Nisselle, King et al. (2021) Measuring physician practice, preparedness and preferences for genomic medicine: a national survey. ***BMJ Open***

SUPPLEMENTARY MATERIALS

Nisselle, King et al. (2021) Measuring physician practice, preparedness and preferences for genomic medicine: a national survey. ***BMJ Open***

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

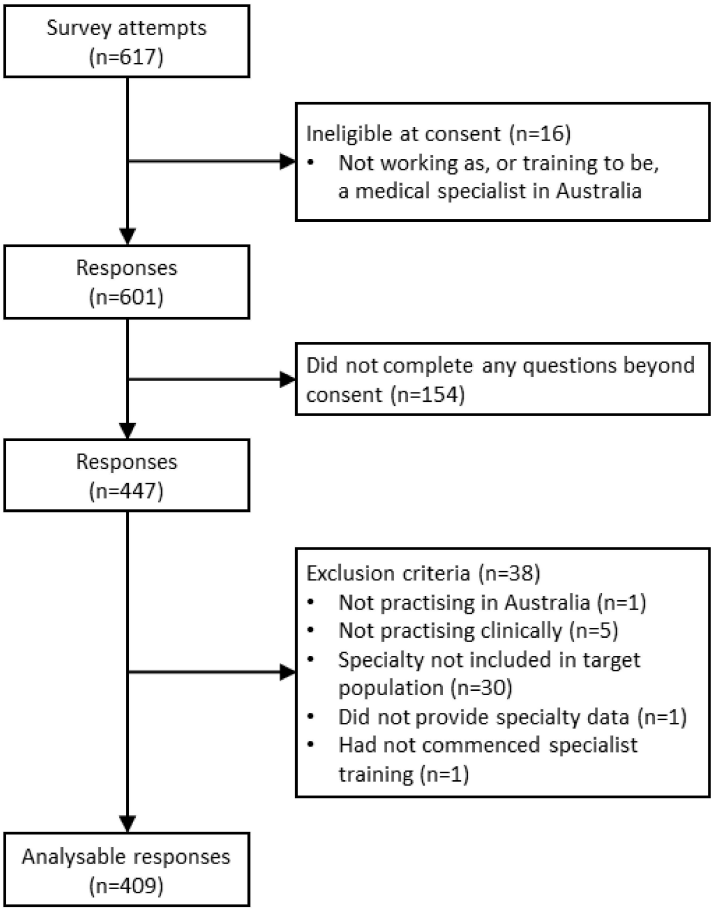

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Table S1. *Conditions for which genetic/genomic testing was covered by Medicare Benefit Scheme at the time of survey deployment in 2017.*[1]

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, 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 **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;

** Definitions were provided for these terms*