete choice experiments. *Genet Med* 2020;22:1311–9.
- Genioz website. Secondary Genioz website. Available: <https://www.genioz.net.au/>
- Metcalf SA, Hickerton C, Savard J, Stackpole E, Tytherleigh R, Tutty E, Terrill B, Turbitt E, Gray K, Middleton A, Wilson B, Newson AJ, Gaff C. Australians' perspectives on

- support around use of personal genomic testing: findings from the Genioz study. *Eur J Med Genet* 2019;62:290–9.
- 19 Savard J, Hickerton C, Tytherleigh R, Terrill B, Turbitt E, Newson AJ, Wilson B, Gray K, Gaff C, Middleton A, Stackpole E, Metcalfe SA. Australians' views and experience of personal genomic testing: survey findings from the Genioz study. *Eur J Hum Genet* 2019;27:711–20.
 - 20 Middleton A, Milne R, Howard H, Niemiec E, Roberts L, Critchley C, Nicol D, Prainsack B, Atutornu J, Vears DF, Smith J, Steed C, Bevan P, Scott ER, Bobe J, Goodhand P, Kleiderman E, Thorogood A, Morley KI, Participant Values Work Stream of the Global Alliance for Genomics and Health. Members of the public in the USA, UK, Canada and Australia expressing genetic exceptionalism say they are more willing to donate genomic data. *Eur J Hum Genet* 2020;28:424–34.
 - 21 Middleton A, Milne R, Thorogood A, Kleiderman E, Niemiec E, Prainsack B, Farley L, Bevan P, Steed C, Smith J, Vears D, Atutornu J, Howard HC, Morley KI. Attitudes of publics who are unwilling to donate DNA data for research. *Eur J Med Genet* 2019;62:316–23.
 - 22 Briscoe F, Ajunwa I, Gaddis A, McCormick J. Evolving public views on the value of one's DNA and expectations for genomic database governance: results from a national survey. *PLoS One* 2020;15:e0229044.
 - 23 Smit AK, Reyes-Marcelino G, Keogh L, Cust AE, Newson AJ. 'There is a lot of good in knowing, but there is also a lot of downs': public views on ethical considerations in population genomic screening. *J Med Ethics* 2021;47:e28.
 - 24 Bowman-Smart H, Savulescu J, Mand C, Gyngell C, Pertile MD, Lewis S, Delatycki MB. 'Is it better not to know certain things?': views of women who have undergone non-invasive prenatal testing on its possible future applications. *J Med Ethics* 2019;45:231–8.
 - 25 Cernat A, De Freitas C, Majid U, Trivedi F, Higgins C, Vanstone M. Facilitating informed choice about non-invasive prenatal testing (NIPT): a systematic review and qualitative meta-synthesis of women's experiences. *BMC Pregnancy Childbirth* 2019;19:27.
 - 26 Kinsella S, Hopkins H, Cooper L, Bonham JR. A public dialogue to inform the use of wider genomic testing when used as part of newborn screening to identify cystic fibrosis. *Int J Neonatal Screen* 2022;8. doi:10.3390/ijns8020032. [Epub ahead of print: 09 May 2022].
 - 27 Van Steijvoort E, Chokoshvili D, Cannon J, Peeters H, Peeraer K, Matthijs G, Borry P. Interest in expanded carrier screening among individuals and couples in the general population: systematic review of the literature. *Hum Reprod Update* 2020;26:335–55.
 - 28 Van Steijvoort E, Devolder H, Geysen I, Van Epperzeel S, Peeters H, Peeraer K, Matthijs G, Borry P. Expanded carrier screening in Flanders (Belgium): an online survey on the perspectives of nonpregnant reproductive-aged women. *Per Med* 2021;18:361–73.
 - 29 McNeil N, McEachern S, Tranter B. *Australian survey of social attitudes*. Australian Consortium for Social and Political Research Incorporated (ACSPRI), 2021.
 - 30 Website of the Australian Survey of Social Attitudes. Secondary website of the Australian survey of social attitudes. Available: <https://www.acspri.org.au/ausa>
 - 31 Grant P, Langlois S, Lynd LD, Austin JC, Elliott AM, GenCOUNSEL Study. Out-of-pocket and private pay in clinical genetic testing: a scoping review. *Clin Genet* 2021;100:504–21.
 - 32 Maxwell S, O'Leary P. Public funding for non-invasive prenatal testing for fetal aneuploidy - It's time. *Aust N Z J Obstet Gynaecol* 2018;58:385–7.
 - 33 Hui L, Barclay J, Poulton A, Hutchinson B, Halliday JL. Prenatal diagnosis and socioeconomic status in the non-invasive prenatal testing era: a population-based study. *Aust N Z J Obstet Gynaecol* 2018;58:404–10.
 - 34 Sulmasy DP. Naked bodies, naked genomes: the special (but not exceptional) nature of genomic information. *Genet Med* 2015;17:331–6.
 - 35 Garrison Nanibaa' A, Brothers KB, Goldenberg AJ, Lynch JA. Genomic contextualism: shifting the rhetoric of genetic exceptionalism. *Am J Bioeth* 2019;19:51–63.
 - 36 Murray TH. Is genetic exceptionalism past its sell-by date? On genomic diaries, context, and content. *Am J Bioeth* 2019;19:13–15.
 - 37 Tiller J, McInerney-Leo A, Belcher A, Boughtwood T, Gleeson P, Delatycki M, Barlow-Stewart K, Winship I, Otlowski M, Keogh L, Lacaze P. Study protocol: the Australian genetics and life insurance moratorium-monitoring the effectiveness and response (A-GLIMMER) project. *BMC Med Ethics* 2021;22:63.
 - 38 Tiller J, Winship I, Otlowski MF, Lacaze PA. Monitoring the genetic testing and life insurance moratorium in Australia: a national research project. *Med J Aust* 2021;214:157–9.
 - 39 Australian Bureau of Statistics. Snapshot of Australia: a picture of the economic, social and cultural make-up of Australia on census night, 2021. Available: <https://www.abs.gov.au/statistics/people/people-and-communities/snapshot-australia/2021> [Accessed 28 Jun 2022].
 - 40 Domchek SM, Friebel TM, Singer CF, Evans DG, Lynch HT, Isaacs C, Garber JE, Neuhausen SL, Matloff E, Eeles R, Pichert G, Van t'Veer L, Tung N, Weitzel JN, Couch FJ, Rubinstein WS, Ganz PA, Daly MB, Olopade OI, Tomlinson G, Schildkraut J, Blum JL, Rebbeck TR. Association of risk-reducing surgery in BRCA1 or BRCA2 mutation carriers with cancer risk and mortality. *JAMA* 2010;304:967–75.
 - 41 Manchanda R, Saridogan E, Abdelraheem A, Johnson M, Rosenthal AN, Benjamin E, Brunell C, Side L, Gessler S, Jacobs I, Menon U. Annual outpatient hysteroscopy and endometrial sampling (OHES) in HNPCC/Lynch syndrome (LS). *Arch Gynecol Obstet* 2012;286:1555–62.
 - 42 Versmissen J, Oosterveer DM, Yazdanpanah M, Defesche JC, Basart DCG, Liem AH, Heeringa J, Witteman JC, Lansberg PJ, Kastelein JJP, Sijbrands EJG. Efficacy of statins in familial hypercholesterolaemia: a long term cohort study. *BMJ* 2008;337:a2423.