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Measuring physician practice, preparedness and preferences for genomic medicine: a national survey

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Measuring physician practice, preparedness and preferences for genomic medicine: a national survey

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Abstract

Objective: Even as genomic medicine is implemented globally, there remains a lack of rigorous, national assessments of physicians' current genomic practice and continuing genomics education needs. The aim of this study was to address this gap.

Design: A cross-sectional survey, informed by qualitative data and behaviour change theory, to assess the current landscape of Australian physicians' genomic medicine practice, perceptions of proximity and individual preparedness, and preferred models of practice and continuing education. The survey was advertised nationally through 10 medical colleges, 24 societies, 62 hospitals, social media, professional networks and snowballing.

Results: 409 medical specialists across Australia responded, representing 30 specialties (majority paediatricians, 20%), from mainly public hospitals (70%) in metropolitan areas (75%). Half (53%) had contacted their local genetics services and half (54%) had ordered or referred for a gene panel or exome/genome sequencing (E/GS) test in the last year. Two-thirds (67%) think genomics will soon impact their practice, with a significant preference for models that involved genetics services (p<0.0001). Currently, respondents mainly perform tasks associated with pre-test family history taking and counselling, but more respondents expect to perform tasks at all stages of testing in the future, including tasks related to the test itself, and reporting results. While a third (34%) recently completed education in genomics, only a quarter (25%) felt prepared to practice. Specialists would like (more) education, particularly on genomic technologies and clinical utility, and prefer this to be through varied educational strategies.

Conclusions: This survey provides data from a breadth of physician specialties that can inform models of genetic service delivery and genomics education. The findings support education providers designing and delivering education that best meet learner needs to build a competent, genomic-literate workforce. Further analyses are underway to characterise early adopters of genomic medicine to inform strategies to increase engagement.

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Strengths and limitations of this study

- The survey tool is based in behavioural change theory and developed from empirical data to capture patterns of genomic practice and preferences, allowing comparisons across different settings and change over time.
- We employed an extensive, multi-staged and overlapping recruitment strategy at a national level to reach as many Australian medical specialists and trainees as possible.
- We successfully gathered data from over 30 specialties, the broadest sample reported in the literature to date.
- Our sample is still relatively small, and over-represented for older specialists and those working in rural and remote areas, which may influence the findings.
- Our study is the first to investigate the genomics education and training needs and preferences of a national sample of a broad range of medical specialties.

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Introduction

Genomic sequencing is shifting from the realm of research to healthcare.[1] A recent review identified five models for the provision of genetic testing globally, including genetics services led by geneticists, referral by primary-care physicians to genetics services, and medical specialist-led testing.[2] The shortage of a specialist genetic workforce suggests that medical specialist-led testing will be necessary.[3][4] A scoping review of genetic specialist workforces internationally emphasised the need for a medical specialist-led model, noting education as a driver of workforce capacity.[5]

A national alliance of over 80 partner organisations, Australian Genomics, formed in 2016 to conduct research supporting adoption of genomics into Australian healthcare.[6] At that time, microarray analysis and a limited number of single gene tests were reimbursed through the federally-funded Medicare Benefit Scheme (MBS). Genomic sequencing tests were largely available through research studies or patient funding until 2020, when exome/genome sequencing (E/GS) for certain conditions was included on the MBS.[7]

Despite national initiatives driving the use of germline genomic tests by medical specialists not qualified in genetics, there are indications that physicians may prefer to refer to genetics services.[8, 9] Cumulative evidence indicates a lack of physician confidence in genomic medicine and low rates of clinical adoption of genomic testing.[10] Studies investigating practice and preparedness span specialties and countries: Dutch cardiologists,[11] European obstetricians and paediatricians,[12] Wisconsin physicians,[13] British gastroenterologists,[14] Australian intensivists,[8] and neurologists worldwide.[15] However, there are no national studies surveying a range of specialties.

Education strategies have been proposed or implemented to support medical professionals' genomic medicine knowledge and skills.[16, 17] Following medical school training,[18, 19] continuing professional development (CPD), whether accredited or not, aims to supplement knowledge and skills for those already in practice.[20, 21] To inform Australian national strategy and local development of genomics CPD, a needs assessment inclusive of a multiple specialties across diverse contexts is required. We previously reported development of a survey underpinned by qualitative data and an empirically-derived framework in which capability, opportunity and motivation are associated with behaviour change.[22]

Here we describe comprehensive deployment of this survey nationally to multiple medical specialities. We present a snapshot of the current landscape of Australian specialists' genomic medicine practice, perceptions of proximity of genomic medicine and individual preparedness, and preferred models of practice and continuing education.

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Methods

In Australia, medical doctors undertake training within a medical college, such as the Royal Australasian College of Surgeons or the Royal Australasian College of Physicians, to specialise in an area of practice, such as surgery, cardiology, paediatrics, etc. Training typically involves completing three years of basic training ('Basic Trainee') followed by three years of advanced training ('Advanced Trainee'). After successful completion of final examinations, they become a Fellow of the relevant medical college.[23] Medical professionals may work in public hospitals, which are the responsibility of State governments, and/or privately. Patients receive some reimbursement for private consultations and specified pathology tests through the Federal Government's MBS. Here we focus on the non-genetic medical workforce and as such define 'medical specialists' as medical doctors who are trained or in training for a specialty other than clinical genetics. Separate studies have been conducted for genetic specialists (e.g., clinical geneticists and genetic counsellors)[4] and general practitioners (Cusack et al., *Australian Journal of General Practice*, in press), and are ongoing for oncologists.

Details of survey development, domains and the full set of questions have been reported elsewhere.[20, 24] For development of the survey, we defined 'genomic medicine' as the use of testing that investigates many regions of the genome at once, such as gene panels and E/GS, but excluding non-invasive prenatal testing using sequencing technologies. The scope of the survey was testing to investigate genetic conditions. The survey was deployed electronically from February to September 2019 using REDCap (Research Electronic Data Capture) software hosted at the Murdoch Children's Research Institute.[25]

This project received ethics approval from the University of Melbourne, Melbourne, Australia (HREC number: 1646785.10). Respondents provided consent by completing the initial screening and consent question.

Recruitment

Inclusion criteria: medical specialists were eligible to complete the survey if they had commenced or completed their specialist training and were currently practising clinically in Australia.

Recruitment was staged through:

- Relevant medical colleges (Mar–Jun 2019) and societies/associations (Apr–Jun 2019).

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- Hospitals (Jun–Oct 2019). 132 hospitals were identified from the 'MyHospitals' search tool on the Australian Institute of Health and Welfare website[26] to represent both public and private hospitals in metropolitan, regional and rural settings across all Australian states.
- Social media (Jun–Jul 2019). Three tweets were posted on the Australian Genomics Twitter account (https://twitter.com/AusGenomics) over 10 business days, then this process was repeated twice, with approximately one week between each cluster of tweets. Content referenced specific survey questions or preliminary data to pique interest of potential participants. For example, *'Early survey results suggest that even though medical specialists are ordering #genomictests for their #patients, many don't feel #prepared for #genomicmedicine. We want to know how you feel [LINK]'* or *'Do you feel ready for #genomics in #clinicalpractice? We want to hear from Australian medical specialists [LINK]'*.
- Investigator networks of national and state-based genomics initiatives, Australian Genomics and Melbourne Genomics (Jul 2019).

Medical colleges, societies and hospitals circulated information about the study to their membership or staff using regular communication channels, e.g., newsletters, e-bulletins, emails, etc. Information was circulated up to three times per organisation, dependent on advertising charges, perceived responder burden and/or internal timelines. The information included a brief description of the study, ethics approval and a link to access the online survey. Recruitment also included professional networks and snowball sampling throughout, with all contacts asked to retweet Australian Genomics tweets if possible. All respondents were asked to share the survey with relevant colleagues.

Data cleaning and analysis

Data were exported to, cleaned and then analysed in Stata 16.0. Cleaning involved removing surveys completed by ineligible respondents or surveys with no data beyond demographic questions. For analysis, career stage was grouped into Basic Trainee, Advanced Trainee or Fellow, as defined above. All categorical questions included an open-ended text option for 'Other'; qualitative data provided for these questions were reviewed by three researchers (AN, EK, MJ) and recoded into existing response categories if possible (see **Supplementary Table S1** for examples). Representative quotes are provided in **Supplementary Table S2** for illustrative purposes where they enhance the understanding of the quantitative results.

Descriptive and inferential statistics were used to analyse the data, including two-sample tests of proportions, chi-square or Fisher's exact tests as appropriate to data characteristics. A p value of <0.05 was considered significant. When determining representativeness of the sample, data were

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referenced against Medical Board of Australia Registrant data,[27] the National Medical Training Advisory Network, and the National Health Workforce Dataset.[28]

Patient and public involvement

There was no patient or public involvement in this research.

Results

Recruitment and response rates

As shown in **Figure 1**, recruitment strategies were staggered and overlapping from March to October 2019. All 10 Australian medical colleges and 24 of 55 medical societies/associations approached agreed to advertise the survey. Of 132 health networks[1] and hospitals contacted,[29] 62 agreed to advertise the survey (67.6% of metropolitan hospitals and 42.9% of remote hospitals), which was subsequently shared with staff at a total of 74 hospitals. There were an estimated 37,000 trainees and fellows in our target specialty audiences at the time of the survey.[27] However, using diverse recruitment approaches that could target one individual in several ways and at several time points meant that it was not possible to determine how many medical specialists were aware of the survey during the recruitment period.

Sample characteristics and representativeness

Of 617 attempts at survey responses, 54 did not meet the inclusion criteria and 154 did not complete any questions beyond consent to participate. A total of 409 responses were therefore included in analyses. Totals differ across questions due to opportunity to provide more than one response, missing data or attrition; where this has occurred, the denominator has been described. **Table 1** presents respondent demographics compared with reference data from the Medical Board of Australia,[27] the National Medical Training Advisory Network[30, 31] and the National Health Workforce Dataset.[28] Our sample had slightly less males (p=0.039), was under-represented for 25–34 year olds (p<0.0001), and over-represented for 55–64 year olds (p<0.0001). As would be expected from this age bias, there was a smaller proportion of Basic and Advanced Trainees than expected from the reference data and a larger proportion of Fellows (p<0.0001). Our sample was broadly representative of primary work locations of medical specialists across Australia. Of the eight Australian states and territories, one was over-represented (Australian Capital Territory; p<0.0001) and two were under-represented (South Australia; p=0.028); Western Australia; p=0.032). Although

[1] Health networks are functional or geographical groups of Australian public hospitals defined by the relevant State Government.

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three-quarters of respondents worked in a major city, those working in remote regions were significantly over-represented in our sample (p=0.0018). The majority of respondents were primarily employed at public hospitals or healthcare providers. A quarter of respondents had been involved in a genomics research project in the last 5 years (n=96, 24.7%). Of these, respondents were involved in clinical (83.3%), laboratory (49.0%), bioinformatics (15.6%) and/or social science (6.3%) projects. Only 7.2% of respondents indicated that they were affiliated with any state- or federally-funded genomic health alliances.

Figure 2 describes proportions of respondent specialties, compared with the proportions expected from reference data.[27] The largest group of respondents were physicians, totalling 289 (70.7%) responses. Our sample was representative of most specialties with some exceptions: there were more haematologists (p=0.01), paediatricians (p<0.0001), infectious disease (p<0.0001) and palliative medicine physicians (p<0.0001), and fewer anaesthetists (p=0.002), psychiatrists (p<0.0001), surgeons (p=0.0001), general medicine physicians (p=0.01), and immunology/allergy physicians (p<0.0001).

Current practice in genomic medicine

Respondents (n=387) answered a series of questions about their current practice in genomic medicine. Just over half of respondents had contacted their local genetics service in the last 12 months (n=203, 52.5%), although this was relatively infrequent, with a third of these 203 respondents indicating this was once or twice in the last 12 months (36.6%). The main reasons for contacting genetics services included: seeking information about a suspected genetic condition (48.0%), advice on how to refer a patient (42.6%) and choosing which genetic or genomic test to order (38.1%). Of those who had not contacted clinical genetics, the majority indicated that this was because they had not yet needed advice (73.5%).

Over half of respondents (n=208, 53.9%) had engaged in genomic sequencing testing in the last 12 months by either ordering a gene panel or E/GS, or referring a patient to a genetics service for those tests. Nearly a third of respondents (n=121, 31.3%) had ordered at least one of these tests, with 29.0% (n=112) ordering a gene panel and 13.0% (n=50) ordering E/GS. When asked about frequency of ordering each test in the previous year, the most common response was once or twice for both gene panels (n=42/112, 37.5%) and E/GS (n=23/50, 46.0%). In contrast, 112 respondents (29.0%) had ordered a microarray in the previous year, most commonly monthly (n=41/112, 36.6%). Funding for tests varied (**Supplementary Table S4**), with microarray tests often funded by the MBS, gene panel tests by the institute/hospital, and E/GS tests by research grants. Overall, 63.3% of respondents

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(n=245/387) had engaged in genetics/genomics in one or more ways: contacting their genetics service, or ordering or referring for a microarray, gene panel or E/GS test.

Respondents were asked to reflect on their confidence about genomic concepts and skills (**Figure 3**). Medical specialists reported the highest level of confidence when taking a family history to elicit information about genetic conditions, and lowest for knowledge about genomics. There was greatest variation in their confidence to make decisions based on genomic information (IQR=2,7).

Current practice compared with expected future practice in genomic medicine

Overall, two-thirds of respondents think genomics will impact their practice in the next two years (n=199/298, 66.8%). Of those medical specialists who think their practice will be impacted, they anticipate it will change the way they manage patients (n=177/199, 88.9%) and practice medicine (n=151/199, 75.8%), more so than impact on workload (n=86/199, 43.2%). For respondents who felt genomics would not impact their practice in the next two years (n=50/298, 16.8%), open-text comments (n=47) suggested this was due to perceived relevance to their specialty, timing and/or pragmatic issues of service delivery (see **Supplementary Table S2** for examples). The remaining 49/298 (16.4%) respondents were 'unsure'.

More respondents currently perform clinical activities before and after E/GS testing (**Figure 4**, n=314, 10.6% to 80.3% across these steps) than are involved in non-clinical activities directly related to the test itself (6.7% to 17.0%). Similar patterns were seen in their expectations of the steps they would perform in the future if they had adequate education, training and support: 40.8% expect to perform all pre-test steps and 23.1% all post-test steps, while 40.3% do not expect to perform any steps relating to the test itself. Notably, there were significant increases in the proportion of specialists who expect to perform each step in future practice ($p \leq 0.004$ across all steps), with the exception of eliciting phenotypic information about genetic conditions as part of a family or medical history for the purpose of assisting with variant interpretation, which was already high (80.3% current, 83.4% future; $p=0.3$).

Preferred future models for delivering genomic medicine

When reflecting on preferred models for delivering genomic medicine in the future, the model most often selected by respondents was referral to their local genetics services to initiate testing and discuss results (**Table 2**). This was the case for both inpatient and outpatient settings. The second most preferred model was delivering testing with support from a local genetics service. The type of support included: advice on whether testing is appropriate (60.0% for inpatients; 66.7% for outpatients); interpreting results (72.0% for inpatients; 75.0% for outpatients); discussing results with families (60.0% for inpatients; 70.8% for outpatients); or follow-up genetic counselling (80.0%

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for inpatients; 83.3% for outpatients). A small number expect to initiate genomic testing themselves with no support from a local genetics service, while some respondents also indicated they did not expect to see patients who would benefit from genomic testing. Overall, significantly more respondents preferred a model that includes involvement of genetics services (for support or referral) than a model of initiating testing themselves: inpatients, 62.4% (95%CI 54.8–69.5) compared with 2.3% (95%CI 0.6–5.6), $p<0.0001$); outpatients, 69.7% (95%CI 62.8–76.1) compared with 4.1%; (95%CI 1.8–7.9, $p<0.0001$).

Preparedness for genomic medicine and preferences for future education

While a third (n=92/273, 33.7%) of respondents had completed education in genomics in the past year, only a quarter (n=73/297, 24.6%) felt prepared to use genomic sequencing testing in their practice. Comments from those who did not feel prepared or were 'unsure' (n=210 combined) primarily suggest this could be addressed through genomics education and training (**Supplementary Table S2**). Forty-two per cent of respondents felt that improved genomic knowledge may alter their clinical practice (n=115/273, 42.1%) but a similar proportion were 'unsure' (n=114/273, 41.8%).

When asked about preferred modes of learning genomics, most respondents (n=250/273; 91.6%) endorsed at least three different modes (**Table 3**). The two most commonly preferred– CPD activities and learning from peers–were also the two most commonly-used currently. In contrast, reading specialty texts was the third most common way of learning about genomics currently, but the eighth preferred. Respondents indicated a preference for genomics education incorporated into their usual work activities (e.g., internal workplace seminars, departmental presentations and clinical meetings).

Despite three-quarters of respondents reporting they had already learned basic concepts of genomics (**Table 4**; n=271), a similar proportion still requested this topic for future education. Six topics were endorsed by over 80% of respondents including current and emerging applications in genomic medicine, the clinical utility of different tests and topics around patient management. Again, respondents could select more than one topic, with 92.3% indicating they wanted to learn about at least five topics in the future, and 26.4% selecting all topics. Nearly two-thirds of respondents indicated they wanted to learn about communication skills with patients, with comments throughout the survey suggesting a need for training in how to explain genomic testing concepts, implications and results to patients.

Discussion

This paper provides a baseline snapshot of Australian non-genetic medical specialists' practice of genomic medicine and perspectives at a point in time before E/GS was widely available to them as a

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funded clinical test. In 2019, 60% of all 409 survey respondents reported some form of interaction with genetics services or genetic/genomic testing. The test ordered most frequently was a microarray, but more than a quarter of all survey respondents indicated they had ordered a genomic sequencing test in the past twelve months. Respondents anticipated their practice would change in the near future, with significantly more respondents expecting to be involved in activities relating to E/GS in the next two years than currently. Consistent with discipline-specific studies from other countries,[13, 15, 32-34] we found the majority of respondents in our survey did not feel prepared to use genomic sequencing testing in their practice and over two-thirds preferred a model that involved genetics services in some way. Our study extends existing literature by providing greater depth of insight into the education needs and preferences of a broad range of medical specialists.

A strength of this snapshot is the use of a survey tool[24] grounded in a theoretical model. The COM-B model posits that behaviour is influenced by capability, opportunity and motivation.[22] *Opportunity* is clearly impacted by the availability of funded genomic tests. The test usage reported by respondents in this study reflects the availability of MBS reimbursement. For instance, microarrays have been established as MBS-reimbursed pathology tests for a decade. At the time of this survey E/GS tests were not reimbursed by the MBS. The relatively lower proportion of respondents who had ordered these tests used a variety of other funding mechanisms, most commonly hospital or research funds, and noted availability of funding as an influence when ordering genomic tests in the future. Since this study was completed, MBS now reimburses genomic sequencing tests for some clinical indications when ordered by paediatricians, enhancing their opportunity to use genomic testing in their clinical practice. It is anticipated that reimbursement for other clinical indications (and medical specialties) will follow in the future.

Respondent's perceived *capability* to respond to the availability of funded tests, however, is limited. Currently, respondents lack confidence in their knowledge, ability to explain genomic concepts and make decisions based on genomic information. This may explain their desire to practice collaboratively with clinical geneticists and genetic counsellors to varying extents. It is possible that these preferences could change as their capability (and confidence in their capability) develops with greater opportunity, experience and learning.[22, 24] Education and training was certainly seen as a solution to feeling unprepared by a substantial proportion of respondents in this study, as also observed by others.[35] In the past two years, continuing education for Australian medical specialists has been produced locally at an introductory level by a number of initiatives and organisations.[2] More is clearly needed: survey respondents are very interested in genomics

[2] For example, https://elearning.racp.edu.au/course and http://learn-genomics.org.au/.

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education and nearly all respondents selected five or more of the topics that they wished to learn about. This is perhaps unsurprising given their perception of being unprepared and expectation of a greater role in the near future, provided they receive adequate support and education. The most popular education topics were related to pre-test aspects of testing, such as identifying appropriate patients to refer and knowing how to refer, consistent with the significantly stronger preference for a genetics-led model for genomic medicine. Educational strategies will need to consider both the diversity of respondents' preferences for modes of learning and timing with respect to clinical implementation. Not only will timing affect perceived relevance to clinical practice, and therefore motivation to learn,[36] but preferences and needs may evolve as implementation progresses.

Our rigorously-developed survey tool can be deployed again in the future to capture changes in workforce practice and preferences over time. Wider use of the tool can provide a basis for documenting and comparing data across specialties and countries. Our experience with deployment of the survey may assist in this regard. Although it is not possible to determine which recruitment approach was most successful because of overlapping timeframes, increases in the number of responses to our survey coincided with recruitment approaches using social media, internal hospital communication channels and investigator networks. This may reflect increasing professional use of social media by medical specialists[37] and greater attention to emails from their employing hospital than a medical college or society. It may also explain the higher representation of Fellows and older specialists in our sample, as trainees were often not on staff mailing lists used by hospitals to distribute the survey. Our staggered and comprehensive recruitment approach did achieve a strong response from rural and remote medical specialists, who are often missed in research. Under- or over-representation of medical specialists in some Australian states may be due to differences in governance (hospital and/or research) and site-based communication policies that limited dissemination of the survey. While it is not possible to determine the response rate, our sample represents 1.2% of 37,000 medical specialist registrants with the Medical Board of Australia[27] and is within the range achieved in similar surveys of American physicians that also recruited participants through medical societies and associations (0.6–2.6%).[13, 38-40]

This national snapshot of medical specialists' current practice in genomic medicine provides the first detailed insight into the continuing genomics education needs of a broad group of subspecialties. It includes some specialties, such as emergency medicine, palliative medicine and infectious disease, for the first time internationally. Those currently involved and/or most interested in genomic medicine may have been more likely to respond, but this would mean our respondents are also those likely to undertake continuing education and engaging with genomics. Consequently, our results can assist genomics education providers to best meet learner needs and develop a

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competent, genomics-literate workforce, as well as genetic and other clinical services implementing models for genomic medicine delivery. Further data analysis will provide insights into any differences between early adopters of genomic medicine and those who have not yet engaged, enabling the development of targeted, tailored genomics education and other capability-building strategies for optimising the adoption of genomics by medical specialists.

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Competing interests

All authors declare no completing interests.

Author contributions

AN and EK were involved in all stages of this work and manuscript preparation. BM, SM and CG were involved with all stages except data acquisition. MJ analysed the qualitative data responses and was involved in manuscript preparation. All authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. We also thank Rigan Tytherleigh for her advice on statistical analyses. We thank Erin Crellin for her assistance in interpretation of the COM-B model and behaviour change framework.

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Figure captions

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Figure 3: *Average confidence about genomic concepts and skills on a scale of 1 'Not at all confident', 5 'Neutral' to 10 'Very confident' (n=273). Boxes represent the interquartile ranges with minimum and maximum value; medians are shown as white bars.*

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Tables

Table 1: *Description of the sample and representativeness (n=409).*

Characteristic	Respondents		Reference data		
	n (%)	95%CI	N (%)	95%CI	*p*
Gender[1]					
Male	213 (52.1)	47.2–56.9	61,700 (57.1)	56.8–57.4	0.039
Female	185 (45.2)	40.4–50.1	46,281 (42.9)	42.6–43.2	0.330
Prefer not to answer	11 (2.7)	1.5–4.8	–	–	–
Age[1]					
≤24 years	–	–	398 (0.4)	–	
25–34 years	29 (7.1)	4.6–9.6	26,827 (24.8)	24.6–25.1	<0.0001
35–44 years	114 (27.9)	23.5–32.2	28,431 (26.3)	26.1–26.6	0.4794
45–54 years	123 (30.1)	25.6–34.7	22,415 (20.8)	20.5–21.0	<0.0001
55–64 years	103 (25.2)	21.2–29.6	18,060 (16.7)	16.5–17.0	<0.0001
≥65 years	40 (9.8)	7.2–13.1	11,852 (11.0)	10.8–11.2	0.4398
Trainee level[2]					
Basic Trainee	9 (2.2)	1.3–4.6	5,858 (12.1)	11.8–12.4	<0.0001
Advanced Trainee	18 (4.4)	2.6–6.7	8,890 (18.3)	18.0–18.7	<0.0001
Fellow	382 (93.4)	89.9–95.0	33,749 (69.6)	69.2–70.0	<0.0001
Australian state or territory[1,3]					
Australian Capital Territory	28 (6.9)	4.4–9.3	702 (1.9)	1.8–2.0	<0.0001
New South Wales	119 (29.1)	24.7–33.5	11,566 (31.2)	30.7–31.7	0.3622
Northern Territory	8 (2.0)	0.6–3.3	373 (1.0)	0.9–1.1	0.0568
Queensland	75 (18.3)	14.8–22.4	7,320 (19.7)	19.3–20.1	0.4777
South Australia	20 (4.9)	2.8–7.0	2,896 (7.8)	7.5–8.1	0.0283
Tasmania	13 (3.2)	1.5–4.9	759 (2.0)	1.9–2.2	0.1091
Victoria	119 (29.1)	24.7–33.5	9,952 (26.8)	26.4–27.3	0.3063
Western Australia	26 (6.4)	4.0–8.7	3,510 (9.5)	9.2–9.8	0.0324
Primary work location[3,4]					
Major city	306 (75.0)	70.6–79.0	72,304 (79.2)	78.9–79.4	0.0391

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Characteristic	Respondents		Reference data		
	n (%)	95%CI	N (%)	95%CI	*p*
Inner regional	59 (14.5)	11.4–18.2	12,422 (13.6)	13.4–13.8	0.6127
Outer regional	31 (7.6)	5.4–10.6	5,299 (5.8)	5.7–6.0	0.1216
Remote	10 (2.5)	1.3–4.5	865 (1.0)	0.9–1.0	0.0018
Very remote	2 (0.5)	0.1–2.0	376 (0.4)	0.4–0.5	0.8048
Primary employer[5]					
Public hospital or healthcare provider	288 (70.4)	65.8–74.7			
Private hospital or healthcare provider	17 (4.2)	2.6–6.6			
Self-employed/ private practice	83 (20.3)	16.7–24.5			
Other (government, research institute, etc.)	21 (5.1)	3.4–7.8			

Reference data were: [1] Registration Data Table 2019 [27]; [2] Medical Education and Training in Australia 1st Edition report 2017 [31]; [3]n=408 for state and location; [4] Medical Workforce 2016 Factsheet [28]; [5] There were no comparable reference data for this category.

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Table 2: *Medical specialists' preferred models for delivering a genomic sequencing test in inpatient and outpatient settings (n=218).*

	INPATIENT n=178[1]		**OUTPATIENT n=195[1]**	
	n (%)	**95%CI**	**n (%)**	**95%CI**
You **initiate** testing and discuss results with patients/families	4 (2.3)	0.6–5.6	8 (4.1)	1.8–7.9
You initiate testing and discuss results with patients/families, with **support** from a clinical genetics team as needed	43 (24.2)	18.15–31.1	49 (25.1)	19.2–31.8
You **refer** to a clinical genetics team to initiate testing and discuss results with patients/families	68 (38.2)	31.0–45.8	87 (44.6)	37.5–51.9
You **do not see**, and do not expect to see, patients who would benefit from genomic testing	33 (18.5)	13.1–25.0	23 (11.8)	7.6–17.2
Unsure at this stage	30 (16.9)	11.7–23.2	28 (14.4)	9.8–20.1

[1] *A total of 218 respondents completed this question, indicating a preference for either the inpatient or outpatient setting, or both.*

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Table 3: *Current and preferred modes of learning about genomics (n=273).*[1]

Mode of learning about genomics	Currently use (%)	Prefer to use (%)
Continuing Professional Development/Continuing Medical Education activities	51.8	79.8
Consult colleagues and peer	54.0	79.4
Internal workplace specialty seminars, conferences or similar	34.1	74.0
Departmental presentations	35.8	72.0
Clinical meetings	34.8	71.4
External specialty seminars, conferences, etc.	36.0	67.3
Internal workplace genetic or genomic seminars, conferences, etc.	24.9	66.3
Reading specialty texts	48.2	63.2
Online webinars, courses, MOOCs, etc.	15.8	59.6
Certification/fellowship activities	34.4	56.4
External genetic or genomic seminars, conferences, etc.	18.4	50.0
Small group tutorials	8.1	44.9
Study days at place of employment	12.5	41.9
Genomic research project	17.6	32.6
Time in a service or laboratory with genomics expertise	6.2	17.6
Mass media	12.5	14.0
Social media	7.4	11.0
Other (e.g., fact sheet written by geneticist)	0.0	0.4

[1] *Respondents could select more than one mode.*

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Table 4: *Topics relevant to genomics medicine that medical specialists have learnt about or would like to learn (more) about (n=271).*[1]

Education topic	Have learnt about (%)	Want to learn (more) about (%)
Genetic/genomic knowledge		
Basic concepts	77.5	77.1
Disorders and diseases	74.2	83.4
Current applications in genomic medicine	60.9	88.9
Emerging applications in genomic medicine	55.7	87.8
Genetic/genomic testing and technology		
Types of genetic tests	64.9	76.4
Types of genomic tests	58.7	77.1
Applications of somatic genomic tests	45.4	75.6
Applications of germline genomic tests	37.6	69.7
Clinical utility of tests	57.6	88.6
Classification of genomic data during testing	41.3	67.9
Limitations of testing	50.2	79.7
Pre- or post-test aspects		
Recognising patients who may benefit from genomic testing	60.9	83.0
Communication skills with patients	70.8	63.1
Performing genetic risk assessments	57.6	67.5
Referring appropriately for a genomic test	59.4	81.5
Requesting a genomic test for a patient	53.9	70.8
Interpreting genomic test results	52.0	74.9
Cascade testing	53.9	68.6
Ethical, legal and social implications		
Ethical implications	59.0	75.6
Legal implications	52.4	75.3
Psychosocial implications	57.2	74.9

[1] *Respondents could select more than one topic.*

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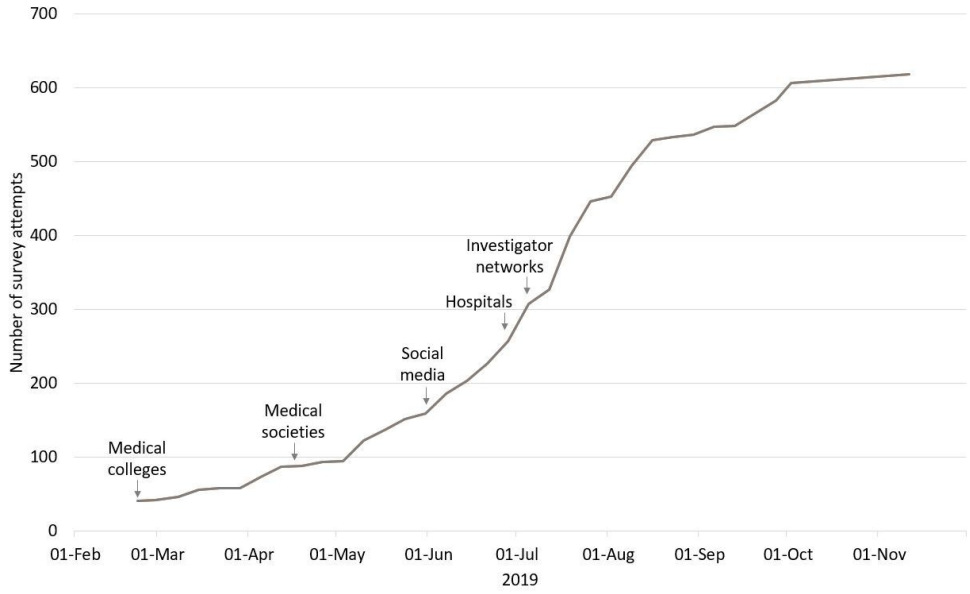

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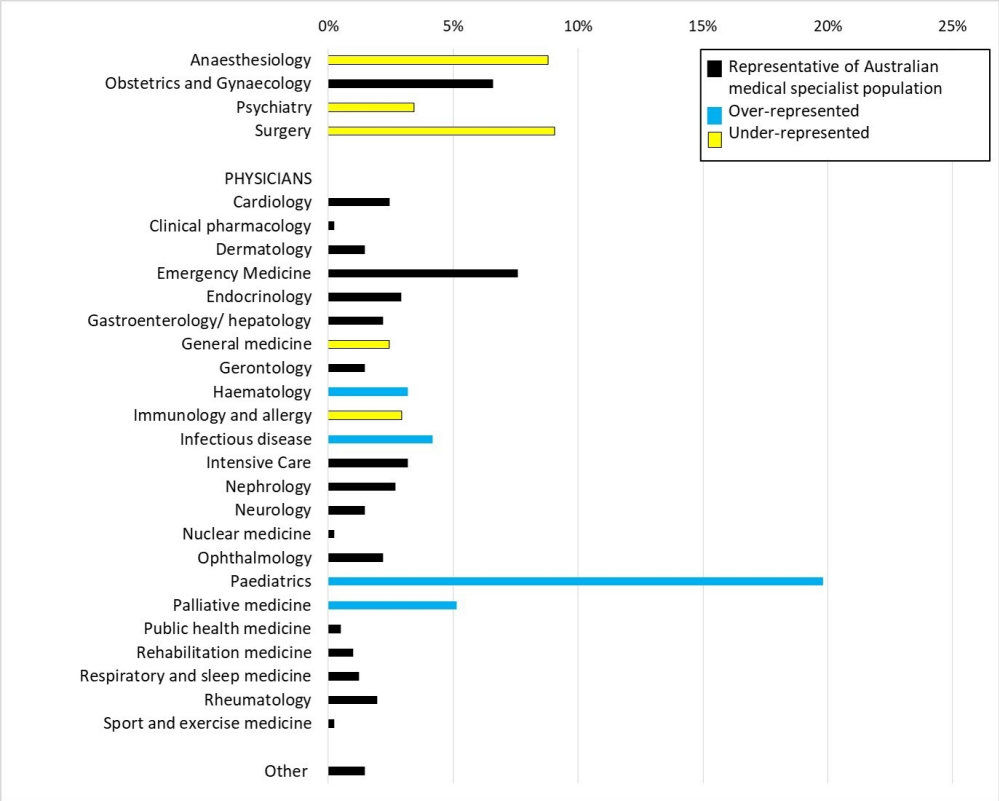

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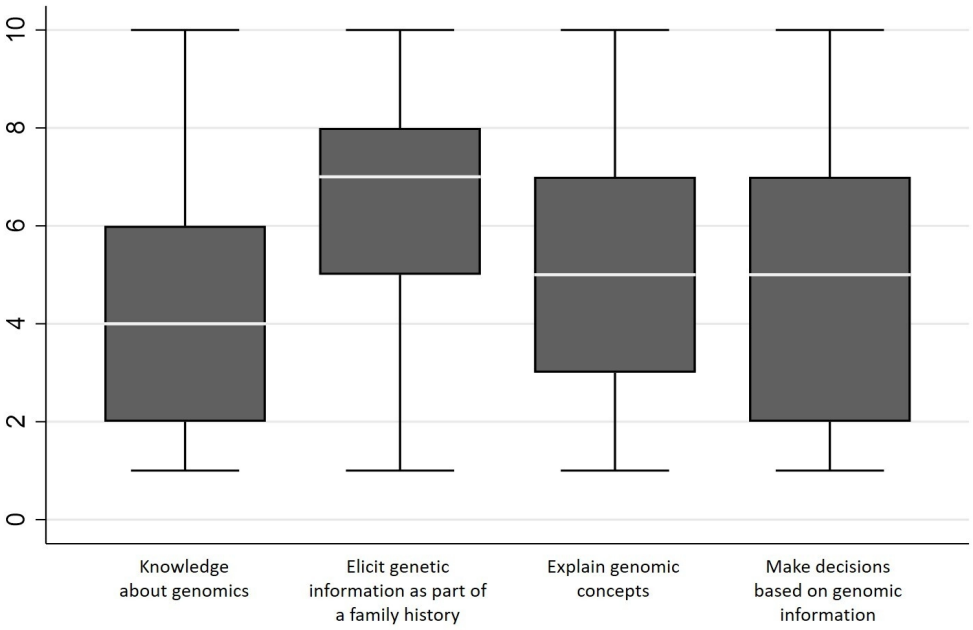

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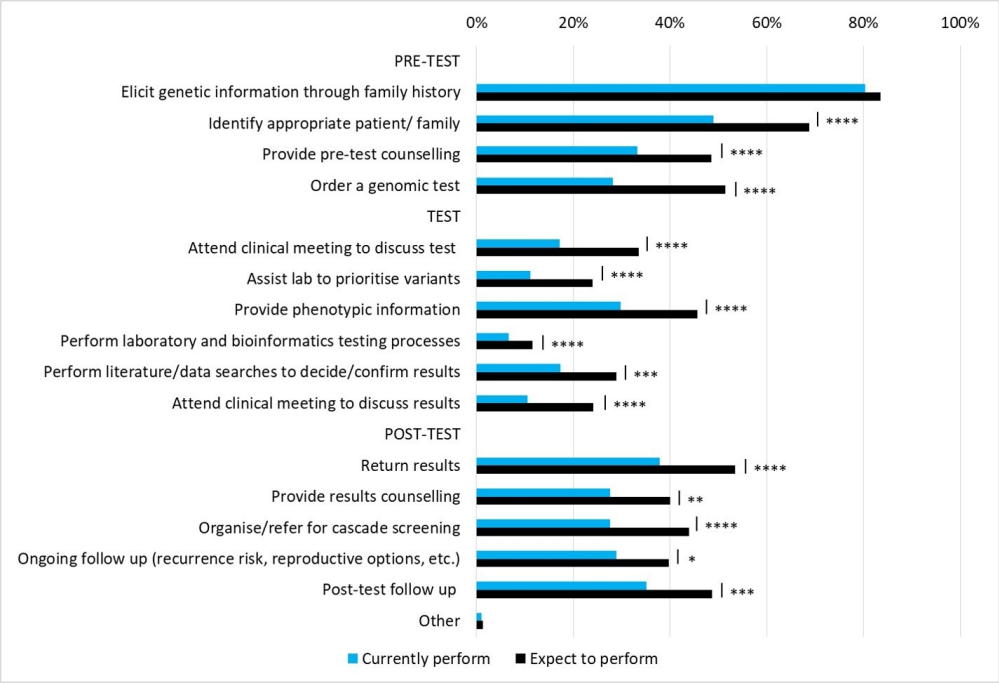

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Nisselle, King et al. Measuring physician practice, preparedness and preferences for genomic medicine: a national survey.

SUPPLEMENTARY MATERIALS

Table S1. *Examples of recoded open-text responses where a respondent selected 'Other (please specify……)' for a categorical question.*

Question	Open text response [ID, specialty]	Recoded category
[If contacted clinical genetics team or service in last 12 months]: Why did you contact your clinical genetics team or service? [a] Information about a suspected genetic condition [b] Advice on what type of genetic or genomic test to order [c] Advice on how to refer the patient to my clinical genetics team or service [d] Assistance with genetic counselling before the test [e] Assistance with genetic counselling after the test [f] Other (please specify)……	*"Referral"* [135, surgery]	[c]
	"Facilitating genomic testing so that genetic counselling can be given to patient before test" [145, paediatrics]	[d]
[If did not contact clinical genetics team or service in last 12 months]: Why haven't you contacted your clinical genetics team or service? [a] Genetics and genomics are not relevant to my practice [b] I have not yet needed advice from a clinical genetics team or service in my practice [c] I can manage my patients without advice from a clinical genetics service [d] I'm not sure how to contact my clinical genetics team or service [e] I do not have access to a clinical genetics team or service [f] Other (please specify)……	*"My cohort of patients generally do not need genetic service input"* [129, gerontology]	[a]
	"We do some of this inhouse" [282, general medicine]	[c]
Below is a list of some of the steps involved in genomic sequencing testing from pre-test to post-test *[see **1 Full** question provided in **Table S1** ; [2] following the question on confidence in four genomic knowledge and skills areas, presented in **Figure 1**; [3] following the question on steps involved in genomic sequencing*	*"Going over letters and reports from genetics, explaining things again in context"* [221, paediatrics]	[k]
	"I continue to see patients after their diagnostic test, which hopefully occurs as part of the	[n]

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Question	Open text response [ID, specialty]	Recoded category
testing, presented in Error! Reference source not found. and ***Table S1.*** ** Definitions were provided for these terms*	*evaluation of their condition"* [3, gerontology]	
Table S3*].* Please indicate which steps you currently perform and which ones you expect to perform in the future if you had adequate education, training and support. If you selected "Other" step, please specify.		
What is/would be your preferred model for delivering a genomic sequencing test in an outpatient setting in your clinical practice, assuming you have appropriate education, training and funding? [a] You initiate testing and discuss results with patients/families [b] You initiate testing and discuss results with patients/families, with support from a clinical genetics team as needed [c] You refer to a clinical genetics team to initiate testing and discuss results with patients/families [d] You do not see, and do not expect to see, patients who would benefit from genomic testing [e] Unsure at this stage [f] Other (please specify)……	*"Not relevant to my specialty"* [140, palliative medicine] *"Same as for inpatient"* [109, palliative medicine; selected [b] for Inpatient response]	[d] [b]
[If selected 'yes' to genomics will impact practice within two years]: What areas will be impacted? [a] The way I practice medicine [b] My workload [c] Patient management [d] Other (please specify)……	*Clinical outcome and prognostications* [123, intensive care]	[c]
[If selected 'yes' to attending genomic professional development education or training in past year]: Was this: [a] In-house (internal) program/s [b] External program/s [c] Online training (webinar, MOOC, etc.) [d] Other (please specify)……	*"Recent commencement of multidisciplinary meeting"* [416, cardiology] *"International Clinical Cardiovascular Genetics conference"* [430, paediatrics]	[a] [b]

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Table S2. *Illustrative quotes from open-text survey comments.*

Domain	Quote
Current practice compared with future practice in genomic medicine *Q: Do you think genomics will impact your practice in the next 2 years?*	
Expect genomics *will* impact practice in next two years	*"Becoming increasingly available and of measurable significance"* [513, surgery] *"I expect it [genomics] will increasingly impact on the practice of medicine in terms of diagnoses, prognoses and treatment"* [281, paediatrics] *"Increased patient requests"* [271, obstetrics and gynaecology]
Expect genomics will *not* impact practice in next two years	*"Emergency department have more important competing interests in treatment delivery to patients"* [383, emergency medicine] *"Timeframe remains too short to see this implemented in a regional area"* [535, anaesthesiology]
Preferred future models for delivering genomic medicine *Q: What is/would be your preferred model for delivering a genomic sequencing test* in your clinical practice, assuming you have appropriate education, training and funding?*[1]	
Referring to genetics services to initiate testing and discuss results	*"For my patients and practice, having an accessible [genetics] clinic for this would be best. I would be very keen to be involved as far as possible, but do not have time to keep up with this rapidly developing field. I would like to be invited to my patients' MDT [multidisciplinary team] discussions. That way I am involved, and have the knowledge to answer follow-up and clarification questions. It would also be a way to increase my knowledge"* [100, nephrologist]
Delivering testing with support from genetics services	*"[Genetics support for both inpatients and outpatients] would streamline the process, improve access and possibly reduce Clinical Genetics load by filtering patients and families I can manage while they still see the patients or results beyond my expertise"* [220, paediatrics, community child health] *"We (clinicians) may be more familiar with the disease phenotype than the Genetics team"* [33, immunopathology] *"Clinicians should be able to initiate testing but will need support with interpretation and counselling, particularly initially until genomic medicine is core practice"* [350, palliative medicine]
Initiating genomic testing themselves with no support from genetics	*"I expect to be able to manage simpler conditions/results, with access to more specialist input when needed"* [129, gerontology]
Will not see patients who would benefit from genomic sequencing tests	*"Relevance to decision making in real time"* [459, emergency medicine] *"Not sure of any relevance to my practice"* [541, anaethesiology]

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Domain	Quote
Preparedness for genomic medicine and preferences for future education	
Q: Do you feel prepared to use genomic sequencing testing in your practice?*	
	"I have little to no training in genetics and genomic medicine. We had a total of 4 genetics lectures at medical school, and there is limited assessment of genetics/genomics in the [college fellowship examination]. Genomic testing is not routinely used in our practice" [73, intensive care]
	"My knowledge of this whole area is woefully inadequate. I can cope with karyotype analysis and testing for CF [cystic fibrosis]. I can also discuss prenatal diagnosis options, PGT-A [pre-implantation genetic testing] and expanded carrier testing but that's about it…… It clearly will be an important part of medical practice in the future" [213, obstetrics and gynaecology]
	"I'm happy to do [genomic testing] but need training." [342, surgery]
	"Need further information, education on who would best benefit from this test, how to consent for it and then how to interpret results" [414, general paediatrics]
Preferences for learning about genomics	
Q: What would help improve your confidence?[2]	*"Further training in counselling [would improve my confidence]–in ability to explain concepts and then clinical implications and follow-on from this"* [27, paediatric neurology]
Q: Please explain why you do not expect to perform the selected steps [involved in genomic sequencing testing]*[3]	*"Would welcome some education on use of these tests in orthopaedics"* [391, surgery]

*[1] Full question provided in **Table S1** ; [2] following the question on confidence in four genomic knowledge and skills areas, presented in **Figure 1**; [3] following the question on steps involved in genomic sequencing testing, presented in Error! Reference source not found. and **Table S1.***

** Definitions were provided for these terms*

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Table S3. *The full wording of each step involved in genomic testing as presented in the survey.*[1]

Pre-test
[a] Eliciting information about genetic conditions as part of a family or medical history
[b] Identifying a patient suitable for a genomic test
[c] Pre-test counselling to assist in making an informed decision, e.g., genetics, test limitations, variants of uncertain/unknown significance*, incidental/secondary findings, unexpected non-paternity or consanguinity
[d] Ordering a genomic test for a patient
Test
[e] Attending multidisciplinary team meeting to discuss the genomic test (e.g., intake meeting)
[f] Assisting the lab to narrow down the genes of interest (creating a gene list to prioritise variant analysis)[2]
[g] Providing phenotypic information to the lab to prioritise variant analysis
[h] Laboratory and bioinformatics testing processes[2]
[i] Searching the literature and databases for evidence of variant pathogenicity*[,2]
[j] Attending a multidisciplinary team meeting to discuss variant prioritisation*, interpretation and classification*
Post-test
[k] Provide test results to patients/ families
[l] Provide genetic counselling to patients/families, e.g., explain variants of uncertain/unknown significance*, incidental/secondary findings, unexpected non-paternity or consanguinity
[m] Organising/ referring for further testing of family members if required, e.g., cascade testing or segregation studies
[n] Ongoing management of the patient, e.g., clarify recurrence risk and discuss reproductive planning options
[o] Post-test follow up of patient to check understanding of result/ ask any additional questions
[p] Other (please specify)……

[1] The survey is available as supplementary material in [24]; [2] These steps are considered non-clinical, i.e., laboratory;

** Definitions were provided for these terms*

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Table S4. *Participant-reported funding for genomic tests ordered in the past year.*[1]

	Microarray n=112	**Gene panel** n=112	**Exome/genome sequencing** n=50
Medicare Benefit Scheme	48.2%	17.0%	2.0%[2]
Institute/hospital	41.1%	52.6%	44.0%
State government	13.4%	17.0%	12.0%
Research grant	2.7%	11.6%	60.0%
Patient	12.5%	24.1%	4.0%
Unsure	11.6%	8.0%	6.0%

[1] *Respondents could select more than one funding source per test type.*

[2] *At the time of the survey the MBS scheme did not fund E/GS, so this response (n=1) is incorrect.*

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Reporting checklist for cross sectional study.

Based on the **STROBE** cross sectional guidelines.

Instructions to authors

Complete this checklist by entering the page numbers from your manuscript where readers will find each of the items listed below.

Your article may not currently address all the items on the checklist. Please modify your text to include the missing information. If you are certain that an item does not apply, please write "n/a" and provide a short explanation.

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		Reporting Item	*Page*
Title and abstract			
Title	#1a	Indicate the study's design with a commonly used term in the title or the abstract	1
Abstract	#1b	Provide in the abstract an informative and balanced summary of what was done and what was found	2
Introduction			
Background / rationale	#2	Explain the scientific background and rationale for the investigation being reported	4
Objectives	#3	State specific objectives, including any prespecified hypotheses	4
Methods			
Study design	#4	Present key elements of study design early in the paper	5-6
Setting	#5	Describe the setting, locations, and relevant dates, including periods of recruitment, exposure, follow-up, and data collection	5-6
Eligibility criteria	#6a	Give the eligibility criteria, and the sources and methods of selection of participants.	5-6
	#7	Clearly define all outcomes, exposures, predictors, potential confounders, and effect modifiers. Give diagnostic criteria, if applicable	5
Data sources / measurement	#8	For each variable of interest give sources of data and details of methods of assessment (measurement). Describe comparability of assessment methods	5-6

		Reporting Item	*Page*
		if there is more than one group. Give information separately for exposed and unexposed groups if applicable.	
Bias	#9	Describe any efforts to address potential sources of bias	5-6, 12
Study size	#10	Explain how the study size was arrived at	5-6, 12
Quantitative variables	#11	Explain how quantitative variables were handled in the analyses. If applicable, describe which groupings were chosen, and why	6-10
Statistical methods	#12a	Describe all statistical methods, including those used to control for confounding	6
Statistical methods	#12b	Describe any methods used to examine subgroups and interactions	6
Statistical methods	#12c	Explain how missing data were addressed	7
Statistical methods	#12d	If applicable, describe analytical methods taking account of sampling strategy *Not required as sampling strategy was same across single cohort*	N/A
Statistical methods	#12e	Describe any sensitivity analyses *Not required*	N/A
Results			
Participants	#13a	Report numbers of individuals at each stage of study—eg numbers potentially eligible, examined for eligibility, confirmed eligible, included in the study, completing follow-up, and analysed. Give information separately for for exposed and unexposed groups if applicable.	7-10, 17-22, vi
Participants	#13b	Give reasons for non-participation at each stage	7
Participants	#13c	Consider use of a flow diagram *Not required*	N/A
Descriptive data	#14a	Give characteristics of study participants (eg demographic, clinical, social) and information on exposures and potential confounders. Give information separately for exposed and unexposed groups if applicable.	7-8, 17-19
Descriptive data	#14b	Indicate number of participants with missing data for each variable of interest	7-10, 17-22, vi
Outcome data	#15	Report numbers of outcome events or summary measures. Give information separately for exposed and unexposed groups if applicable.	7-10, 17-22, i-vi
Main results	#16a	Give unadjusted estimates and, if applicable, confounder-adjusted estimates and their precision (eg, 95% confidence interval). Make clear which confounders were adjusted for and why they were included	N/A

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		Reporting Item	*Page*
		Not relevant	
Main results	#16b	Report category boundaries when continuous variables were categorized	8, 17
Main results	#16c	If relevant, consider translating estimates of relative risk into absolute risk for a meaningful time period *Not relevant*	N/A
Other analyses	#17	Report other analyses done—e.g., analyses of subgroups and interactions, and sensitivity analyses	7-10, 17-22, i-vi
Discussion			
Key results	#18	Summarise key results with reference to study objectives	10-12
Limitations	#19	Discuss limitations of the study, taking into account sources of potential bias or imprecision. Discuss both direction and magnitude of any potential bias.	12
Interpretation	#20	Give a cautious overall interpretation considering objectives, limitations, multiplicity of analyses, results from similar studies, and other relevant evidence.	10-12
Generalisability	#21	Discuss the generalisability (external validity) of the study results	12
Other Information			
Funding	#22	Give the source of funding and the role of the funders for the present study and, if applicable, for the original study on which the present article is based	13

Notes:

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Measuring physician practice, preparedness and preferences for genomic medicine: a national survey

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Measuring physician practice, preparedness and preferences for genomic medicine: a national survey

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Abstract

Objective: Even as genomic medicine is implemented globally, there remains a lack of rigorous, national assessments of physicians' current genomic practice and continuing genomics education needs. The aim of this study was to address this gap.

Design: A cross-sectional survey, informed by qualitative data and behaviour change theory, to assess the current landscape of Australian physicians' genomic medicine practice, perceptions of proximity and individual preparedness, and preferred models of practice and continuing education. The survey was advertised nationally through 10 medical colleges, 24 societies, 62 hospitals, social media, professional networks and snowballing.

Results: 409 medical specialists across Australia responded, representing 30 specialties (majority paediatricians, 20%), from mainly public hospitals (70%) in metropolitan areas (75%). Half (53%) had contacted their local genetics services and half (54%) had ordered or referred for a gene panel or exome/genome sequencing (E/GS) test in the last year. Two-thirds (67%) think genomics will soon impact their practice, with a significant preference for models that involved genetics services (p<0.0001). Currently, respondents mainly perform tasks associated with pre-test family history taking and counselling, but more respondents expect to perform tasks at all stages of testing in the future, including tasks related to the test itself, and reporting results. While a third (34%) recently completed education in genomics, only a quarter (25%) felt prepared to practice. Specialists would like (more) education, particularly on genomic technologies and clinical utility, and prefer this to be through varied educational strategies.

Conclusions: This survey provides data from a breadth of physician specialties that can inform models of genetic service delivery and genomics education. The findings support education providers designing and delivering education that best meet learner needs to build a competent, genomic-literate workforce. Further analyses are underway to characterise early adopters of genomic medicine to inform strategies to increase engagement.

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Strengths and limitations of this study

- The survey tool is based in behavioural change theory and developed from empirical data to capture patterns of genomic practice and preferences, allowing comparisons across different settings and change over time.
- We employed an extensive, multi-staged and overlapping recruitment strategy at a national level to reach as many Australian medical specialists and trainees as possible.
- We successfully gathered data from over 30 specialties, the broadest sample reported in the literature to date.
- Our sample is still relatively small, and over-represented for older specialists and those working in rural and remote areas, which may influence the findings.
- Our study is the first to investigate the genomics education and training needs and preferences of a national sample of a broad range of medical specialties.

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Introduction

Genomic sequencing is shifting from the realm of research to healthcare.[1] A recent review identified five models for the provision of genetic testing globally, including genetics services led by geneticists, referral by primary-care physicians to genetics services, and medical specialist-led testing.[2] The shortage of a specialist genetic workforce suggests that medical specialist-led testing will be necessary.[3][4] A scoping review of genetic specialist workforces internationally emphasised the need for a medical specialist-led model, noting education as a driver of workforce capacity.[5]

A national alliance of over 80 partner organisations, Australian Genomics, formed in 2016 to conduct research supporting adoption of genomics into Australian healthcare.[6] At that time, microarray analysis and a limited number of single gene tests were reimbursed through the federally-funded Medicare Benefit Scheme (MBS). Genomic sequencing tests were largely available through research studies or patient funding until 2020, when exome/genome sequencing (E/GS) for certain conditions was included on the MBS.[7]

Despite national initiatives driving the use of germline genomic tests by medical specialists not qualified in genetics, there are indications that physicians may prefer to refer to genetics services.[8, 9] Cumulative evidence indicates a lack of physician confidence in genomic medicine and low rates of clinical adoption of genomic testing.[10] Studies investigating practice and preparedness span specialties and countries: Dutch cardiologists,[11] European obstetricians and paediatricians,[12] Wisconsin physicians,[13] British gastroenterologists,[14] Australian intensivists,[8] and neurologists worldwide.[15] However, there are no national studies surveying a range of specialties.

Education strategies have been proposed or implemented to support medical professionals' genomic medicine knowledge and skills.[16, 17] Following medical school training,[18, 19] continuing professional development (CPD), whether accredited or not, aims to supplement knowledge and skills for those already in practice.[20, 21] To inform Australian national strategy and local development of genomics CPD, a needs assessment inclusive of a multiple specialties across diverse contexts is required. We previously reported development of a survey underpinned by qualitative data and an empirically-derived framework of behaviour change in which capability, opportunity and motivation influence, and are influenced by, behaviour (the COM-B model).[22]

Here we describe comprehensive deployment of this survey nationally to multiple medical specialities. We present a snapshot of the current landscape of Australian specialists' genomic medicine practice, perceptions of proximity of genomic medicine and individual preparedness, and preferred models of practice and continuing education.

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Methods

In Australia, medical doctors undertake training within a medical college to train as medical specialists, e.g., the Royal Australasian College of Surgeons trains surgeons. Training typically involves completing three years of basic training ('Basic Trainee') followed by three years of advanced training ('Advanced Trainee'). Training programs are specific to the college and the specialty of interest with varied exposure to genetics/genomics. Recognising that the term 'physician' has different meanings in different countries, here we define 'physicians' as doctors whose primary affiliation is with the RACP. After successful completion of final examinations, they become a Fellow of the relevant medical college.[23] Medical professionals may work in public hospitals, which are the responsibility of State governments, and/or privately. Patients receive some reimbursement for private consultations and specified pathology tests through the Federal Government's MBS. At the time of the survey, there were 20 genetics conditions for which tests were reimbursed through the MBS (see **Supplementary Table S1**). Clinical genetics services provide screening, diagnostic and genetic counselling services to patients on referral by a medical practitioner. They are based primarily in publicly-funded hospitals and staffed by health professionals trained in genetics (e.g., clinical geneticists, genetic counsellors. Here we focus on the non-genetic medical workforce and as such define 'medical specialists' as medical doctors who are trained or in training for a specialty other than clinical genetics. We excluded general practitioners (family physicians) who practise general medicine in the community and genetic specialists (e.g., clinical geneticists and genetic counsellors) as separate studies were conducted for those subspecialties.[4](Cusack et al., *Australian Journal of General Practice*, in press). We also excluded radiologists and pathologists as in Australia they typically perform investigations than requesting genomic tests, and oncologists, as they are the focus of other ongoing national studies.

Details of survey development, domains and the full set of questions have been reported elsewhere.[20, 24] In brief, the survey is informed by the COM-B model and includes 28 questions across five key domains: personal characteristics, current practice with genomic medicine, perception of preparedness to practice genomic medicine, perception of how proximal genomic medicine is to clinical practice, and preferences for future models of practice and education. We defined 'genomic medicine' as the use of testing that investigates many regions of the genome at once, such as gene panels and E/GS, but excluding non-invasive prenatal testing using sequencing technologies. The scope of the survey was testing to investigate genetic conditions. The survey was deployed electronically from February to September 2019 using REDCap (Research Electronic Data Capture) software hosted at the Murdoch Children's Research Institute.[25]

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This project received ethics approval from the University of Melbourne, Melbourne, Australia (HREC number: 1646785.10). Respondents provided consent by completing the initial screening and consent question.

Recruitment

Inclusion criteria: medical specialists were eligible to complete the survey if they had commenced or completed their specialist training and were currently practising clinically in Australia.

Recruitment was staged through:

- Relevant medical colleges (Mar–Jun 2019) and societies/associations (Apr–Jun 2019).
- Hospitals (Jun–Oct 2019). 132 hospitals were identified from the 'MyHospitals' search tool on the Australian Institute of Health and Welfare website[26] to represent both public and private hospitals in metropolitan, regional and rural settings across all Australian states.
- Social media (Jun–Jul 2019). Three tweets were posted on the Australian Genomics Twitter account (https://twitter.com/AusGenomics) over 10 business days, then this process was repeated twice, with approximately one week between each cluster of tweets. Content referenced specific survey questions or preliminary data to pique interest of potential participants. For example, *'Early survey results suggest that even though medical specialists are ordering #genomictests for their #patients, many don't feel #prepared for #genomicmedicine. We want to know how you feel [LINK]'* or *'Do you feel ready for #genomics in #clinicalpractice? We want to hear from Australian medical specialists [LINK]'*.
- Investigator networks of national and state-based genomics initiatives, Australian Genomics and Melbourne Genomics (Jul 2019).

Medical colleges, societies and hospitals circulated information about the study to their membership or staff using regular communication channels, e.g., newsletters, e-bulletins, emails, etc. Information was circulated up to three times per organisation, dependent on advertising charges, perceived responder burden and/or internal timelines. The information included a brief description of the study, ethics approval and a link to access the online survey. Recruitment also included professional networks and snowball sampling throughout, with all contacts asked to retweet Australian Genomics tweets if possible. All respondents were asked to share the survey with relevant colleagues.

Data cleaning and analysis

Data were exported to, cleaned and then analysed in Stata 16.0. Cleaning involved removing surveys completed by ineligible respondents or surveys with no data beyond demographic questions. For analysis, career stage was grouped into Basic Trainee, Advanced Trainee or Fellow, as defined above.

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Specialists were grouped according to self-reported primary college affiliation. All categorical questions included an open-ended text option for 'Other'; qualitative data provided for these questions were reviewed by three researchers (AN, EK, MJ) and recoded into existing response categories if possible (see **Supplementary Table S2** for examples). Representative quotes are provided in **Supplementary Table S3** for illustrative purposes where they enhance the understanding of the quantitative results.

Descriptive and inferential statistics were used to analyse the data, including two-sample tests of proportions, chi-square or Fisher's exact tests as appropriate to data characteristics. A *p* value of <0.05 was considered significant. When determining representativeness of the sample, data were referenced against Medical Board of Australia Registrant data,[27] the National Medical Training Advisory Network, and the National Health Workforce Dataset.[28]

Patient and public involvement

There was no patient or public involvement in this research.

Results

Recruitment and response rates

As shown in **Figure 1**, recruitment strategies were staggered and overlapping from March to October 2019. All 10 Australian medical colleges and 24 of 55 medical societies/associations approached agreed to advertise the survey. Of 132 health networks[1] and hospitals contacted,[29] 62 agreed to advertise the survey (67.6% of metropolitan hospitals and 42.9% of remote hospitals), which was subsequently shared with staff at a total of 74 hospitals. There were an estimated 37,000 trainees and fellows in our target specialty audiences at the time of the survey.[27] However, using diverse recruitment approaches that could target one individual in several ways and at several time points meant that it was not possible to determine how many medical specialists were aware of the survey during the recruitment period.

Sample characteristics and representativeness

Of 617 attempts at survey responses, 54 did not meet the inclusion criteria and 154 did not complete any questions beyond consent to participate (see **Supplementary Figure S1** for detail). A total of 409 responses were therefore included in analyses. Totals differ across questions due to opportunity to provide more than one response, missing data or attrition; where this has occurred, the denominator has been described.

[1] Health networks are functional or geographical groups of Australian public hospitals defined by the relevant State Government.

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Table 1 presents respondent demographics compared with reference data from the Medical Board of Australia,[27] the National Medical Training Advisory Network[30, 31] and the National Health Workforce Dataset.[28] Our sample had slightly less males (p=0.039), was under-represented for 25–34 year olds (p<0.0001), and over-represented for 55–64 year olds (p<0.0001). As would be expected from this age bias, there was a smaller proportion of Basic and Advanced Trainees than expected from the reference data and a larger proportion of Fellows (p<0.0001). Our sample was broadly representative of primary work locations of medical specialists across Australia. Of the eight Australian states and territories, one was over-represented (Australian Capital Territory; p<0.0001) and two were under-represented (South Australia; p=0.028); Western Australia; p=0.032). Although three-quarters of respondents worked in a major city, those working in remote regions were significantly over-represented in our sample (p=0.0018). The majority of respondents were primarily employed at public hospitals or healthcare providers. A quarter of respondents had been involved in a genomics research project in the last 5 years (n=96, 24.7%). Of these, respondents were involved in clinical (83.3%), laboratory (49.0%), bioinformatics (15.6%) and/or social science (6.3%) projects. Only 7.2% of respondents indicated that they were affiliated with any state- or federally-funded genomic health alliances. **Error! Reference source not found.** describes proportions of respondent specialties, compared with the proportions expected from reference data.[27] The largest group of respondents were physicians, totalling 232 (56.7%) responses. Our sample was representative of most specialties with some exceptions: there were more physicians (p<0.0001 and fewer anaesthetists (p=0.002), psychiatrists (p<0.0001) and surgeons (p=0.0001).

Current practice in genomic medicine

Respondents (n=387) answered a series of questions about their current practice in genomic medicine. Just over half of respondents had contacted their local genetics service in the last 12 months (n=203, 52.5%), although this was relatively infrequent, with a third of these 203 respondents indicating this was once or twice in the last 12 months (36.6%). The main reasons for contacting genetics services included: seeking information about a suspected genetic condition (48.0%), advice on how to refer a patient (42.6%) and choosing which genetic or genomic test to order (38.1%). Of those who had not contacted clinical genetics, the majority indicated that this was because they had not yet needed advice (73.5%).

Over half of respondents (n=208, 53.9%) had engaged in genomic sequencing testing in the last 12 months by either ordering a gene panel or E/GS, or referring a patient to a genetics service for those tests. Nearly a third of respondents (n=121, 31.3%) had ordered at least one of these tests, with 29.0% (n=112) ordering a gene panel and 13.0% (n=50) ordering E/GS. When asked about frequency of ordering each test in the previous year, the most common response was once or twice for both

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gene panels (n=42/112, 37.5%) and E/GS (n=23/50, 46.0%). In contrast, 112 respondents (29.0%) had ordered a microarray in the previous year, most commonly monthly (n=41/112, 36.6%). Funding for tests varied (**Supplementary Table S4**), with microarray tests often funded by the MBS, gene panel tests by the institute/hospital, and E/GS tests by research grants. Overall, 63.3% of respondents (n=245/387) had engaged in genetics/genomics in one or more ways: contacting their genetics service, or ordering or referring for a microarray, gene panel or E/GS test.

Respondents were asked to reflect on their confidence about genomic concepts and skills (**Error! Reference source not found.**). Medical specialists reported the highest level of confidence when taking a family history to elicit information about genetic conditions, and lowest for knowledge about genomics. There was greatest variation in their confidence to make decisions based on genomic information (IQR=2,7).

Current practice compared with expected future practice in genomic medicine

Overall, two-thirds of respondents think genomics will impact their practice in the next two years (n=199/298, 66.8%). Of those medical specialists who think their practice will be impacted, they anticipate it will change the way they manage patients (n=177/199, 88.9%) and practice medicine (n=151/199, 75.8%), more so than impact on workload (n=86/199, 43.2%). For respondents who felt genomics would not impact their practice in the next two years (n=50/298, 16.8%), open-text comments (n=47) suggested this was due to perceived relevance to their specialty, timing and/or pragmatic issues of service delivery (see **Supplementary Table S3** for examples). The remaining 49/298 (16.4%) respondents were 'unsure'.

More respondents currently perform clinical activities before and after E/GS testing (**Error! Reference source not found.**, n=314, 10.6% to 80.3% across these steps) than are involved in non-clinical activities directly related to the test itself (6.7% to 17.0%). Similar patterns were seen in their expectations of the steps they would perform in the future if they had adequate education, training and support: 40.8% expect to perform all pre-test steps and 23.1% all post-test steps, while 40.3% do not expect to perform any steps relating to the test itself. Notably, there were significant increases in the proportion of specialists who expect to perform each step in future practice ($p \leq 0.004$ across all steps), with the exception of eliciting phenotypic information about genetic conditions as part of a family or medical history for the purpose of assisting with variant interpretation, which was already high (80.3% current, 83.4% future; $p=0.3$).

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Preferred future models for delivering genomic medicine

When reflecting on preferred models for delivering genomic medicine in the future, the model most often selected by respondents was referral to their local genetics services to initiate testing and discuss results (

Table 2). This was the case for both inpatient and outpatient settings. The second most preferred model was delivering testing with support from a local genetics service. The type of support included: advice on whether testing is appropriate (60.0% for inpatients; 66.7% for outpatients); interpreting results (72.0% for inpatients; 75.0% for outpatients); discussing results with families (60.0% for inpatients; 70.8% for outpatients); or follow-up genetic counselling (80.0% for inpatients; 83.3% for outpatients). A small number expect to initiate genomic testing themselves with no support from a local genetics service, while some respondents also indicated they did not expect to see patients who would benefit from genomic testing. Overall, significantly more respondents preferred a model that includes involvement of genetics services (for support or referral) than a model of initiating testing themselves: inpatients, 62.4% (95%CI 54.8–69.5) compared with 2.3% (95%CI 0.6–5.6), $p<0.0001$); outpatients, 69.7% (95%CI 62.8–76.1) compared with 4.1%; (95%CI 1.8–7.9, $p<0.0001$).

Preparedness for genomic medicine and preferences for future education

While a third (n=92/273, 33.7%) of respondents had completed education in genomics in the past year, only a quarter (n=73/297, 24.6%) felt prepared to use genomic sequencing testing in their practice. Comments from those who did not feel prepared or were 'unsure' (n=210 combined) primarily suggest this could be addressed through genomics education and training (**Supplementary Table S3**). Forty-two per cent of respondents felt that improved genomic knowledge may alter their clinical practice (n=115/273, 42.1%) but a similar proportion were 'unsure' (n=114/273, 41.8%).

When asked about preferred modes of learning genomics, most respondents (n=250/273; 91.6%) endorsed at least three different modes (

Table 3). The two most commonly preferred – CPD activities and learning from peers – were also the two most commonly-used currently. In contrast, reading specialty texts was the third most common way of learning about genomics currently, but the eighth preferred. Respondents indicated a preference for genomics education incorporated into their usual work activities (e.g., internal workplace seminars, departmental presentations and clinical meetings).

Despite three-quarters of respondents reporting they had already learned basic concepts of genomics (

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Table 4; n=271), a similar proportion still requested this topic for future education. Six topics were endorsed by over 80% of respondents including current and emerging applications in genomic medicine, the clinical utility of different tests and topics around patient management. Again, respondents could select more than one topic, with 92.3% indicating they wanted to learn about at least five topics in the future, and 26.4% selecting all topics. Nearly two-thirds of respondents indicated they wanted to learn about communication skills with patients, with comments throughout the survey suggesting a need for training in how to explain genomic testing concepts, implications and results to patients.

Discussion

This paper provides a baseline snapshot of Australian non-genetic medical specialists' practice of genomic medicine and perspectives at a point in time before E/GS was widely available to them as a funded clinical test. In 2019, 60% of all 409 survey respondents reported some form of interaction with genetics services or genetic/genomic testing. The test ordered most frequently was a microarray, but more than a quarter of all survey respondents indicated they had ordered a genomic sequencing test in the past twelve months. Respondents anticipated their practice would change in the near future, with significantly more respondents expecting to be involved in activities relating to E/GS in the next two years than currently. Consistent with discipline-specific studies from other countries,[13, 15, 32-34] we found the majority of respondents in our survey did not feel prepared to use genomic sequencing testing in their practice and over two-thirds preferred a model that involved genetics services in some way. Our study extends existing literature by providing greater depth of insight into the education needs and preferences of a broad range of medical specialists.

A strength of this snapshot is the use of a survey tool[24] grounded in a theoretical model. The COM-B model posits that behaviour is influenced by capability, opportunity and motivation.[22] *Opportunity* is clearly impacted by the availability of funded genomic tests. The test usage reported by respondents in this study reflects the availability of MBS reimbursement. For instance, microarrays for developmental delay have been established as MBS-reimbursed pathology tests for a decade. Tests reimbursed at the time of this survey are most typically requested by oncologists, clinical geneticists, haematologists, immunologists, paediatricians, obstetricians, nephrologists and neurologists.[35] At the time of this survey E/GS tests were not reimbursed by the MBS. The relatively lower proportion of respondents who had ordered these tests used a variety of other funding mechanisms, most commonly hospital or research funds, and noted availability of funding as an influence when ordering genomic tests in the future. Since this study was completed, MBS now reimburses genomic sequencing tests for some clinical indications when ordered by paediatricians,

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enhancing their opportunity to use genomic testing in their clinical practice. It is anticipated that reimbursement for other clinical indications (and medical specialties) will follow in the future.

Respondent's perceived *capability* to respond to the availability of funded tests, however, is limited. Currently, respondents lack confidence in their knowledge, ability to explain genomic concepts and make decisions based on genomic information. This may explain their desire to practice collaboratively with clinical geneticists and genetic counsellors to varying extents. It is possible that these preferences could change as their capability (and confidence in their capability) develops with greater opportunity, experience and learning.[22, 24] Education and training was certainly seen as a solution to feeling unprepared by a substantial proportion of respondents in this study, as also observed by others.[36] In the past two years, continuing education for Australian medical specialists has been produced locally at an introductory level by a number of initiatives and organisations.[2] More is clearly needed: survey respondents are very interested in genomics education and nearly all respondents selected five or more of the topics that they wished to learn about. This is perhaps unsurprising given their perception of being unprepared and expectation of a greater role in the near future, provided they receive adequate support and education. The most popular education topics were related to pre-test aspects of testing, such as identifying appropriate patients to refer and knowing how to refer, consistent with the significantly stronger preference for a genetics-led model for genomic medicine. Educational strategies will need to consider both the diversity of respondents' preferences for modes of learning and timing with respect to clinical implementation. Not only will timing affect perceived relevance to clinical practice, and therefore motivation to learn,[37] but preferences and needs may evolve as implementation progresses.

Our rigorously-developed survey tool can be deployed again in the future to capture changes in workforce practice and preferences over time. It could also be repurposed to inform needs for national education initiatives targeted to specific specialties or to assess change in their knowledge, practice or preferences. Wider use of the tool can also provide a basis for documenting and comparing data across specialties and countries. Our experience with deployment of the survey may assist in this regard, as we purposefully staggered recruitment methods to monitor response rates. Although it is not possible to determine which recruitment approach was most successful because of overlapping timeframes, increases in the number of responses to our survey coincided with recruitment approaches using social media, internal hospital communication channels and investigator networks. This may reflect increasing professional use of social media by medical specialists[38] and greater attention to emails from their employing hospital than a medical college

[2] For example, https://elearning.racp.edu.au/course and http://learn-genomics.org.au/.

or society. It may also explain the higher representation of Fellows and older specialists in our sample, as trainees were often not on staff mailing lists used by hospitals to distribute the survey. Our staggered and comprehensive recruitment approach also achieved a strong response from rural and remote medical specialists, who are often missed in research. Under- or over-representation of medical specialists in some Australian states may be due to differences in governance (hospital and/or research) and site-based communication policies that limited dissemination of the survey. One could assume specialists who graduated more recently may be more engaged with genomic medicine but previous research from our group described varied genomic literacy and experience at each career stage.[20] Similarly, specialists working in metropolitan areas, where almost all genetics services are based, might have been expected to be likely to complete our survey but this was not seen in our sample. While it is not possible to determine the response rate, our sample represents 1.2% of 37,000 medical specialist registrants with the Medical Board of Australia[27] and is within the range achieved in similar surveys of American physicians that also recruited participants through medical societies and associations (0.6–2.6%).[13, 39-41]

This national snapshot of medical specialists' current practice in genomic medicine provides the first detailed insight into the continuing genomics education needs of a broad group of subspecialties. It includes some specialties, such as emergency medicine, palliative medicine and infectious disease, for the first time internationally. Those currently involved and/or most interested in genomic medicine may have been more likely to respond, meaning these results may present an overestimation of current practice in Australia, but this might also mean our respondents are those likely to undertake continuing education and engaging with genomics. Consequently, our results can assist providers to best meet learner needs when developing and implementing genomics education to ultimately create a competent, genomics-literate workforce. The findings will also be helpful to genetics and other clinical services implementing models for genomic medicine delivery. Further data analysis will provide insights into any differences between early adopters of genomic medicine and those who have not yet engaged, enabling the development of targeted, tailored genomics education and other capability-building strategies for optimising the adoption of genomics by medical specialists.

Author contributions

AN and EK were involved in all stages of this work and manuscript preparation. BM, SM and CG were involved with all stages except data acquisition. MJ analysed the qualitative data responses and was involved in manuscript preparation. All authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately

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Competing interests

All authors declare no completing interests.

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Data sharing statement

Data are available upon reasonable request.

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Endocrinology Group; Australian Paediatric Research Network; Australian Paediatric Society; Australian Rheumatology Association; Australian Society of Anaesthetists; Australian Society of Ophthalmologists; College of Intensive Care Medicine of Australia & New Zealand; Endocrine Society of Australia; Gastroenterological Society of Australia; Royal Australasian College of Physicians; Royal Australasian College of Surgeons; Royal Australian & New Zealand College of Ophthalmologists; Royal Australian & New Zealand College of Psychiatrists; Royal Australian College of Obstetricians & Gynaecologists (RANZCOG); Rural Doctors Association Australia; Society of Obstetric Medicine of Australia & New Zealand; and Thoracic Society of Australia & New Zealand. Publication fees were waived by ACSEP and reduced by ANZCA and RANZCOG.

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Figure captions

Figure 1: *Number of survey attempts shown with recruitment strategies and timelines after pilot data were complete (n=41). Recruitment start dates are shown and overlapped from March through October 2019 (as described in the Methods). Snowball recruitment may have continued beyond these periods (e.g., forwarding a newsletter or retweeting) but this could not be monitored.*

Figure 2: *Proportion of each reported primary specialty in the sample (n=409) grouped by primary medical college affiliation. Grey bars signify specialties where proportions were representative of the medical specialist population when compared with reference data.[27] The black bar signifies a specialty which was over-represented (physicians; p<0.0001). White bars signify specialties which were under-represented: anaesthesiology (p=0.002), psychiatry (p<0.0001) and surgery (p<0.0001). The reference data did not include a classification for 'rural and remote medicine' so representativeness could not be determined for this specialty (pale grey bar).*

Figure 3: *Average confidence about genomic concepts and skills on a scale of 1 'Not at all confident', 5 'Neutral' to 10 'Very confident' (n=273). Boxes represent the interquartile ranges with minimum and maximum value; medians are shown as white bars.*

Figure 4: *Steps in genomic testing that respondents (n=314) currently perform (blue bars) compared to steps they expect to perform in the future, if they had adequate support, education and training (black bars). Non-clinical steps are indicated by [a]. Differences between proportions for 'Currently perform' and 'Expect to perform' are indicated by *p=0.004, **p=0.001, ***p=0.0006, ****p<0.0001. The difference for the first step – Elicit genetic information through family history – was not significant (p=0.3). The full wording of each step is provided in* ***Supplementary Table S5****.*

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Tables

Table 1: *Description of the sample and representativeness (n=409).*

Characteristic	Respondents		Reference data		
	n (%)	95%CI	N (%)	95%CI	*p*
Gender[1]					
Male	213 (52.1)	47.2–56.9	61,700 (57.1)	56.8–57.4	0.039
Female	185 (45.2)	40.4–50.1	46,281 (42.9)	42.6–43.2	0.330
Prefer not to answer	11 (2.7)	1.5–4.8	–	–	–
Age[1]					
≤24 years	–	–	398 (0.4)	–	
25–34 years	29 (7.1)	4.6–9.6	26,827 (24.8)	24.6–25.1	<0.0001
35–44 years	114 (27.9)	23.5–32.2	28,431 (26.3)	26.1–26.6	0.4794
45–54 years	123 (30.1)	25.6–34.7	22,415 (20.8)	20.5–21.0	<0.0001
55–64 years	103 (25.2)	21.2–29.6	18,060 (16.7)	16.5–17.0	<0.0001
≥65 years	40 (9.8)	7.2–13.1	11,852 (11.0)	10.8–11.2	0.4398
Trainee level[2]					
Basic Trainee	9 (2.2)	1.3–4.6	5,858 (12.1)	11.8–12.4	<0.0001
Advanced Trainee	18 (4.4)	2.6–6.7	8,890 (18.3)	18.0–18.7	<0.0001
Fellow	382 (93.4)	89.9–95.0	33,749 (69.6)	69.2–70.0	<0.0001
Australian state or territory[1,3]					
Australian Capital Territory	28 (6.9)	4.4–9.3	702 (1.9)	1.8–2.0	<0.0001
New South Wales	119 (29.1)	24.7–33.5	11,566 (31.2)	30.7–31.7	0.3622
Northern Territory	8 (2.0)	0.6–3.3	373 (1.0)	0.9–1.1	0.0568
Queensland	75 (18.3)	14.8–22.4	7,320 (19.7)	19.3–20.1	0.4777
South Australia	20 (4.9)	2.8–7.0	2,896 (7.8)	7.5–8.1	0.0283
Tasmania	13 (3.2)	1.5–4.9	759 (2.0)	1.9–2.2	0.1091
Victoria	119 (29.1)	24.7–33.5	9,952 (26.8)	26.4–27.3	0.3063
Western Australia	26 (6.4)	4.0–8.7	3,510 (9.5)	9.2–9.8	0.0324
Primary work location[3,4]					

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Characteristic	Respondents		Reference data		
	n (%)	95%CI	N (%)	95%CI	*p*
Major city	306 (75.0)	70.6–79.0	72,304 (79.2)	78.9–79.4	0.0391
Inner regional	59 (14.5)	11.4–18.2	12,422 (13.6)	13.4–13.8	0.6127
Outer regional	31 (7.6)	5.4–10.6	5,299 (5.8)	5.7–6.0	0.1216
Remote	10 (2.5)	1.3–4.5	865 (1.0)	0.9–1.0	0.0018
Very remote	2 (0.5)	0.1–2.0	376 (0.4)	0.4–0.5	0.8048
***Primary employer*[5]**					
Public hospital or healthcare provider	288 (70.4)	65.8–74.7			
Private hospital or healthcare provider	17 (4.2)	2.6–6.6			
Self-employed/ private practice	83 (20.3)	16.7–24.5			
Other (government, research institute, etc.)	21 (5.1)	3.4–7.8			

Reference data were: [1] Registration Data Table 2019 [27]; [2] Medical Education and Training in Australia 1st Edition report 2017 [31]; [3]n=408 for state and location; [4] Medical Workforce 2016 Factsheet [28]; [5] There were no comparable reference data for this category.

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Table 2: *Medical specialists' preferred models for delivering a genomic sequencing test in inpatient and outpatient settings (n=218).*

	INPATIENT n=178[1]		OUTPATIENT n=195[1]	
	n (%)	**95%CI**	**n (%)**	**95%CI**
You **initiate** testing and discuss results with patients/families	4 (2.3)	0.6–5.6	8 (4.1)	1.8–7.9
You initiate testing and discuss results with patients/families, with **support** from a clinical genetics team as needed	43 (24.2)	18.15–31.1	49 (25.1)	19.2–31.8
You **refer** to a clinical genetics team to initiate testing and discuss results with patients/families	68 (38.2)	31.0–45.8	87 (44.6)	37.5–51.9
You **do not see**, and do not expect to see, patients who would benefit from genomic testing	33 (18.5)	13.1–25.0	23 (11.8)	7.6–17.2
Unsure at this stage	30 (16.9)	11.7–23.2	28 (14.4)	9.8–20.1

[1] *A total of 218 respondents completed this question, indicating a preference for either the inpatient or outpatient setting, or both.*

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Table 3: *Current and preferred modes of learning about genomics (n=273).*[1]

Mode of learning about genomics	Currently use (%)	Prefer to use (%)
Continuing Professional Development/Continuing Medical Education activities	51.8	79.8
Consult colleagues and peer	54.0	79.4
Internal workplace specialty seminars, conferences or similar	34.1	74.0
Departmental presentations	35.8	72.0
Clinical meetings	34.8	71.4
External specialty seminars, conferences, etc.	36.0	67.3
Internal workplace genetic or genomic seminars, conferences, etc.	24.9	66.3
Reading specialty texts	48.2	63.2
Online webinars, courses, MOOCs, etc.	15.8	59.6
Certification/fellowship activities	34.4	56.4
External genetic or genomic seminars, conferences, etc.	18.4	50.0
Small group tutorials	8.1	44.9
Study days at place of employment	12.5	41.9
Genomic research project	17.6	32.6
Time in a service or laboratory with genomics expertise	6.2	17.6
Mass media	12.5	14.0
Social media	7.4	11.0
Other (e.g., fact sheet written by geneticist)	0.0	0.4

[1] *Respondents could select more than one mode.*

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Table 4: *Topics relevant to genomics medicine that medical specialists have learnt about or would like to learn (more) about (n=271).*[1]

Education topic	Have learnt about (%)	Want to learn (more) about (%)
Genetic/genomic knowledge		
Basic concepts	77.5	77.1
Disorders and diseases	74.2	83.4
Current applications in genomic medicine	60.9	88.9
Emerging applications in genomic medicine	55.7	87.8
Genetic/genomic testing and technology		
Types of genetic tests	64.9	76.4
Types of genomic tests	58.7	77.1
Applications of somatic genomic tests	45.4	75.6
Applications of germline genomic tests	37.6	69.7
Clinical utility of tests	57.6	88.6
Classification of genomic data during testing	41.3	67.9
Limitations of testing	50.2	79.7
Pre- or post-test aspects		
Recognising patients who may benefit from genomic testing	60.9	83.0
Communication skills with patients	70.8	63.1
Performing genetic risk assessments	57.6	67.5
Referring appropriately for a genomic test	59.4	81.5
Requesting a genomic test for a patient	53.9	70.8
Interpreting genomic test results	52.0	74.9
Cascade testing	53.9	68.6
Ethical, legal and social implications		
Ethical implications	59.0	75.6
Legal implications	52.4	75.3
Psychosocial implications	57.2	74.9

[1] *Respondents could select more than one topic.*

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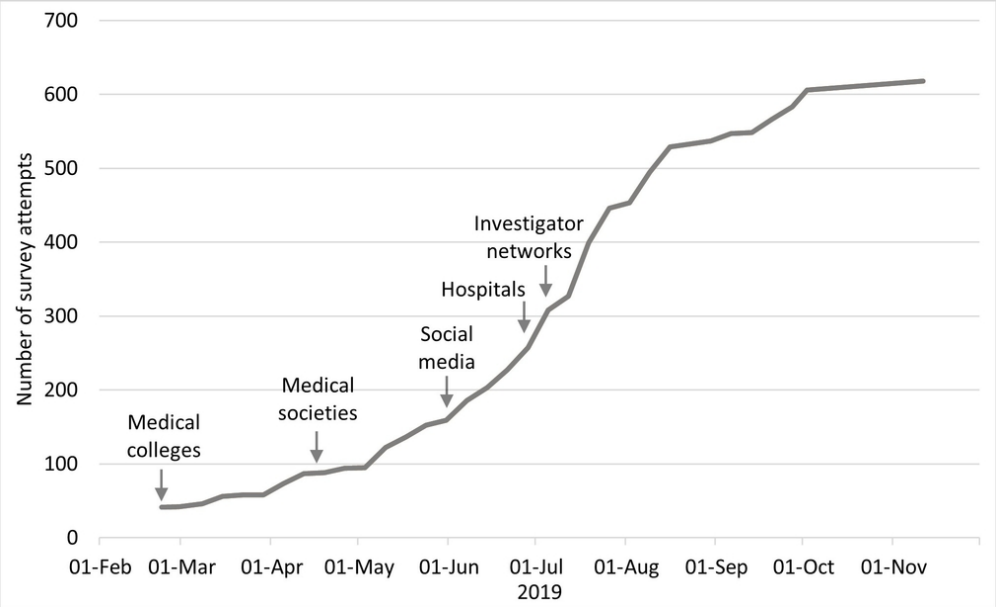

Figure 1: Number of survey attempts shown with recruitment strategies and timelines after pilot data were complete (n=41). Recruitment start dates are shown and overlapped from March through October 2019 (as described in the Methods). Snowball recruitment may have continued beyond these periods (e.g., forwarding a newsletter or retweeting) but this could not be monitored.

85x52mm (300 x 300 DPI)

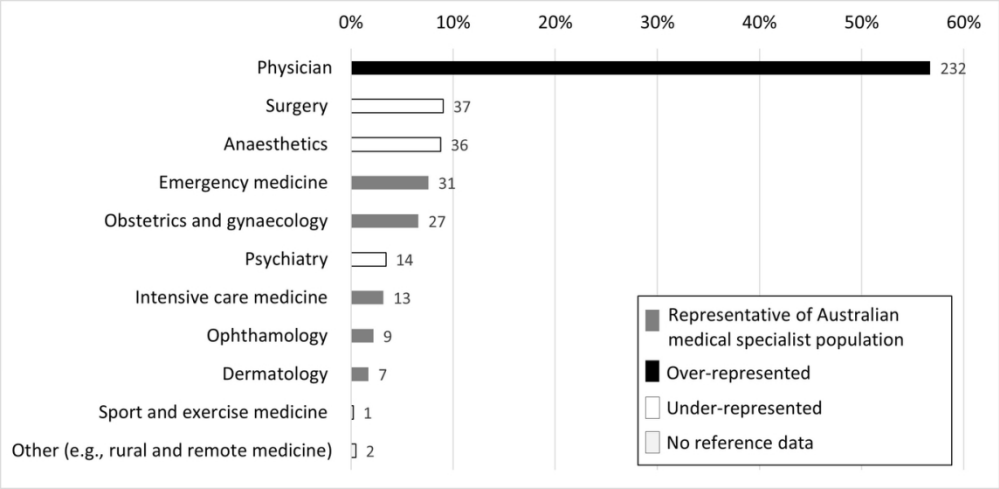

Figure 2: Proportion of each reported primary specialty in the sample (n=409) grouped by primary medical college affiliation. Grey bars signify specialties where proportions were representative of the medical specialist population when compared with reference data.[27] The black bar signifies a specialty which was over-represented (physicians; $p<0.0001$). White bars signify specialties which were under-represented: anaesthesiology ($p=0.002$), psychiatry ($p<0.0001$) and surgery ($p<0.0001$). The reference data did not include a classification for 'rural and remote medicine' so representativeness could not be determined for this specialty (pale grey bar).

116x56mm (300 x 300 DPI)

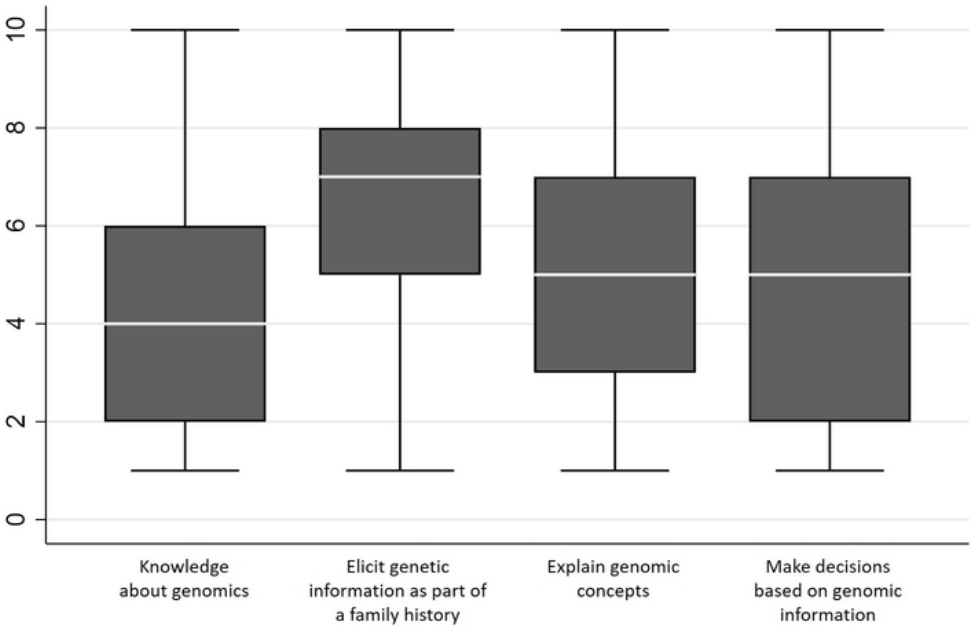

Figure 3: Average confidence about genomic concepts and skills on a scale of 1 'Not at all confident', 5 'Neutral' to 10 'Very confident' (n=273). Boxes represent the interquartile ranges with minimum and maximum value; medians are shown as white bars.

61x40mm (300 x 300 DPI)

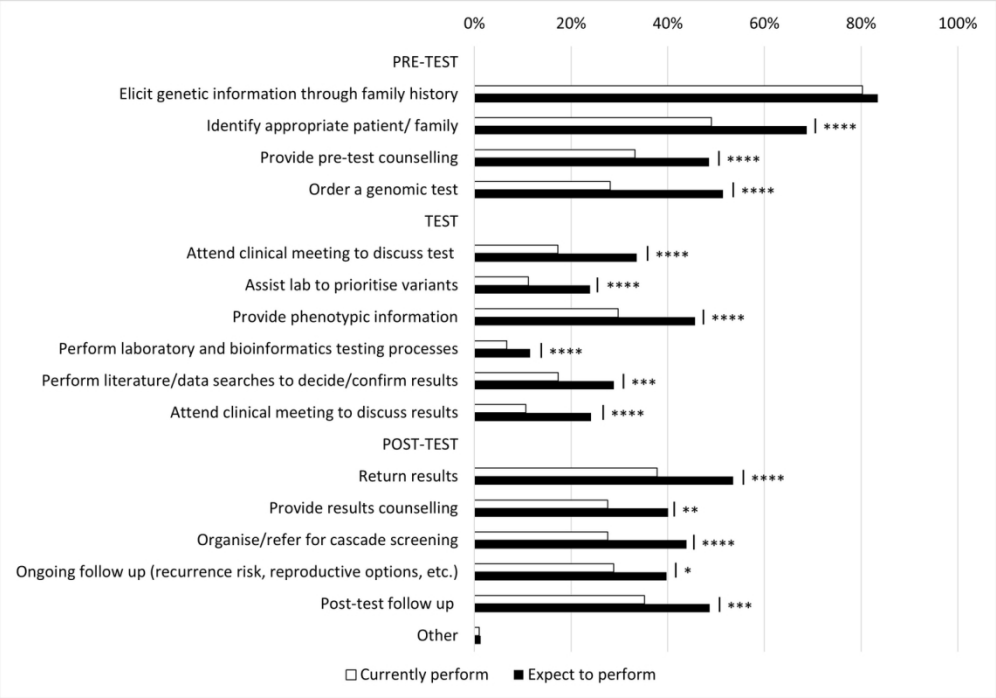

Figure 4: Steps in genomic testing that respondents (n=314) currently perform (blue bars) compared to steps they expect to perform in the future, if they had adequate support, education and training (black bars). Non-clinical steps are indicated by a. Differences between proportions for 'Currently perform' and 'Expect to perform' are indicated by *p=0.004, **p=0.001, ***p=0.0006, ****p<0.0001. The difference for the first step – Elicit genetic information through family history – was not significant (p=0.3). The full wording of each step is provided in Supplementary Table S5.

133x93mm (300 x 300 DPI)

Nisselle, King et al. Measuring physician practice, preparedness and preferences for genomic medicine: a national survey.

SUPPLEMENTARY MATERIALS

Figure S1. *Summary of survey attempts, responses and final sample for analysis*

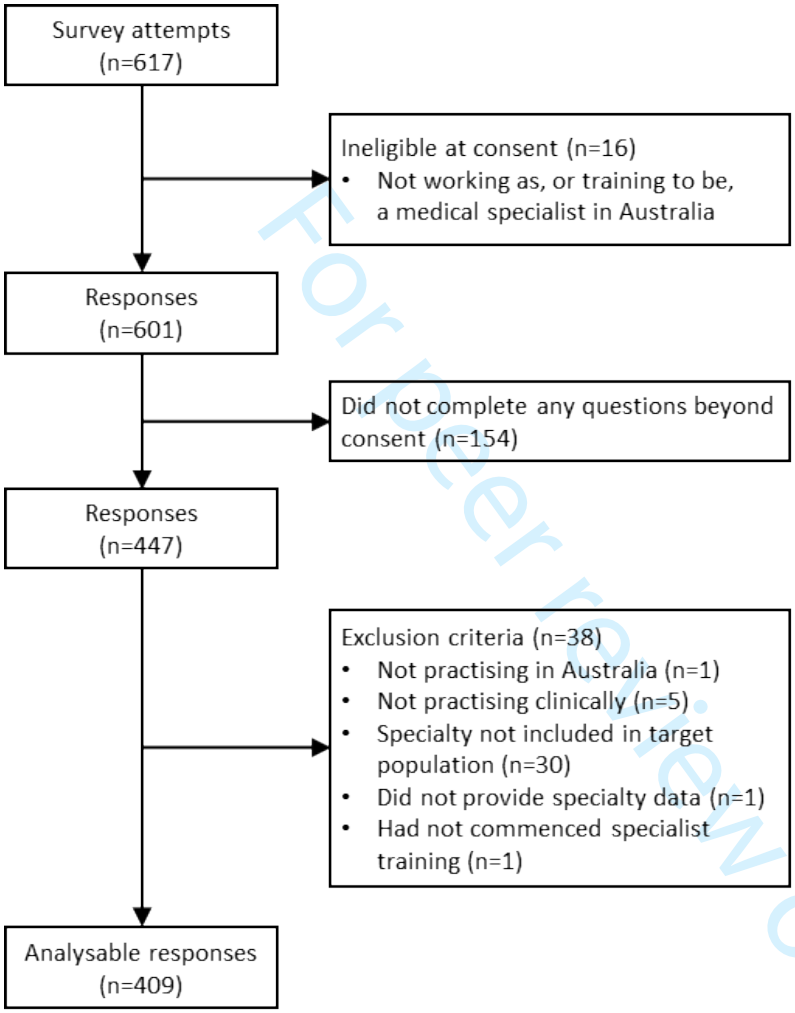

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Table S1. *Conditions for which genetic/genomic testing was covered by Medicare Benefit Scheme at the time of survey deployment in 2017.*[1]

Condition
1. Cytogenetics in general (pregnancies) and products of conception
2. Developmental delay
3. Peripheral neuropathy
4. Alport's Syndrome
5. Ataxia
6. Factor V Leiden Deficiency
7. Haemochromatosis
8. Polycythaemia/thrombocytopaenia
9. Drug toxicity (thiopurine)
10. Cystic fibrosis
11. Haematological malignancies
12. *BRCA* testing for breast/ovarian cancer
13. Leukemias
14. Mast cell disease/hypereosinophilia/eosinophil leukemia
15. *In situ* hybridisation tests for cancers
16. Von Hippel Lindau Syndrome (predisposition to various cancers)
17. Metastatic melanoma
18. Metastatic colorectal cancer
19. Metastatic adenocarcinoma stomach
20. Non-small cell lung cancer

[1]. *Australian Government Department of Health. Medicare Benefits Schedule Book. ISBN: 978-1-76007-375-3. Publications Number: 12289. Australian Government; 2019 [accessed 6 January 2021]. Available from: http://www.mbsonline.gov.au/.*

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Table ~~S1~~S2. *Examples of recoded open-text responses where a respondent selected 'Other (please specify……)' for a categorical question.*

Question	Open text response [ID, specialty]	Recoded category
[If contacted clinical genetics team or service in last 12 months]: Why did you contact your clinical genetics team or service? [a] Information about a suspected genetic condition [b] Advice on what type of genetic or genomic test to order [c] Advice on how to refer the patient to my clinical genetics team or service [d] Assistance with genetic counselling before the test [e] Assistance with genetic counselling after the test [f] Other (please specify)……	*"Referral"* [135, surgery] *"Facilitating genomic testing so that genetic counselling can be given to patient before test"* [145, paediatrics]	[c] [d]
[If did not contact clinical genetics team or service in last 12 months]: Why haven't you contacted your clinical genetics team or service? [a] Genetics and genomics are not relevant to my practice [b] I have not yet needed advice from a clinical genetics team or service in my practice [c] I can manage my patients without advice from a clinical genetics service [d] I'm not sure how to contact my clinical genetics team or service [e] I do not have access to a clinical genetics team or service [f] Other (please specify)……	*"My cohort of patients generally do not need genetic service input"* [129, gerontology] *"We do some of this inhouse"* [282, general medicine]	[a] [c]
Below is a list of some of the steps involved in genomic sequencing testing from pre-test to post-test *[see **Table S5**]*. Please indicate which steps you currently perform and which ones you expect to perform in the future if you had adequate education, training and support. If you selected "Other" step, please specify.	*"Going over letters and reports from genetics, explaining things again in context"* [221, paediatrics] *"I continue to see patients after their diagnostic test, which hopefully occurs as part of the evaluation of their condition"* [3, gerontology]	[k] [n]
What is/would be your preferred model for delivering a genomic sequencing test in an outpatient setting in your clinical practice, assuming you have appropriate education, training and funding?	*"Not relevant to my specialty"* [140, palliative medicine]	[d] [b]

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Question	Open text response [ID, specialty]	Recoded category
[a] You initiate testing and discuss results with patients/families [b] You initiate testing and discuss results with patients/families, with support from a clinical genetics team as needed [c] You refer to a clinical genetics team to initiate testing and discuss results with patients/families [d] You do not see, and do not expect to see, patients who would benefit from genomic testing [e] Unsure at this stage [f] Other (please specify)……	*"Same as for inpatient"* [109, palliative medicine; selected [b] for Inpatient response]	
[If selected 'yes' to genomics will impact practice within two years]: What areas will be impacted? [a] The way I practice medicine [b] My workload [c] Patient management [d] Other (please specify)……	*Clinical outcome and prognostications* [123, intensive care]	[c]
[If selected 'yes' to attending genomic professional development education or training in past year]: Was this: [a] In-house (internal) program/s [b] External program/s [c] Online training (webinar, MOOC, etc.) [d] Other (please specify)……	*"Recent commencement of multidisciplinary meeting"* [416, cardiology] *"International Clinical Cardiovascular Genetics conference"* [430, paediatrics]	[a] [b]

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Table S32. *Illustrative quotes from open-text survey comments.*

Domain	Quote
Current practice compared with future practice in genomic medicine *Q: Do you think genomics will impact your practice in the next 2 years?*	
Expect genomics *will* impact practice in next two years	*"Becoming increasingly available and of measurable significance"* [513, surgery] *"I expect it [genomics] will increasingly impact on the practice of medicine in terms of diagnoses, prognoses and treatment"* [281, paediatrics] *"Increased patient requests"* [271, obstetrics and gynaecology]
Expect genomics will *not* impact practice in next two years	*"Emergency department have more important competing interests in treatment delivery to patients"* [383, emergency medicine] *"Timeframe remains too short to see this implemented in a regional area"* [535, anaesthesiology]
Preferred future models for delivering genomic medicine *Q: What is/would be your preferred model for delivering a genomic sequencing test* in your clinical practice, assuming you have appropriate education, training and funding?*[1]	
Referring to genetics services to initiate testing and discuss results	*"For my patients and practice, having an accessible [genetics] clinic for this would be best. I would be very keen to be involved as far as possible, but do not have time to keep up with this rapidly developing field. I would like to be invited to my patients' MDT [multidisciplinary team] discussions. That way I am involved, and have the knowledge to answer follow-up and clarification questions. It would also be a way to increase my knowledge"* [100, nephrologist]
Delivering testing with support from genetics services	*"[Genetics support for both inpatients and outpatients] would streamline the process, improve access and possibly reduce Clinical Genetics load by filtering patients and families I can manage while they still see the patients or results beyond my expertise"* [220, paediatrics, community child health] *"We (clinicians) may be more familiar with the disease phenotype than the Genetics team"* [33, immunopathology] *"Clinicians should be able to initiate testing but will need support with interpretation and counselling, particularly initially until genomic medicine is core practice"* [350, palliative medicine]
Initiating genomic testing themselves with no support from genetics	*"I expect to be able to manage simpler conditions/results, with access to more specialist input when needed"* [129, gerontology]
Will not see patients who would benefit from genomic sequencing tests	*"Relevance to decision making in real time"* [459, emergency medicine] *"Not sure of any relevance to my practice"* [541, anaethesiology]

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Domain	Quote
Preparedness for genomic medicine and preferences for future education *Q: Do you feel prepared to use genomic sequencing testing* in your practice?*	
	"I have little to no training in genetics and genomic medicine. We had a total of 4 genetics lectures at medical school, and there is limited assessment of genetics/genomics in the [college fellowship examination]. Genomic testing is not routinely used in our practice" [73, intensive care]
	"My knowledge of this whole area is woefully inadequate. I can cope with karyotype analysis and testing for CF [cystic fibrosis]. I can also discuss prenatal diagnosis options, PGT-A [pre-implantation genetic testing] and expanded carrier testing but that's about it…… It clearly will be an important part of medical practice in the future" [213, obstetrics and gynaecology]
	"I'm happy to do [genomic testing] but need training." [342, surgery]
	"Need further information, education on who would best benefit from this test, how to consent for it and then how to interpret results" [414, general paediatrics]
Preferences for learning about genomics	
Q: What would help improve your confidence?[2]	*"Further training in counselling [would improve my confidence]–in ability to explain concepts and then clinical implications and follow-on from this"* [27, paediatric neurology]
Q: Please explain why you do not expect to perform the selected steps [involved in genomic sequencing testing]*[3]	*"Would welcome some education on use of these tests in orthopaedics"* [391, surgery]

*[1] Full question provided in **Table S2~~Table S1~~**; [2] following the question on confidence in four genomic knowledge and skills areas, presented in **Figure 1**; [3] following the question on steps involved in genomic sequencing testing, presented in **Error! Reference source not found.** and **Table S2~~Table S1~~**.*

** Definitions were provided for these terms*

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Table S3. *The full wording of each step involved in genomic testing as presented in the survey.*[1]

Pre-test
[a] Eliciting information about genetic conditions as part of a family or medical history
[b] Identifying a patient suitable for a genomic test
[c] Pre-test counselling to assist in making an informed decision, e.g., genetics, test limitations, variants of uncertain/unknown significance*, incidental/secondary findings, unexpected non-paternity or consanguinity
[d] Ordering a genomic test for a patient
Test
[e] Attending multidisciplinary team meeting to discuss the genomic test (e.g., intake meeting)
[f] Assisting the lab to narrow down the genes of interest (creating a gene list to prioritise variant analysis)[2]
[g] Providing phenotypic information to the lab to prioritise variant analysis
[h] Laboratory and bioinformatics testing processes[2]
[i] Searching the literature and databases for evidence of variant pathogenicity*,[2]
[j] Attending a multidisciplinary team meeting to discuss variant prioritisation*, interpretation and classification*
Post-test
[k] Provide test results to patients/ families
[l] Provide genetic counselling to patients/families, e.g., explain variants of uncertain/unknown significance*, incidental/secondary findings, unexpected non-paternity or consanguinity
[m] Organising/ referring for further testing of family members if required, e.g., cascade testing or segregation studies
[n] Ongoing management of the patient, e.g., clarify recurrence risk and discuss reproductive planning options
[o] Post-test follow up of patient to check understanding of result/ ask any additional questions
[p] Other (please specify).....

[1] The survey is available as supplementary material in [24]; [2] These steps are considered non-clinical, i.e., laboratory;

** Definitions were provided for these terms*

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Table S2. *Participant-reported funding for genomic tests ordered in the past year.*[1]

	Microarray n=112	**Gene panel** n=112	**Exome/genome sequencing** n=50
Medicare Benefit Scheme	48.2%	17.0%	2.0%[2]
Institute/hospital	41.1%	52.6%	44.0%
State government	13.4%	17.0%	12.0%
Research grant	2.7%	11.6%	60.0%
Patient	12.5%	24.1%	4.0%
Unsure	11.6%	8.0%	6.0%

[1] *Respondents could select more than one funding source per test type.*

[2] *At the time of the survey the MBS scheme did not fund E/GS, so this response (n=1) is incorrect.*

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Table S5. *The full wording of each step involved in genomic testing as presented in the survey.*[1]

Pre-test
[a] Eliciting information about genetic conditions as part of a family or medical history
[b] Identifying a patient suitable for a genomic test
[c] Pre-test counselling to assist in making an informed decision, e.g., genetics, test limitations, variants of uncertain/unknown significance*, incidental/secondary findings, unexpected non-paternity or consanguinity
[d] Ordering a genomic test for a patient
Test
[e] Attending multidisciplinary team meeting to discuss the genomic test (e.g., intake meeting)
[f] Assisting the lab to narrow down the genes of interest (creating a gene list to prioritise variant analysis)[2]
[g] Providing phenotypic information to the lab to prioritise variant analysis
[h] Laboratory and bioinformatics testing processes[2]
[i] Searching the literature and databases for evidence of variant pathogenicity*,[2]
[j] Attending a multidisciplinary team meeting to discuss variant prioritisation*, interpretation and classification*
Post-test
[k] Provide test results to patients/ families
[l] Provide genetic counselling to patients/families, e.g., explain variants of uncertain/unknown significance*, incidental/secondary findings, unexpected non-paternity or consanguinity
[m] Organising/ referring for further testing of family members if required, e.g., cascade testing or segregation studies
[n] Ongoing management of the patient, e.g., clarify recurrence risk and discuss reproductive planning options
[o] Post-test follow up of patient to check understanding of result/ ask any additional questions
[p] Other (please specify)……

[1] The survey is available as supplementary material in [24]; [2] These steps are considered non-clinical, i.e., laboratory;

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SUPPLEMENTARY MATERIALS

Figure S1. *Summary of survey attempts, responses and final sample for analysis*

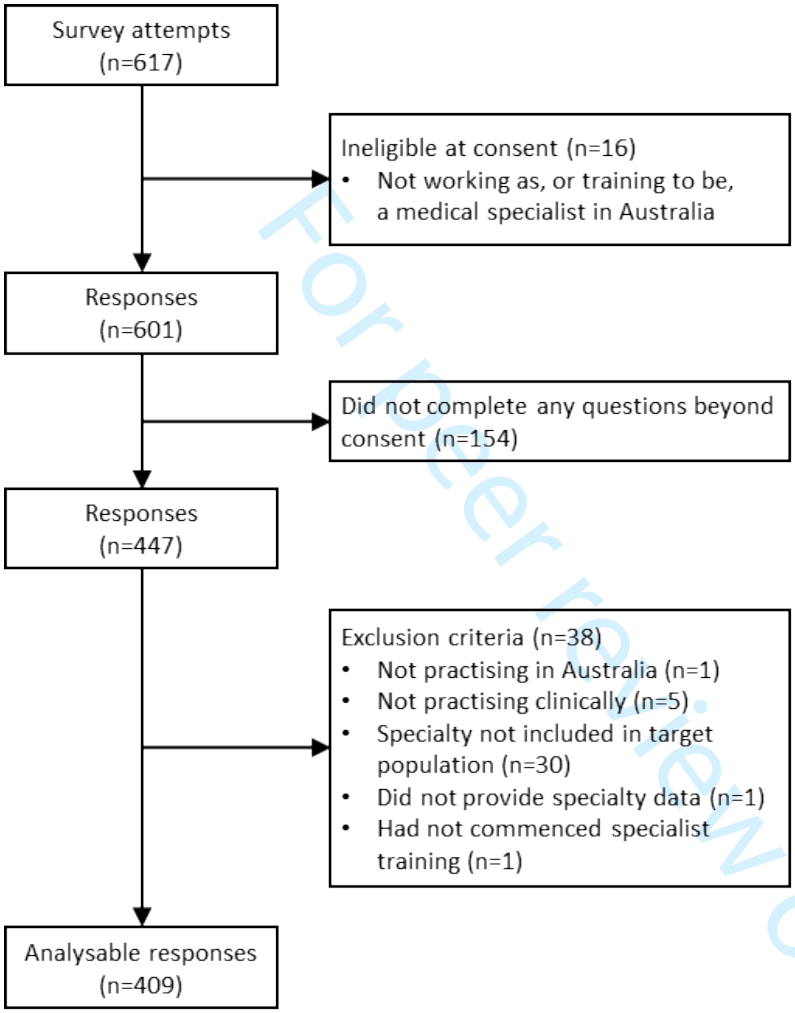

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Table S1. *Conditions for which genetic/genomic testing was covered by Medicare Benefit Scheme at the time of survey deployment in 2017.*[1]

Condition
1. Cytogenetics in general (pregnancies) and products of conception
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6. Factor V Leiden Deficiency
7. Haemochromatosis
8. Polycythaemia/thrombocytopaenia
9. Drug toxicity (thiopurine)
10. Cystic fibrosis
11. Haematological malignancies
12. *BRCA* testing for breast/ovarian cancer
13. Leukemias
14. Mast cell disease/hypereosinophilia/eosinophil leukemia
15. *In situ* hybridisation tests for cancers
16. Von Hippel Lindau Syndrome (predisposition to various cancers)
17. Metastatic melanoma
18. Metastatic colorectal cancer
19. Metastatic adenocarcinoma stomach
20. Non-small cell lung cancer

[1]. *Australian Government Department of Health. Medicare Benefits Schedule Book. ISBN: 978-1-76007-375-3. Publications Number: 12289. Australian Government; 2019 [accessed 6 January 2021]. Available from: http://www.mbsonline.gov.au/.*

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Table S2. *Examples of recoded open-text responses where a respondent selected 'Other (please specify……)' for a categorical question.*

Question	Open text response [ID, specialty]	Recoded category
[If contacted clinical genetics team or service in last 12 months]: Why did you contact your clinical genetics team or service? [a] Information about a suspected genetic condition [b] Advice on what type of genetic or genomic test to order [c] Advice on how to refer the patient to my clinical genetics team or service [d] Assistance with genetic counselling before the test [e] Assistance with genetic counselling after the test [f] Other (please specify)……	*"Referral"* [135, surgery]	[c]
	"Facilitating genomic testing so that genetic counselling can be given to patient before test" [145, paediatrics]	[d]
[If did not contact clinical genetics team or service in last 12 months]: Why haven't you contacted your clinical genetics team or service? [a] Genetics and genomics are not relevant to my practice [b] I have not yet needed advice from a clinical genetics team or service in my practice [c] I can manage my patients without advice from a clinical genetics service [d] I'm not sure how to contact my clinical genetics team or service [e] I do not have access to a clinical genetics team or service [f] Other (please specify)……	*"My cohort of patients generally do not need genetic service input"* [129, gerontology]	[a]
	"We do some of this inhouse" [282, general medicine]	[c]
Below is a list of some of the steps involved in genomic sequencing testing from pre-test to post-test *[see **Table S5**]*. Please indicate which steps you currently perform and which ones you expect to perform in the future if you had adequate education, training and support. If you selected "Other" step, please specify.	*"Going over letters and reports from genetics, explaining things again in context"* [221, paediatrics]	[k]
	"I continue to see patients after their diagnostic test, which hopefully occurs as part of the evaluation of their condition" [3, gerontology]	[n]
What is/would be your preferred model for delivering a genomic sequencing test in an outpatient setting in your clinical practice, assuming you have appropriate education, training and funding?	*"Not relevant to my specialty"* [140, palliative medicine]	[d]
		[b]

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Question	Open text response [ID, specialty]	Recoded category
[a] You initiate testing and discuss results with patients/families [b] You initiate testing and discuss results with patients/families, with support from a clinical genetics team as needed [c] You refer to a clinical genetics team to initiate testing and discuss results with patients/families [d] You do not see, and do not expect to see, patients who would benefit from genomic testing [e] Unsure at this stage [f] Other (please specify)……	*"Same as for inpatient"* [109, palliative medicine; selected [b] for Inpatient response]	
[If selected 'yes' to genomics will impact practice within two years]: What areas will be impacted? [a] The way I practice medicine [b] My workload [c] Patient management [d] Other (please specify)……	*Clinical outcome and prognostications* [123, intensive care]	[c]
[If selected 'yes' to attending genomic professional development education or training in past year]: Was this: [a] In-house (internal) program/s [b] External program/s [c] Online training (webinar, MOOC, etc.) [d] Other (please specify)……	*"Recent commencement of multidisciplinary meeting"* [416, cardiology] *"International Clinical Cardiovascular Genetics conference"* [430, paediatrics]	[a] [b]

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Table S3. *Illustrative quotes from open-text survey comments.*

Domain	Quote
Current practice compared with future practice in genomic medicine *Q: Do you think genomics will impact your practice in the next 2 years?*	
Expect genomics *will* impact practice in next two years	*"Becoming increasingly available and of measurable significance"* [513, surgery] *"I expect it [genomics] will increasingly impact on the practice of medicine in terms of diagnoses, prognoses and treatment"* [281, paediatrics] *"Increased patient requests"* [271, obstetrics and gynaecology]
Expect genomics will *not* impact practice in next two years	*"Emergency department have more important competing interests in treatment delivery to patients"* [383, emergency medicine] *"Timeframe remains too short to see this implemented in a regional area"* [535, anaesthesiology]
Preferred future models for delivering genomic medicine *Q: What is/would be your preferred model for delivering a genomic sequencing test* in your clinical practice, assuming you have appropriate education, training and funding?*[1]	
Referring to genetics services to initiate testing and discuss results	*"For my patients and practice, having an accessible [genetics] clinic for this would be best. I would be very keen to be involved as far as possible, but do not have time to keep up with this rapidly developing field. I would like to be invited to my patients' MDT [multidisciplinary team] discussions. That way I am involved, and have the knowledge to answer follow-up and clarification questions. It would also be a way to increase my knowledge"* [100, nephrologist]
Delivering testing with support from genetics services	*"[Genetics support for both inpatients and outpatients] would streamline the process, improve access and possibly reduce Clinical Genetics load by filtering patients and families I can manage while they still see the patients or results beyond my expertise"* [220, paediatrics, community child health] *"We (clinicians) may be more familiar with the disease phenotype than the Genetics team"* [33, immunopathology] *"Clinicians should be able to initiate testing but will need support with interpretation and counselling, particularly initially until genomic medicine is core practice"* [350, palliative medicine]
Initiating genomic testing themselves with no support from genetics	*"I expect to be able to manage simpler conditions/results, with access to more specialist input when needed"* [129, gerontology]
Will not see patients who would benefit from genomic sequencing tests	*"Relevance to decision making in real time"* [459, emergency medicine] *"Not sure of any relevance to my practice"* [541, anaethesiology]

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Domain	Quote
Preparedness for genomic medicine and preferences for future education	
Q: Do you feel prepared to use genomic sequencing testing in your practice?*	
	"I have little to no training in genetics and genomic medicine. We had a total of 4 genetics lectures at medical school, and there is limited assessment of genetics/genomics in the [college fellowship examination]. Genomic testing is not routinely used in our practice" [73, intensive care]
	"My knowledge of this whole area is woefully inadequate. I can cope with karyotype analysis and testing for CF [cystic fibrosis]. I can also discuss prenatal diagnosis options, PGT-A [pre-implantation genetic testing] and expanded carrier testing but that's about it…… It clearly will be an important part of medical practice in the future" [213, obstetrics and gynaecology]
	"I'm happy to do [genomic testing] but need training." [342, surgery]
	"Need further information, education on who would best benefit from this test, how to consent for it and then how to interpret results" [414, general paediatrics]
Preferences for learning about genomics	
Q: What would help improve your confidence?[2]	*"Further training in counselling [would improve my confidence]–in ability to explain concepts and then clinical implications and follow-on from this"* [27, paediatric neurology]
Q: Please explain why you do not expect to perform the selected steps [involved in genomic sequencing testing]*[3]	*"Would welcome some education on use of these tests in orthopaedics"* [391, surgery]

*[1] Full question provided in **Table S2**; [2] following the question on confidence in four genomic knowledge and skills areas, presented in **Figure 1**; [3] following the question on steps involved in genomic sequencing testing, presented in **Error! Reference source not found.** and **Table S2.***

** Definitions were provided for these terms*

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Table S4. *Participant-reported funding for genomic tests ordered in the past year.*[1]

	Microarray n=112	**Gene panel** n=112	**Exome/genome sequencing** n=50
Medicare Benefit Scheme	48.2%	17.0%	2.0%[2]
Institute/hospital	41.1%	52.6%	44.0%
State government	13.4%	17.0%	12.0%
Research grant	2.7%	11.6%	60.0%
Patient	12.5%	24.1%	4.0%
Unsure	11.6%	8.0%	6.0%

[1] *Respondents could select more than one funding source per test type.*

[2] *At the time of the survey the MBS scheme did not fund E/GS, so this response (n=1) is incorrect.*

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Table S5. *The full wording of each step involved in genomic testing as presented in the survey.*[1]

Pre-test
[a] Eliciting information about genetic conditions as part of a family or medical history
[b] Identifying a patient suitable for a genomic test
[c] Pre-test counselling to assist in making an informed decision, e.g., genetics, test limitations, variants of uncertain/unknown significance*, incidental/secondary findings, unexpected non-paternity or consanguinity
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[g] Providing phenotypic information to the lab to prioritise variant analysis
[h] Laboratory and bioinformatics testing processes[2]
[i] Searching the literature and databases for evidence of variant pathogenicity*[,2]
[j] Attending a multidisciplinary team meeting to discuss variant prioritisation*, interpretation and classification*
Post-test
[k] Provide test results to patients/ families
[l] Provide genetic counselling to patients/families, e.g., explain variants of uncertain/unknown significance*, incidental/secondary findings, unexpected non-paternity or consanguinity
[m] Organising/ referring for further testing of family members if required, e.g., cascade testing or segregation studies
[n] Ongoing management of the patient, e.g., clarify recurrence risk and discuss reproductive planning options
[o] Post-test follow up of patient to check understanding of result/ ask any additional questions
[p] Other (please specify)……

[1] The survey is available as supplementary material in [24]; [2] These steps are considered non-clinical, i.e., laboratory;

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Reporting checklist for cross sectional study.

Based on the **STROBE** cross sectional guidelines.

Instructions to authors

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		Reporting Item	*Page*
Title and abstract			
Title	#1a	Indicate the study's design with a commonly used term in the title or the abstract	1
Abstract	#1b	Provide in the abstract an informative and balanced summary of what was done and what was found	2
Introduction			
Background / rationale	#2	Explain the scientific background and rationale for the investigation being reported	4
Objectives	#3	State specific objectives, including any prespecified hypotheses	4
Methods			
Study design	#4	Present key elements of study design early in the paper	5-6
Setting	#5	Describe the setting, locations, and relevant dates, including periods of recruitment, exposure, follow-up, and data collection	5-6
Eligibility criteria	#6a	Give the eligibility criteria, and the sources and methods of selection of participants.	5-6
	#7	Clearly define all outcomes, exposures, predictors, potential confounders, and effect modifiers. Give diagnostic criteria, if applicable	5
Data sources / measurement	#8	For each variable of interest give sources of data and details of methods of assessment (measurement). Describe comparability of assessment methods	5-6

		Reporting Item	*Page*
		if there is more than one group. Give information separately for exposed and unexposed groups if applicable.	
Bias	#9	Describe any efforts to address potential sources of bias	5-6, 12
Study size	#10	Explain how the study size was arrived at	5-6, 12
Quantitative variables	#11	Explain how quantitative variables were handled in the analyses. If applicable, describe which groupings were chosen, and why	6-10
Statistical methods	#12a	Describe all statistical methods, including those used to control for confounding	6
Statistical methods	#12b	Describe any methods used to examine subgroups and interactions	6
Statistical methods	#12c	Explain how missing data were addressed	7
Statistical methods	#12d	If applicable, describe analytical methods taking account of sampling strategy *Not required as sampling strategy was same across single cohort*	N/A
Statistical methods	#12e	Describe any sensitivity analyses *Not required*	N/A
Results			
Participants	#13a	Report numbers of individuals at each stage of study—eg numbers potentially eligible, examined for eligibility, confirmed eligible, included in the study, completing follow-up, and analysed. Give information separately for for exposed and unexposed groups if applicable.	7-10, 17-22, vi
Participants	#13b	Give reasons for non-participation at each stage	7
Participants	#13c	Consider use of a flow diagram *Not required*	N/A
Descriptive data	#14a	Give characteristics of study participants (eg demographic, clinical, social) and information on exposures and potential confounders. Give information separately for exposed and unexposed groups if applicable.	7-8, 17-19
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Outcome data	#15	Report numbers of outcome events or summary measures. Give information separately for exposed and unexposed groups if applicable.	7-10, 17-22, i-vi
Main results	#16a	Give unadjusted estimates and, if applicable, confounder-adjusted estimates and their precision (eg, 95% confidence interval). Make clear which confounders were adjusted for and why they were included	N/A

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		Reporting Item	*Page*
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Main results	#16b	Report category boundaries when continuous variables were categorized	8, 17
Main results	#16c	If relevant, consider translating estimates of relative risk into absolute risk for a meaningful time period *Not relevant*	N/A
Other analyses	#17	Report other analyses done—e.g., analyses of subgroups and interactions, and sensitivity analyses	7-10, 17-22, i-vi
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Measuring physician practice, preparedness and preferences for genomic medicine: a national survey

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Measuring physician practice, preparedness and preferences for genomic medicine: a national survey

Amy Nisselle,[1,2,3,*,†] Emily King,[1,2,3*] Belinda McClaren,[1,2,3,] Monika Janinski,[1,2] Sylvia Metcalfe[1,2,3] and Clara Gaff[1,2,3] on behalf of the Australian Genomics Workforce & Education Working Group

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Abstract

Objective: Even as genomic medicine is implemented globally, there remains a lack of rigorous, national assessments of physicians' current genomic practice and continuing genomics education needs. The aim of this study was to address this gap.

Design: A cross-sectional survey, informed by qualitative data and behaviour change theory, to assess the current landscape of Australian physicians' genomic medicine practice, perceptions of proximity and individual preparedness, and preferred models of practice and continuing education. The survey was advertised nationally through 10 medical colleges, 24 societies, 62 hospitals, social media, professional networks and snowballing.

Results: 409 medical specialists across Australia responded, representing 30 specialties (majority paediatricians, 20%), from mainly public hospitals (70%) in metropolitan areas (75%). Half (53%) had contacted their local genetics services and half (54%) had ordered or referred for a gene panel or exome/genome sequencing (E/GS) test in the last year. Two-thirds (67%) think genomics will soon impact their practice, with a significant preference for models that involved genetics services (p<0.0001). Currently, respondents mainly perform tasks associated with pre-test family history taking and counselling, but more respondents expect to perform tasks at all stages of testing in the future, including tasks related to the test itself, and reporting results. While a third (34%) recently completed education in genomics, only a quarter (25%) felt prepared to practice. Specialists would like (more) education, particularly on genomic technologies and clinical utility, and prefer this to be through varied educational strategies.

Conclusions: This survey provides data from a breadth of physician specialties that can inform models of genetic service delivery and genomics education. The findings support education providers designing and delivering education that best meet learner needs to build a competent, genomic-literate workforce. Further analyses are underway to characterise early adopters of genomic medicine to inform strategies to increase engagement.

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Strengths and limitations of this study

- The survey tool is based in behavioural change theory and developed from empirical data to capture patterns of genomic practice and preferences, allowing comparisons across different settings and change over time.
- We employed an extensive, multi-staged and overlapping recruitment strategy at a national level to reach as many Australian medical specialists and trainees as possible.
- We successfully gathered data from over 30 specialties, the broadest sample reported in the literature to date.
- Our sample is still relatively small, and over-represented for older specialists and those working in rural and remote areas, which may influence the findings.
- Our study is the first to investigate the genomics education and training needs and preferences of a national sample of a broad range of medical specialties.

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Introduction

Genomic sequencing is shifting from the realm of research to healthcare.[1] A recent review identified five models for the provision of genetic testing globally, including genetics services led by geneticists, referral by primary-care physicians to genetics services, and medical specialist-led testing.[2] The shortage of a specialist genetic workforce suggests that medical specialist-led testing will be necessary.[3][4] A scoping review of genetic specialist workforces internationally emphasised the need for a medical specialist-led model, noting education as a driver of workforce capacity.[5]

A national alliance of over 80 partner organisations, Australian Genomics, formed in 2016 to conduct research supporting adoption of genomics into Australian healthcare.[6] At that time, microarray analysis and a limited number of single gene tests were reimbursed through the federally-funded Medicare Benefit Scheme (MBS). Genomic sequencing tests were largely available through research studies or patient funding until 2020, when exome/genome sequencing (E/GS) for certain conditions was included on the MBS.[7]

Despite national initiatives driving the use of germline genomic tests by medical specialists not qualified in genetics, there are indications that physicians may prefer to refer to genetics services.[8, 9] Cumulative evidence indicates a lack of physician confidence in genomic medicine and low rates of clinical adoption of genomic testing.[10] Studies investigating practice and preparedness span specialties and countries: Dutch cardiologists,[11] European obstetricians and paediatricians,[12] Wisconsin physicians,[13] British gastroenterologists,[14] Australian intensivists,[8] and neurologists worldwide.[15] However, there are no national studies surveying a range of specialties.

Education strategies have been proposed or implemented to support medical professionals' genomic medicine knowledge and skills.[16, 17] Following medical school training,[18, 19] continuing professional development (CPD), whether accredited or not, aims to supplement knowledge and skills for those already in practice.[20, 21] To inform Australian national strategy and local development of genomics CPD, a needs assessment inclusive of a multiple specialties across diverse contexts is required. We previously reported development of a survey underpinned by qualitative data and an empirically-derived framework of behaviour change in which capability, opportunity and motivation influence, and are influenced by, behaviour (the COM-B model).[22]

Here we describe comprehensive deployment of this survey nationally to multiple medical specialities. We present a snapshot of the current landscape of Australian specialists' genomic medicine practice, perceptions of proximity of genomic medicine and individual preparedness, and preferred models of practice and continuing education.

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Methods

In Australia, after obtaining a medical degree, doctors undertake specialty training.[23] This typically involves completing three years of basic training ('Basic Trainee') followed by three years of advanced training ('Advanced Trainee'). Medical colleges provide the training relevant to the medical specialty, e.g., the Royal Australasian College of Surgeons trains surgeons, the Royal Australasian College of Physicians trains physicians, etc. Exposure to genetics/genomics varies across training programs. After successful completion of final college examinations, they become a Fellow of the relevant medical college. Recognising that the term 'physician' has different meanings in different countries, here we define 'physicians' as doctors whose primary affiliation is with the Royal Australasian College of Physicians . Medical professionals may work in public hospitals, which are the responsibility of State governments, and/or privately. Patients receive some reimbursement for private consultations and specified pathology tests through the Federal Government's MBS. At the time of the survey, there were 20 genetics conditions for which tests were reimbursed through the MBS (see **Supplementary Table S1**). Clinical genetics services provide screening, diagnostic and genetic counselling services to patients on referral by a medical practitioner. They are based primarily in publicly-funded hospitals and staffed by health professionals trained in genetics (e.g., clinical geneticists, genetic counsellors. Here we focus on the non-genetic medical workforce and as such define 'medical specialists' as medical doctors who are trained or in training for a specialty other than clinical genetics. We excluded general practitioners (family physicians) who practise general medicine in the community and genetic specialists (e.g., clinical geneticists and genetic counsellors) as separate studies were conducted for those subspecialties.[4](Cusack et al., *Australian Journal of General Practice*, in press). We also excluded radiologists and pathologists as in Australia they typically perform investigations rather than requesting genomic tests, and oncologists, as they are the focus of other ongoing national studies.

Details of survey development, domains and the full set of questions have been reported elsewhere.[20, 24] In brief, the survey is informed by the COM-B model and includes 28 questions across five key domains: personal characteristics, current practice with genomic medicine, perception of preparedness to practice genomic medicine, perception of how proximal genomic medicine is to clinical practice, and preferences for future models of practice and education. We defined 'genomic medicine' as the use of testing that investigates many regions of the genome at once, such as gene panels and E/GS, but excluding non-invasive prenatal testing using sequencing technologies. The scope of the survey was testing to investigate genetic conditions. The survey was deployed

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electronically from February to September 2019 using REDCap (Research Electronic Data Capture) software hosted at the Murdoch Children's Research Institute.[25]

This project received ethics approval from the University of Melbourne, Melbourne, Australia (HREC number: 1646785.10). Respondents provided consent by completing the initial screening and consent question.

Recruitment

Inclusion criteria: medical specialists were eligible to complete the survey if they had commenced or completed their specialist training and were currently practising clinically in Australia.

Recruitment was staged through:

- Relevant medical colleges (Mar–Jun 2019) and societies/associations (Apr–Jun 2019).
- Hospitals (Jun–Oct 2019). 132 hospitals were identified from the 'MyHospitals' search tool on the Australian Institute of Health and Welfare website[26] to represent both public and private hospitals in metropolitan, regional and rural settings across all Australian states.
- Social media (Jun–Jul 2019). Three tweets were posted on the Australian Genomics Twitter account (https://twitter.com/AusGenomics) over 10 business days, then this process was repeated twice, with approximately one week between each cluster of tweets. Content referenced specific survey questions or preliminary data to pique interest of potential participants. For example, *'Early survey results suggest that even though medical specialists are ordering #genomictests for their #patients, many don't feel #prepared for #genomicmedicine. We want to know how you feel [LINK]'* or *'Do you feel ready for #genomics in #clinicalpractice? We want to hear from Australian medical specialists [LINK]'*.
- Investigator networks of national and state-based genomics initiatives, Australian Genomics and Melbourne Genomics (Jul 2019).

Medical colleges, societies and hospitals circulated information about the study to their membership or staff using regular communication channels, e.g., newsletters, e-bulletins, emails, etc. Information was circulated up to three times per organisation, dependent on advertising charges, perceived responder burden and/or internal timelines. The information included a brief description of the study, ethics approval and a link to access the online survey. Recruitment also included professional networks and snowball sampling throughout, with all contacts asked to retweet Australian Genomics tweets if possible. All respondents were asked to share the survey with relevant colleagues.

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Data cleaning and analysis

Data were exported to, cleaned and then analysed in Stata 16.0. Cleaning involved removing surveys completed by ineligible respondents or surveys with no data beyond demographic questions. For analysis, career stage was grouped into Basic Trainee, Advanced Trainee or Fellow, as defined above. Specialists were grouped according to self-reported primary college affiliation. All categorical questions included an open-ended text option for 'Other'; qualitative data provided for these questions were reviewed by three researchers (AN, EK, MJ) and recoded into existing response categories if possible (see **Supplementary Table S2** for examples). Representative quotes are provided in **Supplementary Table S3** for illustrative purposes where they enhance the understanding of the quantitative results.

Descriptive and inferential statistics were used to analyse the data, including two-sample tests of proportions, chi-square or Fisher's exact tests as appropriate to data characteristics. A *p* value of <0.05 was considered significant. When determining representativeness of the sample, data were referenced against Medical Board of Australia Registrant data,[27] the National Medical Training Advisory Network, and the National Health Workforce Dataset.[28]

Patient and public involvement

There was no patient or public involvement in this research.

Results

Recruitment and response rates

As shown in **Figure 1**, recruitment strategies were staggered and overlapping from March to October 2019. All 10 Australian medical colleges and 24 of 55 medical societies/associations approached agreed to advertise the survey. Of 132 health networks[1] and hospitals contacted,[29] 62 agreed to advertise the survey (67.6% of metropolitan hospitals and 42.9% of remote hospitals), which was subsequently shared with staff at a total of 74 hospitals. There were an estimated 37,000 trainees and fellows in our target specialty audiences at the time of the survey.[27] However, using diverse recruitment approaches that could target one individual in several ways and at several time points meant that it was not possible to determine how many medical specialists were aware of the survey during the recruitment period.

[1] Health networks are functional or geographical groups of Australian public hospitals defined by the relevant State Government.

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Sample characteristics and representativeness

Of 617 attempts at survey responses, 54 did not meet the inclusion criteria and 154 did not complete any questions beyond consent to participate (see **Supplementary Figure S1** for detail). A total of 409 responses were therefore included in analyses. Totals differ across questions due to opportunity to provide more than one response, missing data or attrition; where this has occurred, the denominator has been described.

Table 1 presents respondent demographics compared with reference data from the Medical Board of Australia,[27] the National Medical Training Advisory Network[30, 31] and the National Health Workforce Dataset.[28] Our sample had slightly less males (p=0.039), was under-represented for 25–34 year olds (p<0.0001), and over-represented for 55–64 year olds (p<0.0001). As would be expected from this age bias, there was a smaller proportion of Basic and Advanced Trainees than expected from the reference data and a larger proportion of Fellows (p<0.0001). Our sample was broadly representative of primary work locations of medical specialists across Australia. Of the eight Australian states and territories, one was over-represented (Australian Capital Territory; p<0.0001) and two were under-represented (South Australia; p=0.028); Western Australia; p=0.032). Although three-quarters of respondents worked in a major city, those working in remote regions were significantly over-represented in our sample (p=0.0018). The majority of respondents were primarily employed at public hospitals or healthcare providers. A quarter of respondents had been involved in a genomics research project in the last 5 years (n=96, 24.7%). Of these, respondents were involved in clinical (83.3%), laboratory (49.0%), bioinformatics (15.6%) and/or social science (6.3%) projects. Only 7.2% of respondents indicated that they were affiliated with any state- or federally-funded genomic health alliances. **Error! Reference source not found.** describes proportions of respondent specialties, compared with the proportions expected from reference data.[27] The largest group of respondents were physicians, totalling 232 (56.7%) responses. Our sample was representative of most specialties with some exceptions: there were more physicians (p<0.0001 and fewer anaesthetists (p=0.002), psychiatrists (p<0.0001) and surgeons (p=0.0001).

Current practice in genomic medicine

Respondents (n=387) answered a series of questions about their current practice in genomic medicine. Just over half of respondents had contacted their local genetics service in the last 12 months (n=203, 52.5%), although this was relatively infrequent, with a third of these 203 respondents indicating this was once or twice in the last 12 months (36.6%). The main reasons for contacting genetics services included: seeking information about a suspected genetic condition (48.0%), advice on how to refer a patient (42.6%) and choosing which genetic or genomic test to order (38.1%). Of those who had not contacted clinical genetics, the majority indicated that this was because they had not yet needed advice (73.5%).

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Over half of respondents (n=208, 53.9%) had engaged in genomic sequencing testing in the last 12 months by either ordering a gene panel or E/GS, or referring a patient to a genetics service for those tests. Nearly a third of respondents (n=121, 31.3%) had ordered at least one of these tests, with 29.0% (n=112) ordering a gene panel and 13.0% (n=50) ordering E/GS. When asked about frequency of ordering each test in the previous year, the most common response was once or twice for both gene panels (n=42/112, 37.5%) and E/GS (n=23/50, 46.0%). In contrast, 112 respondents (29.0%) had ordered a microarray in the previous year, most commonly monthly (n=41/112, 36.6%). Funding for tests varied (**Supplementary Table S4**), with microarray tests often funded by the MBS, gene panel tests by the institute/hospital, and E/GS tests by research grants. Overall, 63.3% of respondents (n=245/387) had engaged in genetics/genomics in one or more ways: contacting their genetics service, or ordering or referring for a microarray, gene panel or E/GS test.

Respondents were asked to reflect on their confidence about genomic concepts and skills (**Error! Reference source not found.**). Medical specialists reported the highest level of confidence when taking a family history to elicit information about genetic conditions, and lowest for knowledge about genomics. There was greatest variation in their confidence to make decisions based on genomic information (IQR=2,7).

Current practice compared with expected future practice in genomic medicine

Overall, two-thirds of respondents think genomics will impact their practice in the next two years (n=199/298, 66.8%). Of those medical specialists who think their practice will be impacted, they anticipate it will change the way they manage patients (n=177/199, 88.9%) and practice medicine (n=151/199, 75.8%), more so than impact on workload (n=86/199, 43.2%). For respondents who felt genomics would not impact their practice in the next two years (n=50/298, 16.8%), open-text comments (n=47) suggested this was due to perceived relevance to their specialty, timing and/or pragmatic issues of service delivery (see **Supplementary Table S3** for examples). The remaining 49/298 (16.4%) respondents were 'unsure'.

More respondents currently perform clinical activities before and after E/GS testing (**Error! Reference source not found.**, n=314, 10.6% to 80.3% across these steps) than are involved in non-clinical activities directly related to the test itself (6.7% to 17.0%). Similar patterns were seen in their expectations of the steps they would perform in the future if they had adequate education, training and support: 40.8% expect to perform all pre-test steps and 23.1% all post-test steps, while 40.3% do not expect to perform any steps relating to the test itself. Notably, there were significant increases in the proportion of specialists who expect to perform each step in future practice ($p \leq 0.004$ across all steps), with the exception of eliciting phenotypic information about genetic

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conditions as part of a family or medical history for the purpose of assisting with variant interpretation, which was already high (80.3% current, 83.4% future; *p*=0.3).

Preferred future models for delivering genomic medicine

When reflecting on preferred models for delivering genomic medicine in the future, the model most often selected by respondents was referral to their local genetics services to initiate testing and discuss results (

Table 2). This was the case for both inpatient and outpatient settings. The second most preferred model was delivering testing with support from a local genetics service. The type of support included: advice on whether testing is appropriate (60.0% for inpatients; 66.7% for outpatients); interpreting results (72.0% for inpatients; 75.0% for outpatients); discussing results with families (60.0% for inpatients; 70.8% for outpatients); or follow-up genetic counselling (80.0% for inpatients; 83.3% for outpatients). A small number expect to initiate genomic testing themselves with no support from a local genetics service, while some respondents also indicated they did not expect to see patients who would benefit from genomic testing. Overall, significantly more respondents preferred a model that includes involvement of genetics services (for support or referral) than a model of initiating testing themselves: inpatients, 62.4% (95%CI 54.8–69.5) compared with 2.3% (95%CI 0.6–5.6), *p*<0.0001); outpatients, 69.7% (95%CI 62.8–76.1) compared with 4.1%; (95%CI 1.8–7.9, *p*<0.0001).

Preparedness for genomic medicine and preferences for future education

While a third (n=92/273, 33.7%) of respondents had completed education in genomics in the past year, only a quarter (n=73/297, 24.6%) felt prepared to use genomic sequencing testing in their practice. Comments from those who did not feel prepared or were 'unsure' (n=210 combined) primarily suggest this could be addressed through genomics education and training (**Supplementary Table S3**). Forty-two per cent of respondents felt that improved genomic knowledge may alter their clinical practice (n=115/273, 42.1%) but a similar proportion were 'unsure' (n=114/273, 41.8%).

When asked about preferred modes of learning genomics, most respondents (n=250/273; 91.6%) endorsed at least three different modes (

Table 3). The two most commonly preferred – CPD activities and learning from peers – were also the two most commonly-used currently. In contrast, reading specialty texts was the third most common way of learning about genomics currently, but the eighth preferred. Respondents indicated a preference for genomics education incorporated into their usual work activities (e.g., internal workplace seminars, departmental presentations and clinical meetings).

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Despite three-quarters of respondents reporting they had already learned basic concepts of genomics (

Table 4; n=271), a similar proportion still requested this topic for future education. Six topics were endorsed by over 80% of respondents including current and emerging applications in genomic medicine, the clinical utility of different tests and topics around patient management. Again, respondents could select more than one topic, with 92.3% indicating they wanted to learn about at least five topics in the future, and 26.4% selecting all topics. Nearly two-thirds of respondents indicated they wanted to learn about communication skills with patients, with comments throughout the survey suggesting a need for training in how to explain genomic testing concepts, implications and results to patients.

Discussion

This paper provides a baseline snapshot of Australian non-genetic medical specialists' practice of genomic medicine and perspectives at a point in time before E/GS was widely available to them as a funded clinical test. In 2019, 60% of all 409 survey respondents reported some form of interaction with genetics services or genetic/genomic testing. The test ordered most frequently was a microarray, but more than a quarter of all survey respondents indicated they had ordered a genomic sequencing test in the past twelve months. Respondents anticipated their practice would change in the near future, with significantly more respondents expecting to be involved in activities relating to E/GS in the next two years than currently. Consistent with discipline-specific studies from other countries,[13, 15, 32-34] we found the majority of respondents in our survey did not feel prepared to use genomic sequencing testing in their practice and over two-thirds preferred a model that involved genetics services in some way. Our study extends existing literature by providing greater depth of insight into the education needs and preferences of a broad range of medical specialists.

A strength of this snapshot is the use of a survey tool[24] grounded in a theoretical model. The COM-B model posits that behaviour is influenced by capability, opportunity and motivation.[22] *Opportunity* is clearly impacted by the availability of funded genomic tests. The test usage reported by respondents in this study reflects the availability of MBS reimbursement. For instance, microarrays for developmental delay have been established as MBS-reimbursed pathology tests for a decade. Tests reimbursed at the time of this survey are most typically requested by oncologists, clinical geneticists, haematologists, immunologists, paediatricians, obstetricians, nephrologists and neurologists.[35] Our survey sample included these specialties, barring clinical geneticists and oncologists, who were not the focus of this study. At the time of this survey E/GS tests were not

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reimbursed by the MBS. The relatively lower proportion of respondents who had ordered these tests used a variety of other funding mechanisms, most commonly hospital or research funds, and noted availability of funding as an influence when ordering genomic tests in the future. Since this study was completed, MBS now reimburses genomic sequencing tests for some clinical indications when ordered by paediatricians, enhancing their opportunity to use genomic testing in their clinical practice. It is anticipated that reimbursement for other clinical indications (and medical specialties) will follow in the future.

Broadening the responsibility for delivering genomic medicine to non-genetic medical specialties may address issues such as patient access, genetics workforce capacity or long wait times for genetics consultations. However, the medical specialists surveyed in our study show a clear preference for a model of genomic medicine that involves support from genetics services, rather than ordering tests and managing patients themselves. This may relate in part to their *capacity* to respond, such as constraints on their own time or competing health priorities. However, it is clear that there is a gap in respondents' perceived *capability* to respond to the availability of funded tests is limited. Currently, respondents lack confidence in their knowledge and ability to explain genomic concepts, and make decisions based on genomic information. This may explain their desire to practice collaboratively with clinical geneticists and genetic counsellors to varying extents. It is possible that these service model preferences could change as their capability (and confidence in their capability) develops with greater opportunity, experience and learning.[22, 24] Education and training was certainly seen as a solution to feeling unprepared by a substantial proportion of respondents in this study, as also observed by others.[36] In the past two years, continuing education for Australian medical specialists has been produced locally at an introductory level by a number of initiatives and organisations.[2] More is clearly needed: survey respondents are very interested in genomics education and nearly all respondents selected five or more of the topics that they wished to learn about. This is perhaps unsurprising given their perception of being unprepared and expectation of a greater role in the near future, provided they receive adequate support and education. The most popular education topics were related to pre-test aspects of testing, such as identifying appropriate patients to refer and knowing how to refer, consistent with the significantly stronger preference for a genetics-led model for genomic medicine. Educational strategies will need to consider both the diversity of respondents' preferences for modes of learning and timing with respect to clinical implementation. Not only will timing affect perceived relevance to clinical

[2] For example, https://elearning.racp.edu.au/course and http://learn-genomics.org.au/.

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practice, and therefore motivation to learn,[37] but preferences and needs may evolve as implementation progresses.

Our rigorously-developed survey tool can be deployed again in the future to capture changes in workforce practice and preferences over time. It could also be repurposed to inform needs for national education initiatives targeted to specific specialties or to assess change in their knowledge, practice or preferences. Wider use of the tool can also provide a basis for documenting and comparing data across specialties and countries. Our experience with deployment of the survey may assist in this regard, as we purposefully staggered recruitment methods to monitor response rates. Although it is not possible to determine which recruitment approach was most successful because of overlapping timeframes, increases in the number of responses to our survey coincided with recruitment approaches using social media, internal hospital communication channels and investigator networks. This may reflect increasing professional use of social media by medical specialists[38] and greater attention to emails from their employing hospital than a medical college or society. It may also explain the higher representation of Fellows and older specialists in our sample, as trainees were often not on staff mailing lists used by hospitals to distribute the survey. Our staggered and comprehensive recruitment approach also achieved a strong response from rural and remote medical specialists, who are often missed in research. Under- or over-representation of medical specialists in some Australian states may be due to differences in governance (hospital and/or research) and site-based communication policies that limited dissemination of the survey. One could assume specialists who graduated more recently may be more engaged with genomic medicine but previous research from our group described varied genomic literacy and experience at each career stage.[20] Similarly, specialists working in metropolitan areas, where almost all genetics services are based, might have been expected to be likely to complete our survey but this was not seen in our sample. While it is not possible to determine the response rate, our sample represents 1.2% of 37,000 medical specialist registrants with the Medical Board of Australia[27] and is within the range achieved in similar surveys of American physicians that also recruited participants through medical societies and associations (0.6–2.6%).[13, 39-41]

This national snapshot of medical specialists' current practice in genomic medicine provides the first detailed insight into the continuing genomics education needs of a broad group of subspecialties. It includes some specialties, such as emergency medicine, palliative medicine and infectious disease, for the first time internationally. Those currently involved and/or most interested in genomic medicine may have been more likely to respond, meaning these results may present an overestimation of current practice in Australia, but this might also mean our respondents are those likely to undertake continuing education and engaging with genomics. Consequently, our results can

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assist providers to best meet learner needs when developing and implementing genomics education to ultimately create a competent, genomics-literate workforce. The findings will also be helpful to genetics and other clinical services implementing models for genomic medicine delivery. Further data analysis will provide insights into any differences between early adopters of genomic medicine and those who have not yet engaged, enabling the development of targeted, tailored genomics education and other capability-building strategies for optimising the adoption of genomics by medical specialists.

Author contributions

AN and EK were involved in all stages of this work and manuscript preparation. BM, SM and CG were involved with all stages except data acquisition. MJ analysed the qualitative data responses and was involved in manuscript preparation. All authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Competing interests

All authors declare no completing interests.

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Data sharing statement

Data are available upon reasonable request.

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Figure captions

Figure 1: *Number of survey attempts shown with recruitment strategies and timelines after pilot data were complete (n=41). Recruitment start dates are shown and overlapped from March through October 2019 (as described in the Methods). Snowball recruitment may have continued beyond these periods (e.g., forwarding a newsletter or retweeting) but this could not be monitored.*

Figure 2: *Proportion of each reported primary specialty in the sample (n=409) grouped by primary medical college affiliation. Grey bars signify specialties where proportions were representative of the medical specialist population when compared with reference data.[27] The black bar signifies a specialty which was over-represented (physicians; p<0.0001). White bars signify specialties which were under-represented: anaesthesiology (p=0.002), psychiatry (p<0.0001) and surgery (p<0.0001). The reference data did not include a classification for 'rural and remote medicine' so representativeness could not be determined for this specialty (pale grey bar).*

Figure 3: *Average confidence about genomic concepts and skills on a scale of 1 'Not at all confident', 5 'Neutral' to 10 'Very confident' (n=273). Boxes represent the interquartile ranges with minimum and maximum value; medians are shown as white bars.*

Figure 4: *Steps in genomic testing that respondents (n=314) currently perform (blue bars) compared to steps they expect to perform in the future, if they had adequate support, education and training (black bars). Non-clinical steps are indicated by [a]. Differences between proportions for 'Currently perform' and 'Expect to perform' are indicated by *p=0.004, **p=0.001, ***p=0.0006, ****p<0.0001. The difference for the first step – Elicit genetic information through family history – was not significant (p=0.3). The full wording of each step is provided in* ***Supplementary Table S5****.*

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Tables

Table 1: *Description of the sample and representativeness (n=409).*

Characteristic	Respondents		Reference data		
	n (%)	95%CI	N (%)	95%CI	*p*
Gender[1]					
Male	213 (52.1)	47.2–56.9	61,700 (57.1)	56.8–57.4	0.039
Female	185 (45.2)	40.4–50.1	46,281 (42.9)	42.6–43.2	0.330
Prefer not to answer	11 (2.7)	1.5–4.8	–	–	–
Age[1]					
≤24 years	–	–	398 (0.4)	–	
25–34 years	29 (7.1)	4.6–9.6	26,827 (24.8)	24.6–25.1	<0.0001
35–44 years	114 (27.9)	23.5–32.2	28,431 (26.3)	26.1–26.6	0.4794
45–54 years	123 (30.1)	25.6–34.7	22,415 (20.8)	20.5–21.0	<0.0001
55–64 years	103 (25.2)	21.2–29.6	18,060 (16.7)	16.5–17.0	<0.0001
≥65 years	40 (9.8)	7.2–13.1	11,852 (11.0)	10.8–11.2	0.4398
Trainee level[2]					
Basic Trainee	9 (2.2)	1.3–4.6	5,858 (12.1)	11.8–12.4	<0.0001
Advanced Trainee	18 (4.4)	2.6–6.7	8,890 (18.3)	18.0–18.7	<0.0001
Fellow	382 (93.4)	89.9–95.0	33,749 (69.6)	69.2–70.0	<0.0001
Australian state or territory[1,3]					
Australian Capital Territory	28 (6.9)	4.4–9.3	702 (1.9)	1.8–2.0	<0.0001
New South Wales	119 (29.1)	24.7–33.5	11,566 (31.2)	30.7–31.7	0.3622
Northern Territory	8 (2.0)	0.6–3.3	373 (1.0)	0.9–1.1	0.0568
Queensland	75 (18.3)	14.8–22.4	7,320 (19.7)	19.3–20.1	0.4777
South Australia	20 (4.9)	2.8–7.0	2,896 (7.8)	7.5–8.1	0.0283
Tasmania	13 (3.2)	1.5–4.9	759 (2.0)	1.9–2.2	0.1091
Victoria	119 (29.1)	24.7–33.5	9,952 (26.8)	26.4–27.3	0.3063
Western Australia	26 (6.4)	4.0–8.7	3,510 (9.5)	9.2–9.8	0.0324
Primary work location[3,4]					

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Characteristic	Respondents		Reference data		
	n (%)	95%CI	N (%)	95%CI	*p*
Major city	306 (75.0)	70.6–79.0	72,304 (79.2)	78.9–79.4	0.0391
Inner regional	59 (14.5)	11.4–18.2	12,422 (13.6)	13.4–13.8	0.6127
Outer regional	31 (7.6)	5.4–10.6	5,299 (5.8)	5.7–6.0	0.1216
Remote	10 (2.5)	1.3–4.5	865 (1.0)	0.9–1.0	0.0018
Very remote	2 (0.5)	0.1–2.0	376 (0.4)	0.4–0.5	0.8048
***Primary employer*[5]**					
Public hospital or healthcare provider	288 (70.4)	65.8–74.7			
Private hospital or healthcare provider	17 (4.2)	2.6–6.6			
Self-employed/ private practice	83 (20.3)	16.7–24.5			
Other (government, research institute, etc.)	21 (5.1)	3.4–7.8			

Reference data were: [1] Registration Data Table 2019 [27]; [2] Medical Education and Training in Australia 1st Edition report 2017 [31]; [3]n=408 for state and location; [4] Medical Workforce 2016 Factsheet [28]; [5] There were no comparable reference data for this category.

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Table 2: *Medical specialists' preferred models for delivering a genomic sequencing test in inpatient and outpatient settings (n=218).*

	INPATIENT n=178[1]		OUTPATIENT n=195[1]	
	n (%)	**95%CI**	**n (%)**	**95%CI**
You **initiate** testing and discuss results with patients/families	4 (2.3)	0.6–5.6	8 (4.1)	1.8–7.9
You initiate testing and discuss results with patients/families, with **support** from a clinical genetics team as needed	43 (24.2)	18.15–31.1	49 (25.1)	19.2–31.8
You **refer** to a clinical genetics team to initiate testing and discuss results with patients/families	68 (38.2)	31.0–45.8	87 (44.6)	37.5–51.9
You **do not see**, and do not expect to see, patients who would benefit from genomic testing	33 (18.5)	13.1–25.0	23 (11.8)	7.6–17.2
Unsure at this stage	30 (16.9)	11.7–23.2	28 (14.4)	9.8–20.1

[1] *A total of 218 respondents completed this question, indicating a preference for either the inpatient or outpatient setting, or both.*

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Table 3: *Current and preferred modes of learning about genomics (n=273).*[1]

Mode of learning about genomics	Currently use (%)	Prefer to use (%)
Continuing Professional Development/Continuing Medical Education activities	51.8	79.8
Consult colleagues and peer	54.0	79.4
Internal workplace specialty seminars, conferences or similar	34.1	74.0
Departmental presentations	35.8	72.0
Clinical meetings	34.8	71.4
External specialty seminars, conferences, etc.	36.0	67.3
Internal workplace genetic or genomic seminars, conferences, etc.	24.9	66.3
Reading specialty texts	48.2	63.2
Online webinars, courses, MOOCs, etc.	15.8	59.6
Certification/fellowship activities	34.4	56.4
External genetic or genomic seminars, conferences, etc.	18.4	50.0
Small group tutorials	8.1	44.9
Study days at place of employment	12.5	41.9
Genomic research project	17.6	32.6
Time in a service or laboratory with genomics expertise	6.2	17.6
Mass media	12.5	14.0
Social media	7.4	11.0
Other (e.g., fact sheet written by geneticist)	0.0	0.4

[1] *Respondents could select more than one mode.*

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Table 4: *Topics relevant to genomics medicine that medical specialists have learnt about or would like to learn (more) about (n=271).*[1]

Education topic	Have learnt about (%)	Want to learn (more) about (%)
Genetic/genomic knowledge		
Basic concepts	77.5	77.1
Disorders and diseases	74.2	83.4
Current applications in genomic medicine	60.9	88.9
Emerging applications in genomic medicine	55.7	87.8
Genetic/genomic testing and technology		
Types of genetic tests	64.9	76.4
Types of genomic tests	58.7	77.1
Applications of somatic genomic tests	45.4	75.6
Applications of germline genomic tests	37.6	69.7
Clinical utility of tests	57.6	88.6
Classification of genomic data during testing	41.3	67.9
Limitations of testing	50.2	79.7
Pre- or post-test aspects		
Recognising patients who may benefit from genomic testing	60.9	83.0
Communication skills with patients	70.8	63.1
Performing genetic risk assessments	57.6	67.5
Referring appropriately for a genomic test	59.4	81.5
Requesting a genomic test for a patient	53.9	70.8
Interpreting genomic test results	52.0	74.9
Cascade testing	53.9	68.6
Ethical, legal and social implications		
Ethical implications	59.0	75.6
Legal implications	52.4	75.3
Psychosocial implications	57.2	74.9

[1] *Respondents could select more than one topic.*

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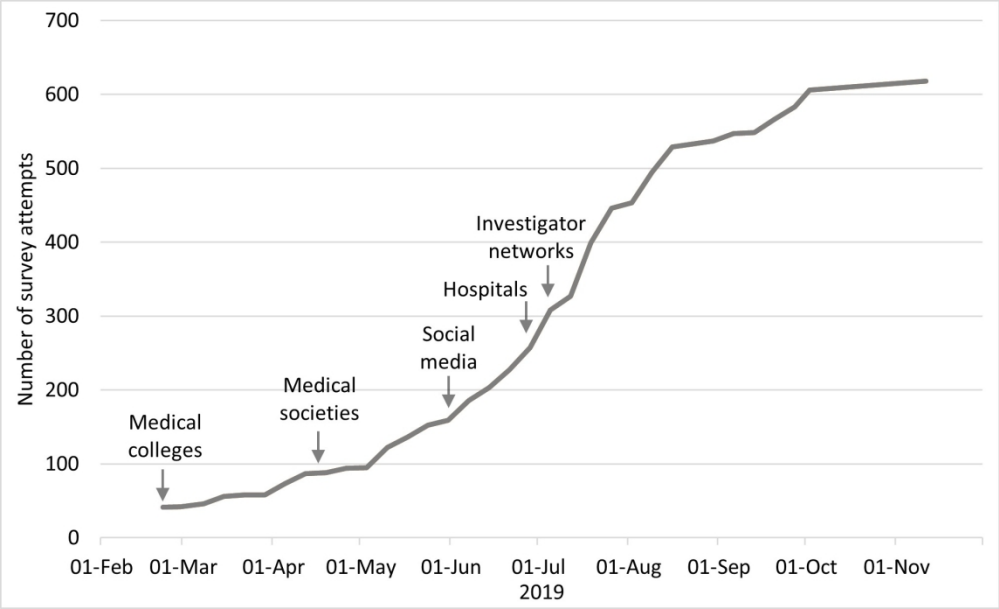

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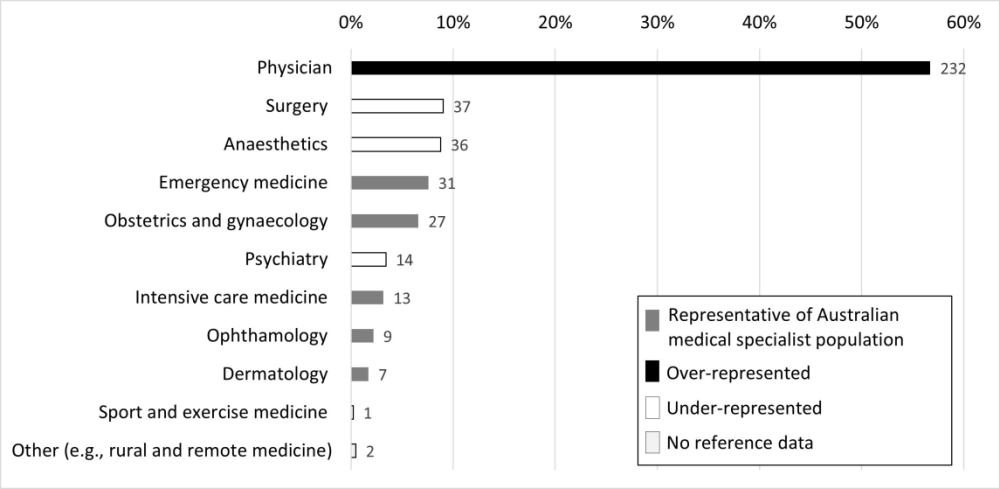

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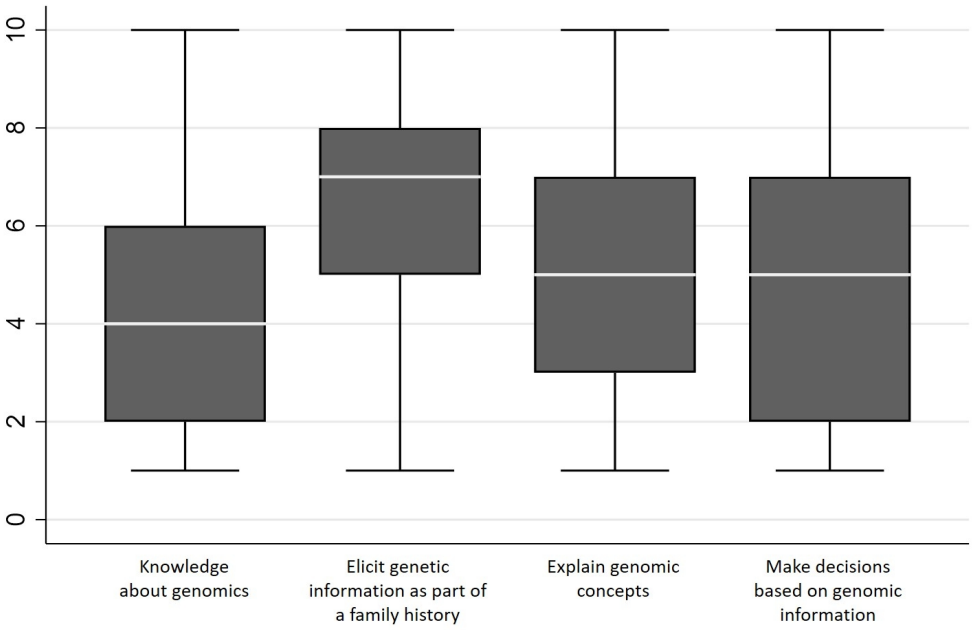

Figure 3: Average confidence about genomic concepts and skills on a scale of 1 'Not at all confident', 5 'Neutral' to 10 'Very confident' (n=273). Boxes represent the interquartile ranges with minimum and maximum value; medians are shown as white bars.

61x40mm (600 x 600 DPI)

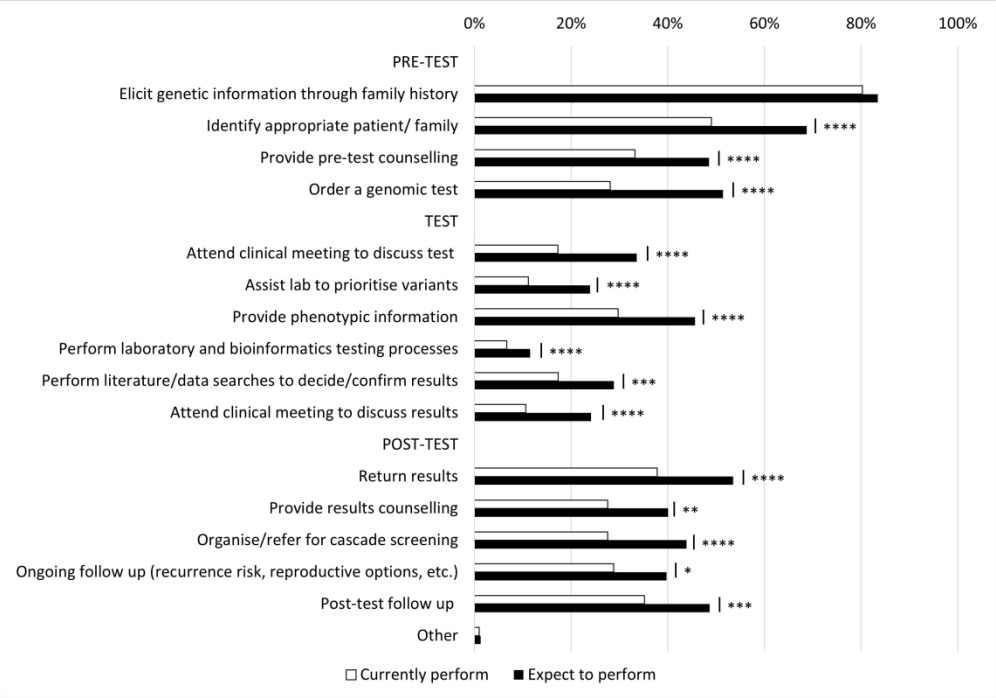

Figure 4: Steps in genomic testing that respondents (n=314) currently perform (blue bars) compared to steps they expect to perform in the future, if they had adequate support, education and training (black bars). Non-clinical steps are indicated by a. Differences between proportions for 'Currently perform' and 'Expect to perform' are indicated by *p=0.004, **p=0.001, ***p=0.0006, ****p<0.0001. The difference for the first step – Elicit genetic information through family history – was not significant (p=0.3). The full wording of each step is provided in Supplementary Table S5.

133x93mm (600 x 600 DPI)

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SUPPLEMENTARY MATERIALS

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Figure S1. Summary of survey attempts, responses and final sample for analysis

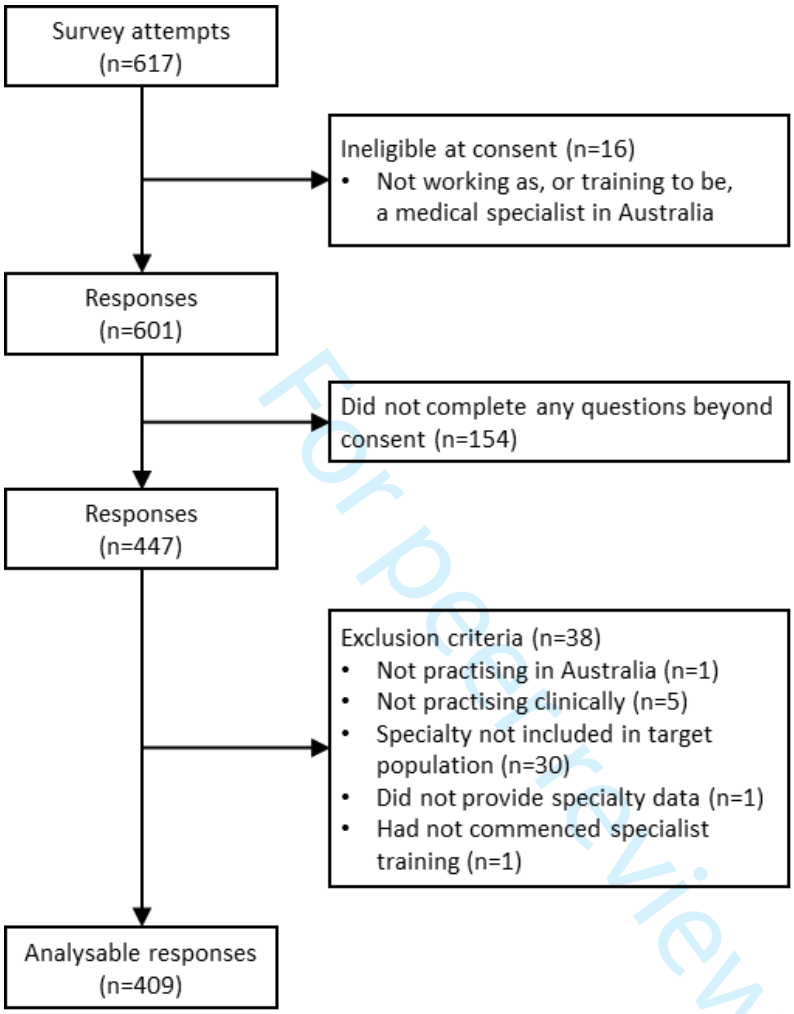

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Table S1. *Conditions for which genetic/genomic testing was covered by Medicare Benefit Scheme at the time of survey deployment in 2017.*[1]

Condition
1. Cytogenetics in general (pregnancies) and products of conception
2. Developmental delay
3. Peripheral neuropathy
4. Alport's Syndrome
5. Ataxia
6. Factor V Leiden Deficiency
7. Haemochromatosis
8. Polycythaemia/thrombocytopaenia
9. Drug toxicity (thiopurine)
10. Cystic fibrosis
11. Haematological malignancies
12. *BRCA* testing for breast/ovarian cancer
13. Leukemias
14. Mast cell disease/hypereosinophilia/eosinophil leukemia
15. *In situ* hybridisation tests for cancers
16. Von Hippel Lindau Syndrome (predisposition to various cancers)
17. Metastatic melanoma
18. Metastatic colorectal cancer
19. Metastatic adenocarcinoma stomach
20. Non-small cell lung cancer

[1]. *Australian Government Department of Health. Medicare Benefits Schedule Book. ISBN: 978-1-76007-375-3. Publications Number: 12289. Australian Government; 2019 [accessed 6 January 2021]. Available from: http://www.mbsonline.gov.au/.*

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Table S2. *Examples of recoded open-text responses where a respondent selected 'Other (please specify……)' for a categorical question.*

Question	Open text response [ID, specialty]	Recoded category
[If contacted clinical genetics team or service in last 12 months]: Why did you contact your clinical genetics team or service? [a] Information about a suspected genetic condition [b] Advice on what type of genetic or genomic test to order [c] Advice on how to refer the patient to my clinical genetics team or service [d] Assistance with genetic counselling before the test [e] Assistance with genetic counselling after the test [f] Other (please specify)……	*"Referral"* [135, surgery]	[c]
	"Facilitating genomic testing so that genetic counselling can be given to patient before test" [145, paediatrics]	[d]
[If did not contact clinical genetics team or service in last 12 months]: Why haven't you contacted your clinical genetics team or service? [a] Genetics and genomics are not relevant to my practice [b] I have not yet needed advice from a clinical genetics team or service in my practice [c] I can manage my patients without advice from a clinical genetics service [d] I'm not sure how to contact my clinical genetics team or service [e] I do not have access to a clinical genetics team or service [f] Other (please specify)……	*"My cohort of patients generally do not need genetic service input"* [129, gerontology]	[a]
	"We do some of this inhouse" [282, general medicine]	[c]
Below is a list of some of the steps involved in genomic sequencing testing from pre-test to post-test *[see **Table S5**]*. Please indicate which steps you currently perform and which ones you expect to perform in the future if you had adequate education, training and support. If you selected "Other" step, please specify.	*"Going over letters and reports from genetics, explaining things again in context"* [221, paediatrics]	[k]
	"I continue to see patients after their diagnostic test, which hopefully occurs as part of the evaluation of their condition" [3, gerontology]	[n]

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Question	Open text response [ID, specialty]	Recoded category
What is/would be your preferred model for delivering a genomic sequencing test in an outpatient setting in your clinical practice, assuming you have appropriate education, training and funding? [a] You initiate testing and discuss results with patients/families [b] You initiate testing and discuss results with patients/families, with support from a clinical genetics team as needed [c] You refer to a clinical genetics team to initiate testing and discuss results with patients/families [d] You do not see, and do not expect to see, patients who would benefit from genomic testing [e] Unsure at this stage [f] Other (please specify)……	*"Not relevant to my specialty"* [140, palliative medicine] *"Same as for inpatient"* [109, palliative medicine; selected [b] for Inpatient response]	[d] [b]
[If selected 'yes' to genomics will impact practice within two years]: What areas will be impacted? [a] The way I practice medicine [b] My workload [c] Patient management [d] Other (please specify)……	*Clinical outcome and prognostications* [123, intensive care]	[c]
[If selected 'yes' to attending genomic professional development education or training in past year]: Was this: [a] In-house (internal) program/s [b] External program/s [c] Online training (webinar, MOOC, etc.) [d] Other (please specify)……	*"Recent commencement of multidisciplinary meeting"* [416, cardiology] *"International Clinical Cardiovascular Genetics conference"* [430, paediatrics]	[a] [b]

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Table S3. *Illustrative quotes from open-text survey comments.*

Domain	Quote
Current practice compared with future practice in genomic medicine *Q: Do you think genomics will impact your practice in the next 2 years?*	
Expect genomics *will* impact practice in next two years	*"Becoming increasingly available and of measurable significance"* [513, surgery] *"I expect it [genomics] will increasingly impact on the practice of medicine in terms of diagnoses, prognoses and treatment"* [281, paediatrics] *"Increased patient requests"* [271, obstetrics and gynaecology]
Expect genomics will *not* impact practice in next two years	*"Emergency department have more important competing interests in treatment delivery to patients"* [383, emergency medicine] *"Timeframe remains too short to see this implemented in a regional area"* [535, anaesthesiology]
Preferred future models for delivering genomic medicine *Q: What is/would be your preferred model for delivering a genomic sequencing test* in your clinical practice, assuming you have appropriate education, training and funding?*[1]	
Referring to genetics services to initiate testing and discuss results	*"For my patients and practice, having an accessible [genetics] clinic for this would be best. I would be very keen to be involved as far as possible, but do not have time to keep up with this rapidly developing field. I would like to be invited to my patients' MDT [multidisciplinary team] discussions. That way I am involved, and have the knowledge to answer follow-up and clarification questions. It would also be a way to increase my knowledge"* [100, nephrologist]
Delivering testing with support from genetics services	*"[Genetics support for both inpatients and outpatients] would streamline the process, improve access and possibly reduce Clinical Genetics load by filtering patients and families I can manage while they still see the patients or results beyond my expertise"* [220, paediatrics, community child health] *"We (clinicians) may be more familiar with the disease phenotype than the Genetics team"* [33, immunopathology] *"Clinicians should be able to initiate testing but will need support with interpretation and counselling, particularly initially until genomic medicine is core practice"* [350, palliative medicine]
Initiating genomic testing themselves with no support from genetics	*"I expect to be able to manage simpler conditions/results, with access to more specialist input when needed"* [129, gerontology]
Will not see patients who would benefit from genomic sequencing tests	*"Relevance to decision making in real time"* [459, emergency medicine] *"Not sure of any relevance to my practice"* [541, anaesthesiology]

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Domain	Quote
Preparedness for genomic medicine and preferences for future education	
Q: Do you feel prepared to use genomic sequencing testing in your practice?*	
	"I have little to no training in genetics and genomic medicine. We had a total of 4 genetics lectures at medical school, and there is limited assessment of genetics/genomics in the [college fellowship examination]. Genomic testing is not routinely used in our practice" [73, intensive care]
	"My knowledge of this whole area is woefully inadequate. I can cope with karyotype analysis and testing for CF [cystic fibrosis]. I can also discuss prenatal diagnosis options, PGT-A [pre-implantation genetic testing] and expanded carrier testing but that's about it…… It clearly will be an important part of medical practice in the future" [213, obstetrics and gynaecology]
	"I'm happy to do [genomic testing] but need training." [342, surgery]
	"Need further information, education on who would best benefit from this test, how to consent for it and then how to interpret results" [414, general paediatrics]
Preferences for learning about genomics	
Q: What would help improve your confidence?[2]	*"Further training in counselling [would improve my confidence]–in ability to explain concepts and then clinical implications and follow-on from this"* [27, paediatric neurology]
Q: Please explain why you do not expect to perform the selected steps [involved in genomic sequencing testing]*[3]	*"Would welcome some education on use of these tests in orthopaedics"* [391, surgery]

*[1] Full question provided in **Table S2**; [2] following the question on confidence in four genomic knowledge and skills areas, presented in **Figure 1**; [3] following the question on steps involved in genomic sequencing testing, presented in **Figure 4** and **Table S2.***

** Definitions were provided for these terms*

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Table S4. *Participant-reported funding for genomic tests ordered in the past year.*[1]

	Microarray n=112	**Gene panel** n=112	**Exome/genome sequencing** n=50
Medicare Benefit Scheme	48.2%	17.0%	2.0%[2]
Institute/hospital	41.1%	52.6%	44.0%
State government	13.4%	17.0%	12.0%
Research grant	2.7%	11.6%	60.0%
Patient	12.5%	24.1%	4.0%
Unsure	11.6%	8.0%	6.0%

[1] *Respondents could select more than one funding source per test type.*

[2] *At the time of the survey the MBS scheme did not fund E/GS, so this response (n=1) is incorrect.*

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Table S5. *The full wording of each step involved in genomic testing as presented in the survey.*[1]

Pre-test
[a] Eliciting information about genetic conditions as part of a family or medical history
[b] Identifying a patient suitable for a genomic test
[c] Pre-test counselling to assist in making an informed decision, e.g., genetics, test limitations, variants of uncertain/unknown significance*, incidental/secondary findings, unexpected non-paternity or consanguinity
[d] Ordering a genomic test for a patient
Test
[e] Attending multidisciplinary team meeting to discuss the genomic test (e.g., intake meeting)
[f] Assisting the lab to narrow down the genes of interest (creating a gene list to prioritise variant analysis)[2]
[g] Providing phenotypic information to the lab to prioritise variant analysis
[h] Laboratory and bioinformatics testing processes[2]
[i] Searching the literature and databases for evidence of variant pathogenicity*,[2]
[j] Attending a multidisciplinary team meeting to discuss variant prioritisation*, interpretation and classification*
Post-test
[k] Provide test results to patients/ families
[l] Provide genetic counselling to patients/families, e.g., explain variants of uncertain/unknown significance*, incidental/secondary findings, unexpected non-paternity or consanguinity
[m] Organising/ referring for further testing of family members if required, e.g., cascade testing or segregation studies
[n] Ongoing management of the patient, e.g., clarify recurrence risk and discuss reproductive planning options
[o] Post-test follow up of patient to check understanding of result/ ask any additional questions
[p] Other (please specify)……

[1] The survey is available as supplementary material in [24]; [2] These steps are considered non-clinical, i.e., laboratory;

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Reporting checklist for cross sectional study.

Based on the **STROBE** cross sectional guidelines.

Instructions to authors

Complete this checklist by entering the page numbers from your manuscript where readers will find each of the items listed below.

Your article may not currently address all the items on the checklist. Please modify your text to include the missing information. If you are certain that an item does not apply, please write "n/a" and provide a short explanation.

Upload your completed checklist as an extra file when you submit to a journal.

In your methods section, say that you used the STROBE cross sectionalreporting guidelines, and cite them as: von Elm E, Altman DG, Egger M, Pocock SJ, Gotzsche PC, Vandenbroucke JP. The Strengthening the Reporting of Observational Studies in Epidemiology (STROBE) Statement: guidelines for reporting observational studies.

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		Reporting Item	*Page*
Title and abstract			
Title	#1a	Indicate the study's design with a commonly used term in the title or the abstract	1
Abstract	#1b	Provide in the abstract an informative and balanced summary of what was done and what was found	2
Introduction			
Background / rationale	#2	Explain the scientific background and rationale for the investigation being reported	4
Objectives	#3	State specific objectives, including any prespecified hypotheses	4
Methods			
Study design	#4	Present key elements of study design early in the paper	5-6
Setting	#5	Describe the setting, locations, and relevant dates, including periods of recruitment, exposure, follow-up, and data collection	5-6
Eligibility criteria	#6a	Give the eligibility criteria, and the sources and methods of selection of participants.	5-6
	#7	Clearly define all outcomes, exposures, predictors, potential confounders, and effect modifiers. Give diagnostic criteria, if applicable	5
Data sources / measurement	#8	For each variable of interest give sources of data and details of methods of assessment (measurement). Describe comparability of assessment methods	5-6

		Reporting Item	*Page*
		if there is more than one group. Give information separately for exposed and unexposed groups if applicable.	
Bias	#9	Describe any efforts to address potential sources of bias	5-6, 13
Study size	#10	Explain how the study size was arrived at	5-6, 13
Quantitative variables	#11	Explain how quantitative variables were handled in the analyses. If applicable, describe which groupings were chosen, and why	7-11
Statistical methods	#12a	Describe all statistical methods, including those used to control for confounding	7
Statistical methods	#12b	Describe any methods used to examine subgroups and interactions	7
Statistical methods	#12c	Explain how missing data were addressed	7
Statistical methods	#12d	If applicable, describe analytical methods taking account of sampling strategy *Not required as sampling strategy was same across single cohort*	N/A
Statistical methods	#12e	Describe any sensitivity analyses *Not required*	N/A
Results			
Participants	#13a	Report numbers of individuals at each stage of study—eg numbers potentially eligible, examined for eligibility, confirmed eligible, included in the study, completing follow-up, and analysed. Give information separately for exposed and unexposed groups if applicable.	7-11, 17-23, vii
Participants	#13b	Give reasons for non-participation at each stage	7-8
Participants	#13c	Consider use of a flow diagram	i
Descriptive data	#14a	Give characteristics of study participants (eg demographic, clinical, social) and information on exposures and potential confounders. Give information separately for exposed and unexposed groups if applicable.	7-8, 18-20
Descriptive data	#14b	Indicate number of participants with missing data for each variable of interest	7-11, 18-23, vii
Outcome data	#15	Report numbers of outcome events or summary measures. Give information separately for exposed and unexposed groups if applicable.	7-11, 18-23, i-vii
Main results	#16a	Give unadjusted estimates and, if applicable, confounder-adjusted estimates and their precision (eg, 95% confidence interval). Make clear which confounders were adjusted for and why they were included *Not relevant*	N/A

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		Reporting Item	*Page*
Main results	#16b	Report category boundaries when continuous variables were categorized	8, 18
Main results	#16c	If relevant, consider translating estimates of relative risk into absolute risk for a meaningful time period *Not relevant*	N/A
Other analyses	#17	Report other analyses done—e.g., analyses of subgroups and interactions, and sensitivity analyses	7-11, 18-23, i-vi
Discussion			
Key results	#18	Summarise key results with reference to study objectives	11-14
Limitations	#19	Discuss limitations of the study, taking into account sources of potential bias or imprecision. Discuss both direction and magnitude of any potential bias.	13
Interpretation	#20	Give a cautious overall interpretation considering objectives, limitations, multiplicity of analyses, results from similar studies, and other relevant evidence.	11-14
Generalisability	#21	Discuss the generalisability (external validity) of the study results	13
Other Information			
Funding	#22	Give the source of funding and the role of the funders for the present study and, if applicable, for the original study on which the present article is based	14

Notes:

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