centre for population genomics

murdoch children's research institute

Garvan Institute of Medical Research

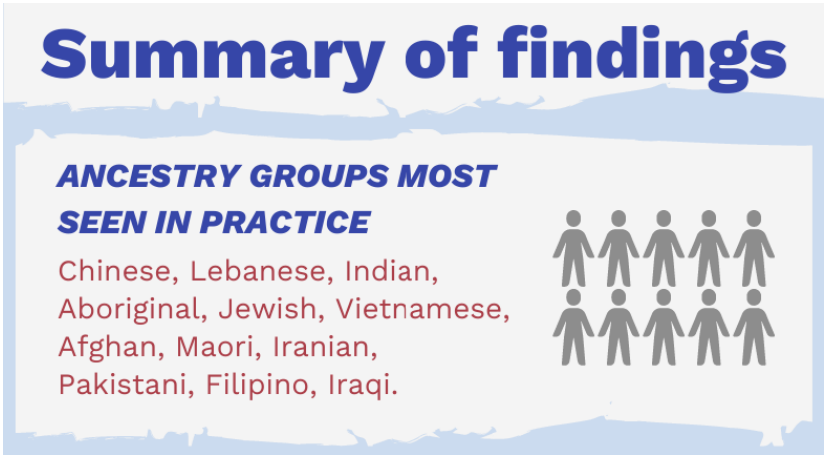
Diversity in genetic services and genomic datasets

A 2021 survey of clinical geneticists and genetic counsellors working in Australia

22 February 2022

Introduction

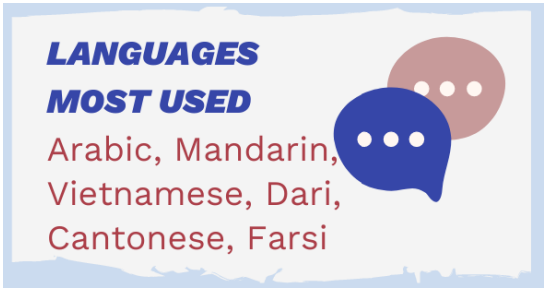
The Centre for Population Genomics (CPG) conducted a survey of Australian clinical geneticists and genetic counsellors between 5 October and 10 December 2021. The online survey was distributed to the members of the Human Genetics Society of Australasia (HGSA), whose membership includes 146 clinical geneticists and 359 genetic counsellors, and to subscribers to the Australian Genomics (AG) newsletter.



The CPG is building a genomic reference database that aims to represent the diversity of the Australian population and address the underrepresentation of people of non-European ancestry in international databases. This work will begin through in-depth community engagement and co-design with individual communities. Through this survey, we aimed to identify communities for whom the absence of reference data is most urgent to help us decide where to start. We asked participants about the groups they see most often in practice, their perceptions of the burden of genetic illness in these groups, and the challenges they experienced in accessing relevant data.

The participants

Forty-seven practising clinicians completed the survey. The majority were genetic counsellors (n=33), with 11 clinical geneticists taking part, and three participants reporting that they were involved in other clinical genetics or genetic counselling work. The majority of participants were practising in New South Wales (n=25, 53.2%), with a quarter in Victoria (n=11). Five participants were from Western Australia and two from Queensland, while four had practised in more than one state including South Australia, Tasmania, and the ACT.



Languages used in clinical services

Ninety-five percent of participants (n=43) said that they had used translators and interpreters in their work. We asked participants to select the top five languages that they had used in order of frequency of use. The language for which clinicians most commonly needed an interpreter or translator was Arabic, followed by Mandarin, Vietnamese, and Dari, with Cantonese and Farsi ranked equal fifth.

Ancestry groups seen in clinical services

We gave participants a list of ancestries grouped under the census categories in which we know there are people under-represented in genomic data: Oceanian,

North African and Middle Eastern, South-East Asian, North-East Asian, Southern and Central Asian, and Sub-Saharan African peoples. We asked participants to select the ancestry groups from this list that they had ever seen in their practice due to a genetic illness. The ancestry groups that participants selected formed the basis of subsequent questions about the burden of disease and diagnostic challenges.

Table 1. Ancestry groups most seen in practice

Rank	Ancestry Group	Score
1	Chinese	68
2	Lebanese	53
3	Indian*	47
4	Aboriginal	39
5	Jewish	32
6	Vietnamese	29
7	Afghan	22
=8	Maori, Iranian, Pakistani	14
9	Filipino	10
10	Iraqi	9

*Does not include specific South Asian ancestry groups (e.g. Bengali, Punjabi etc.)

We then asked participants to rank the groups that they had seen most in their practice in order of frequency from one to five. Ancestry groups were given a score based on this ranking (reverse-scored) and multiplied by the number of times ranked. The ancestry group that participants most commonly reported having seen in practice were Chinese Australians, followed by Lebanese and Indian Australians (See Table 1). The analysis in the following sections focuses on these twelve groups ranked in the top 10, with three groups being equal 8th.

Ease of access to reference data for different ancestry groups

Twenty-four participants said that they used genomic reference databases to inform diagnoses and interpretation of variants. These participants were asked how easy or difficult it was to access relevant reference data for each of the ancestry groups that they reported having seen in their practices. Of the groups most commonly seen, clinicians experienced the most difficulty accessing information for Aboriginal Australians and Maori. Of the participants responding to this question for Aboriginal Australians, an overwhelming majority (83.3%) said that accessing relevant data for this group was very difficult, with 73.3% reporting the equivalent for Maori.

ACCESSING RELEVANT DATA

Very Difficult
Aboriginal, Maori

Moderately Difficult
Vietnamese, Filipino, Afghan, Lebanese, Iranian

Not Difficult
Jewish, Chinese

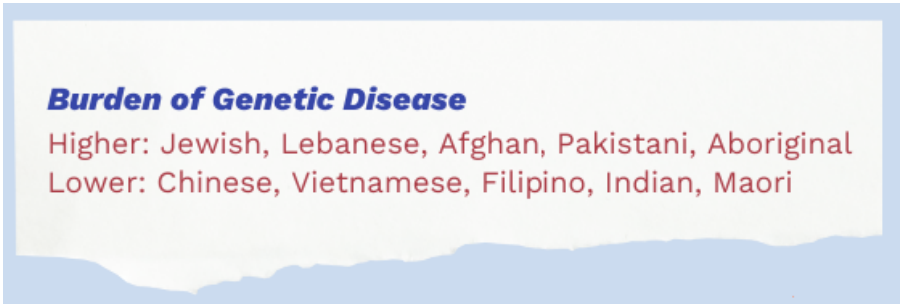
The majority found accessing information for Vietnamese, Filipino, and Afghan Australians to be moderately or very difficult. In contrast, clinicians reported little difficulty in accessing information for Jewish and Chinese Australians. Seventy-five percent said they had no difficulty accessing info for Jewish Australians, while 52.9% reported the same for Chinese Australians.

Overall, participants reported moderate difficulty in accessing data for Middle-Eastern groups. The majority responding to the question for Iraqi

Australians said that it was moderately difficult to access relevant data (86.7%). Similarly, 85% found it moderately difficult to find reference data for their patients of Lebanese descent, and 77.8% for patients of Iranian ancestry.

Diagnostic rates and burden of disease

Of the groups that they had ever seen in practice, we asked clinicians which they perceived to have lower diagnostic rates compared to other Australians because of a lack of reference data. The majority of participants who had ever seen Aboriginal Australians in their practice perceived this group to have lower diagnostic rates (70.7%). A sizable proportion of clinicians (37.1%) similarly perceived Maori to have lower diagnostic rates. About a quarter of clinicians perceived challenges for diagnosis for Vietnamese (27.8%), Afghan (26.7%), and Filipino (25.7%) Australians. Few perceived diagnostic challenges for Jewish (7.7%), Chinese (12.8%), or Iraqi (13.9%) Australians.



Participants were also asked which of the groups they had ever seen in practice they perceived to have a higher than average burden of genetic disease. The groups where a large number of clinicians perceived higher than average burden of genetic disease included Jewish (42.1%), Lebanese (38.7%), Afghan (36.7%), and Pakistani (32.3%) Australians. Over a quarter also perceived Aborginal Australians to experience a higher burden of genetic illness (26.8%). None of our participants perceived Chinese Australians to experience higher than average genetic disease, and few thought this was the case for Vietnamese (2.8%), Filipino (5.7%), Indian (7.9%), or Maori Australians (8.6%).

Groups to prioritise

Participants were asked which groups of the ones they had ever seen in practice they would prioritise for inclusion in CPG's reference database. Of the 41 participants who had ever seen Aboriginal Australian patients, almost half (n=20, 48.8%) said that they would prioritise Aboriginal Australians for CPG's reference database. Four of the 31 participants who had ever seen Lebanese Australians said they would prioritise Lebanese Australians.

Conclusion

Through an ethical, scalable approach to recruitment for CPG's planned reference database, our aim is to eventually include all Australians who are underrepresented in international databases accessible to Australian clinicians. In-depth co-design will only be possible with a small number of groups at any one time. The results of this survey will inform CPG's decisions about which groups to prioritise in the first stages of work.

We will consider these results in the context of a number of other factors. A key criterion for our prioritisation will be the overall size of the population of people from the different ancestry groups living in Australia. We will also consider other projects underway internationally that focus on improving genomic data for Australian ancestry groups, provided the diaspora communities in the project countries reflect the genetic ancestry of Australian communities, and the databases are accessible to Australian clinicians.

Finally, we aim to ensure that the reference database project provides for the equitable representation of Australian people descended from *all* regions currently missing from international databases, regardless of population size. From the beginning of the project, we will work towards addressing the underrepresentation of Australians of diverse backgrounds with ancestries that include Oceanian, North African and Middle Eastern, South-East Asian, North-East Asian, Southern and Central Asian, and Sub-Saharan African.

Next steps

CPG is now in the process of planning our work with the first communities we will engage in indepth co-design. To learn more about how we have been thinking about community engagement and co-design, you can read our literature review informing our approach here:
https://www.garvan.org.au/research/population-genomics/publications/croy-et-al-2021-towards_an_inclusive_genomics-cpg.pdf

If you were unable to fill in the survey but would like to provide us with information that you think will be helpful to us, please get in touch with CPG's Inclusive Genomics Lead, Maia Ambegaokar at
maia.ambegaokar@populationgenomics.org.au

Thank you to all the clinicians who took the time to fill out our survey! You have helped us to prioritise the communities to engage with in the first stages of our work.