PEER REVIEW HISTORY

ARTICLE DETAILS

TITLE (PROVISIONAL)	Measuring physician practice, preparedness and preferences for genomic medicine: a national survey
AUTHORS	Nisselle, Amy; King, Emily; McClaren, Belinda; Janinski, Monika; Metcalfe, Sylvia; Gaff, Clara

VERSION 1 – REVIEW

REVIEWER	O'Daniel, Julianne University of North Carolina at Chapel Hill, Genetics
REVIEW RETURNED	09-Oct-2020

GENERAL COMMENTS	The authors present data from a broad survey of Australian medical specialists regarding current and anticipated genomics practice as well as overview of desire for additional genomics education including topics and preferred formats. This study is timely given the continual growth of clinical genetics spurred by advances in technology and clinical knowledge, as well as expanded access to clinical funding to cover test cost. While the findings are interesting and clear, I believe some additional background as well as data analysis could strengthen the manuscript and provide more insight to the readers. While the broad representation of medical specialists is a strength, it is also noted that some specialists may have better access to tests based on Australian reimbursement policies. Similarly, some specialists may be more likely to have patients that require genetic evaluation or follow a broader pool of patients and therefore more likely to interact with patients who have a genetic diagnosis. Although you have very small numbers for many specialist groups or even practice settings, it would be helpful to stratify your results to see if there are trends. For example, were those that ordered no, 1-2, or more frequent tests more/less likely to indicate they felt unprepared? Or indicate they had received genetics education in the past 12 months? Were those in remote regions more likely to have ordered the tests simply because referral to a genetics specialist was not as available for their patients? Along similar lines, which specialists were more likely to indicate that genomics would not impact their practice. These trends, if available could potentially be used to prioritize educational development and implementation. Additional detail/background could also be helpful in some places. For instance, although the development of your survey instrument has been reported elsewhere, it would be quite helpful for readers to have a brief description of the item number and topical domains. Further, for readers not familiar with Australian medical education, the difference between a specialist and a general practitioner may

	not be clear. Pediatricians and general medicine practitioners may be considered or called 'generalists' elsewhere. In addition, the exclusion of oncologists should be restated in the discussion as that group is likely to be exposed to significant genetics practice. Lastly, while briefly addressed in the discussion, it may be worth expanding on which specialty groups are most likely to be ordering the approved clinical tests in Australia (e.g. microarray, gene panels) as this may help frame consideration of your response data. As you mention, 1% response rate is similar to some broad studies of American physicians. That said, a bit more discussion is warranted of the potential biases in your results and how that should be considered in applying the findings to educational development and implementation. Do you believe additional surveys are needed and if so, targeted at which groups? Thank you for the opportunity to learn about your valuable work.

REVIEWER	Cornel, Martina VU University Medical Center, Clinical Genetics
REVIEW RETURNED	07-Dec-2020

GENERAL COMMENTS	Nisselle et al. have measured physician practice, preparedness and preferences for genomic medicine in Australia, a country with a major Genomics programme (https://www.australiangenomics.org.au/). The challenge that many health care providers will have to cope with genomics results is recognized in many countries, but indeed this is the first initiative to investigate many specialties' educational needs simultaneously. The paper is well written and timely. The changes in tests becoming available (e.g. now being funded in regular health care) makes the preparedness of physicians more urgent. The topics mentioned (Table 4) are well in line with learning needs reported in other studies (e.g. for primary care providers and oncologists). This can indeed guide the development of training modules, as can the diversity of modes of learning requested (Table 3). The participants were collected through many survey attempts. It is already a result that so many attempts were needed. Does this mean that the relevance of genomics is not immediately clear to non-genetic experts? A limitation of a survey that was advertised nationally via a diversity of channels, is that those interested most may have participated, and the overall results may be too optimistic. One would expect physicians that graduated recently to be most interested in genomics medicine, as well as those in university clinics and in proximity to genetic centres, but interestingly older specialists and those in rural and remote areas were overrepresented. I suggest to briefly comment on the long quest for the 409 participants. P 8 l 17 speaks about 70.7% of the respondents being physicians. Apparently experts in anesthesiology, OB/GYN, psychiatry and surgery do not count as physicians, which needs some more explanation. In The Netherlands, these medical specialists would all count as physicians.

REVIEWER	Dunnenberger , Henry M

	NorthShore University HealthSystem
REVIEW RETURNED	30-Dec-2020

GENERAL COMMENTS	This is a well written manuscript. The design and recruitment for the study is great. I only have minor comments. 1) Consider using a different color for the blue in the figures. I found it tough to read. 2) Consider adding a consort plot for your exclusion data. 3) Consider adding description about what is meant by genetics service. What this entails is unclear as a reader outside of Australia. 4) Based on data from the US, the perceived education needs for a provider changes significantly if the provider has some experience with genetic testing. It would be interesting to see if that trend exist in this data set. 5) Adding information about what conditions genetic testing was covered by Medicare Benefit Scheme since this payment had a large effect on the data.

VERSION 1 – AUTHOR RESPONSE

Reviewer 1

While the broad representation of medical specialists is a strength, it is also noted that some specialists may have better access to tests based on Australian reimbursement policies. Similarly, some specialists may be more likely to have patients that require genetic evaluation or follow a broader pool of patients and therefore more likely to interact with patients who have a genetic diagnosis. Although you have very small numbers for many specialist groups or even practice settings, it would be helpful to stratify your results to see if there are trends. For example, were those that ordered no, 1-2, or more **frequent** tests more/less likely to indicate they felt **unprepared**? Or indicate they had received **genetics education** in the past 12 months? Were those in **remote** regions more likely to have ordered the tests simply because referral to a genetics specialist was not as available for their patients? Along similar lines, which **specialists** were more likely to indicate that genomics would not **impact** their practice. These trends, if available could potentially be used to prioritize educational development and implementation.

Reviewer 3

4) Based on data from the US, the **perceived education needs** for a provider changes significantly if the provider has some **experience with genetic testing**. It would be interesting to see if that trend exist in this data set.

We agree with the reviewers that analysing the data for associations and trends will provide important insights. As noted in the Discussion (p.13), we are currently preparing a second manuscript to permit in-depth analyses and reporting of these data, including the associations requested by the Reviewers.

Regarding the specific request from Reviewer 1 to stratify results by frequency of test ordering, we agree this would be interesting but unfortunately the sample size does not permit robust statistical analyses (only 12 respondents indicated they order E/GS weekly or monthly and only 9 answered the other survey questions required for association analyses). Stratifying the data, those ordering most frequently reflected the overall demographic composition of the cohort (data not shown).

ADDITIONAL DETAIL/DISCUSSION

Editor

Please ensure that you have fully discussed the methodological limitations of the study in the discussion section of the main text.

We have included additional discussion points around methodology and limitations, as described below in our responses to both Reviewers' comments.

Reviewer 1

Additional detail/background could also be helpful in some places. For instance, although the development of your survey instrument has been reported elsewhere, it would be quite helpful for readers to have a brief description of the item number and topical domains.

We have added further details to the end of the Introduction and Methods.

Introduction (p.4):

> 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]

Methods (p.5):

> 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. ~~For development of the survey, w~~** 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.

Further, for readers not familiar with Australian medical education, the difference between a specialist and a general practitioner may not be clear. Pediatricians and general medicine practitioners may be considered or called 'generalists' elsewhere.

In addition, the exclusion of oncologists should be restated in the discussion as that group is likely to be exposed to significant genetics practice.

We have amended the Methods (p.5):

> 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 sub-specialties.[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, who are the focus of other ongoing national studies**.

Lastly, while briefly addressed in the discussion, it may be worth expanding on which specialty groups are most likely to be ordering the approved clinical tests in Australia (e.g. microarray, gene panels) as this may help frame consideration of your response data.
We have added more detail to the Discussion (p.11) and added a new reference:

> 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]**

Reviewer 2

The participants were collected through many survey attempts. It is already a result that so many attempts were needed. Does this mean that the relevance of genomics is not immediately clear to non-genetic experts?

I suggest to briefly comment on the long quest for the 409 participants.
Thank you for these comments. We also believe the recruitment process and outcomes provide important insights for other researchers and welcome the opportunity to elaborate on the points suggested. We expected that busy clinicians would not immediately prioritise time to participate in research. Therefore, we purposefully designed the recruitment process to allow time to stagger the four approaches in an attempt to gain insights into more, or less, successful recruitment methods. As noted on p.12–13, we could not draw definitive conclusions as to the most 'effective' strategy, as some overlapped (e.g., a College member may have opened or forwarded a newsletter months after receiving it) but we did see increases in response rate when using social media and hospital mailing lists. These types of more pragmatic insights are not often reported in the literature.

We have highlighted this intentional recruitment approach in the Discussion (p.12):

> Our experience with deployment of the survey may assist in this regard**, as we purposefully staggered recruitment methods to monitor response rates**.

As indicated in the following response, it is certainly possible that those who did not perceive genomics as relevant were less likely to spend time completing the survey.

A limitation of a survey that was advertised nationally via a diversity of channels, is that those interested most may have participated, and the overall results may be too optimistic.
As you mention, 1% response rate is similar to some broad studies of American physicians. That said, a bit more discussion is warranted of the potential biases in your results and how that should be considered in applying the findings to educational development and implementation.

We have added more detail to our existing discussion of the potential implications of responder bias on p.13 of the Discussion:

> 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.

Do you believe additional surveys are needed and if so, targeted at which groups?
We have expanded the description of potential applications of our survey in the Discussion (p.12):

> 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.

One would expect physicians that graduated recently to be most interested in genomics medicine, as well as those in university clinics and in proximity to genetic centres, but interestingly older specialists and those in rural and remote areas were overrepresented.
We were also surprised at the over-representation of older specialists and rural and remote practitioners. We cannot be certain if this reflects level of interest in genomics, the multiple demands on early career practitioners' time, differing recognition of the importance of participating on research or other factors. Previous research from our group revealed conflicting views on the genomic literacy of early versus mid-career or senior specialists (McClaren et al., 2020, reference #20 in the manuscript), somewhat dispelling the notion that recent graduate may be most interested in genomic medicine.

We have expanded the discussion of potential biases on p.13:

> 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.**

P 8 line 17 speaks about 70.7% of the respondents being physicians. Apparently experts in anesthesiology, OB/GYN, psychiatry and surgery do not count as physicians, which needs some more explanation. In The Netherlands, these medical specialists would all count as physicians.
In Australia these sub-disciplines are known as 'medical specialists' with their own professional medical colleges and training programs, separate to the Royal Australasian College of Physicians – the Australian & New Zealand College of Anaesthetists; Royal Australian College of Obstetricians & Gynaecologists; Royal Australian & New Zealand College of Psychiatrists; and Royal Australasian College of Surgeons. The Methods and Results have been amended in three places to provide this clarification.

Methods (p.5):

> 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**.

Methods (p.7):

> 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.

Results (p.8):

> Figure 2 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).

In addition, there is the Australian College of Rural and Remote Medicine, which has over 5,000 members but as this specialty was not recognised as a separate category in the data provided by the Medical Board of Australia, representativeness could not be determined. This is noted in the caption for Figure 2 (p.18):

> 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).**

Reviewer 3

3) Consider adding description about what is meant by genetics service. What this entails is unclear as a reader outside of Australia.

5) Adding information about what conditions genetic testing was covered by Medicare Benefit Scheme since this payment had a large effect on the data.
We have added the following details to the Methods (p.5):

> 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 genetic 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).**

OTHER AMENDMENTS

Editor

As BMJ Open publishes material under a creative commons licence, it is problematic to include copyrighted material. Please remove any supplementary material which is not your own and cannot be published as part of your manuscript under the CC-BY-NC licence.
We confirm all Supplementary Materials are our own.

Reviewer 3

1) Consider using a different color for the blue in the figures. I found it tough to read.
We have converted the figures to black and white.

2) Consider adding a consort plot for your exclusion data.
We have added a CONSORT plot to the Supplementary Materials and referenced the new figure in the Results (p.7):

> 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)**.

VERSION 2 – REVIEW

REVIEWER	O'Daniel, Julianne University of North Carolina at Chapel Hill, Genetics
REVIEW RETURNED	01-Mar-2021

GENERAL COMMENTS	The authors present data from a broad survey of Australian medical specialists regarding current and anticipated genomics practice as well as overview of desire for additional genomics support and education including topics and preferred formats. This study is timely given the continual growth of clinical genetics spurred by advances in technology and clinical knowledge, as well as expanded access to clinical funding to cover test cost. The results are compelling and the authors have, for the most part, responded to the prior reviews. I believe the manuscript could be improved with minimal, additional edits. The discussion could be further enhanced with contextualization regarding the survey population. For instance, following this statement: "Tests reimbursed at the time of this survey are most typically requested by oncologists, clinical geneticists, haematologists, immunologists, paediatricians, obstetricians, nephrologists and neurologists." -considering reminding readers which of these groups are represented in your survey vs those who will be reported elsewhere. It is also worth commenting on practice models and the benefits/detriments of shifting some genetics specialty care to non-geneticists. This may be done for patient access, or workforce shortages and long waits for consultation. Although lack of confidence is a plausible explanation for respondents preference for a model which involved referral to or considerable support from genetics services, could it also be limitations in total visit time and competing priorities of health messages for the non-geneticists with their patients?

REVIEWER	Cornel, Martina VU University Medical Center, Clinical Genetics
REVIEW RETURNED	15-Feb-2021

GENERAL COMMENTS	Many of my suggestions have been adequately addressed. However, the first paragraph of the Methods section still is not quite clear to me. In the beginning of the methods section now “RACP” is no more defined (p5 l 17). May I suggest to move the sentence “After successful completion of final examinations, they become a Fellow of the relevant medical college.[23]” after “surgeons” in line 9 and to write RACP in full.

	To me as non-native speaker, the sentence “We also excluded radiologists and pathologists as in Australia they typically perform investigations than requesting genomic tests” sounds like incorrect grammar. Do you mean: “perform other investigations rather than…” The PDF mentions some Errors.

VERSION 2 – AUTHOR RESPONSE

REVIEWER 1

The discussion could be further enhanced with contextualization regarding the survey population. For instance, following this statement: "Tests reimbursed at the time of this survey are most typically requested by oncologists, clinical geneticists, haematologists, immunologists, paediatricians, obstetricians, nephrologists and neurologists." -considering reminding readers which of these groups are represented in your survey vs those who will be reported elsewhere.

We have included the following sentence at the bottom of p.11:

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 (excepting clinical geneticists and oncologists, who were not the focus of this study). At the time…

It is also worth commenting on practice models and the benefits/detriments of shifting some genetics specialty care to non-geneticists. This may be done for patient access, or workforce shortages and long waits for consultation. Although lack of confidence is a plausible explanation for respondents preference for a model which involved referral to or considerable support from genetics services, could it also be limitations in total visit time and competing priorities of health messages for the non-geneticists with their patients?

We have added further discussion to the paragraph at the top of p.12:

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. Currently, respondents lack confidence…

REVIEWER 2

The first paragraph of the Methods section still is not quite clear to me. In the beginning of the methods section now “RACP” is no more defined (p5 l 17). May I suggest to move the sentence “After successful completion of final examinations, they become a Fellow of the relevant medical college.[23]” after “surgeons” in line 9 and to write RACP in full.

Our apologies for not making this clearer in the earlier draft. We have reordered the paragraph, and added more detail, to reflect the chronological order of medical training in Australia. We only refer to the Royal Australasian College of Physicians here so have deleted the acronym. The Methods on p.5 now read:

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.

To me as non-native speaker, the sentence “We also excluded radiologists and pathologists as in Australia they typically perform investigations than requesting genomic tests” sounds like incorrect grammar. Do you mean: “perform other investigations rather than…”

We have amended the typographical error at the bottom of p.5. Thank you for picking this up.

…as in Australia they typically perform investigations rather than…

VERSION 3 – REVIEW

REVIEWER	O'Daniel, Julianne University of North Carolina at Chapel Hill, Genetics
REVIEW RETURNED	25-May-2021

GENERAL COMMENTS	I believe the authors have fully addressed all comments and concerns. This is a timely study and should be published.