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Sharing Genomic Data From Clinical Testing with Researchers: Public Expectations of Clinical Genomic Data Management in Queensland, Australia.

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Abstract

Background: There has beenconsiderable investment and strategic planning to introduce genomic testing intoAustralia's public health system. As more patients' genomic data is being held by the public health system, there will be increased requests from researchers to access this data. However, sharing of genomic data comes with a unique set of ethical, legal and social considerations that are not necessarily reflected in current health information management policies and legislation. It is important that public policy reflects public expectations for how genomic data that is generated from clinical tests is used.To inform public policy and discussions around genomic data sharing, we sought public opinions on using genomic data contained in medical records for research purposes in the Australian state of Queensland.

Methods: A total of 1,233 participants completed an online questionnaire between February and May 2019.

Results: Most participants wanted to be given the choice to have their genomic data from medical records used in research. Their expectations onhow often they needed to be approached for permission on using their genomic data, depended on whether thedata was identifiable or anonymous. Participants were most concerned with genomics data sharing that could lead to discrimination (insurance and employment), data being used for marketing, data security, orcommercial use.

Conclusions: Most participants were willing to share their genomic data from medical records with researchers. However,the existing policies related to this process in Queensland do not reflect public expectations for how this is achieved. Here we present options available to public health service to better reflect public expectations in clinical genomic data management and use.

Background

Australia is in the midst of a surge in public investment in clinical genomics through programs that focus on accelerated health service implementation and funding for translational research (1). In 2016, the Queensland State government committed $25 million to the accelerated implementation of genomics into public healthcare (2). Queensland has a population of aproximately 5 million and a statewide public health system. The mainstreaming of genomics into Queensland's clinical practice will result in an increasing amount of genomic data being held by the health system.

Australia's public health system is predicated on patient health data, in both aggregated and individual forms, being used for research and quality improvement activities to improve patient services and outcomes (3). In Queensland, there is legislation and associated policies that outline how and when health information can be used for research (4–7). There are currently no specific policies related to sharing of clinically derived genomic data with researchers. Any application to access genomic data for research purposes is considered under the policies related to general health information (4, 5) and by Human Research Ethics Committees (HREC) using national standards (8).

In Queensland, anonymised health information can be shared with researchers for ethically approved projects without individual consent in circumstances where health information cannot be directly or indirectly linked back to the patient (9). Under certain circumstances, identifiable or re-identifiable health information can also be shared with researchers for ethically approved research (4). This can occur when: (a) the patient has provided specific consent to participate in the project, or (b) the Director General of Health, or his/her delegate, has approved a Public Health Act (PHA) application in circumstances where researchers are unable to obtain individual consent, or it is deemed inappropriate or practically infeasible to contact patients. When a PHA application is sought, the research project must fulfil a waiver of consent criteria stipulated in the National Statement (8).

There have been a number of international studies that looked at public, patient and research participant perspectives on various aspects of genomic data sharing. These inquired about trustworthiness, risk, sharing preferences and concerns (10–15). Participant preference for sharing their genomic health information with researchers can differ due to jurisdictional, societal and demographic differences between study populations and types of linkage to personal identifiers proposed for the study (10–15). Recent work in Australia on health information and biospecimen sharing has observed an overall public willingness to participate in research (16–18). This is caveated by differences of opinions in use of identifiable data (17, 18), the need to know who the data recipients are (16, 17), and a desire for autonomy in providing permission (16, 18).

This study explores public opinions related to the sharing of genomic data for research from clinical records. Questions were framed around preferences relating to identifiable and anonymous genomic data or biological samples. The questionnaire was drafted in the context of current Queensland Health (QH) policies around data sharing for research purposes.

Methods

Recruitment

Eligible participants were adults aged 18 years or over who were residents of Australia at that time, according to the postcode of residence provided. Therefore, individuals who reported living overseas were not eligible. Participants were recruited via electronic direct marketing, QIMR Berghofer's magazine (LifeLab), or social media posts on QIMR Berghofer's Facebook and Twitter accounts. The electronic direct marketing contacted individuals who were on a QIMR Berghofer mailing list and who have previously subscribed to be notified about the Institute's future research. The questionnaire was available online for 16 weeks from February 2019 to May 2019.

Questionnaire Design

The cross-sectional online questionnaire (Additional file 1) comprised three sections: (A) socio-demographics, (B) permissions and preferences for either genomic data or biological samples sharing, and (C) concerns about genomic data sharing. The questionnaire contained 16 questions (Additional File 1; Section A: Q1 to Q7, Section B: Q8 to Q14, Section C: Q15 to Q16) and took about 10–15 minutes to complete. Each section used a mixture of questions and answer formats including: single response, multiple responses, categorical responses, Likert scales, and open text box.

In our study, we surveyed both Queensland and non-Queensland residents. The interstate cohort was included since: (1) there are cross jurisdictional agreements for the use of QH services by interstate patients, (2) interstate residents may have previously been residents of Queensland, and (3) some Queensland resident may not have experienced QH services. This questionnaire is based on a perception of QH and that perception can come from multiple sources that are not just based on personal experiences.

Questions related to permission for genomic data sharing were informed by the current QH policy on data sharing and consent (Q8 to Q11) (Additional file 1). Questions related to preferences for genomic data sharing (Q12 to Q14) are based on the categories and options used by the Australian Genomics Health Alliance CTRL platform for dynamic consent (19), which were derived from the Global Alliance for Genomic and Health's Data Use Ontology technical standard (20). Response options provided for concerns about genomic data sharing were based on a previous international public survey (21, 22), and then modified for this study's purpose based on feedback from consumer representatives and expert community members.

Participants were supplied with a definition of the terms *identifiable* and *anonymous* in the questionnaire (Additional file 1). Depending on the genomic data set requested by researchers, these categories and the provided definitions are a simplification of the complexity of anonymising genomic data in practice. The choice of definitions provided was based on the terms used in QH data sharing policies (9) and testing of comprehension by wider members of the community.

Analysis

For comparison of groups, the response categories of some demographics were grouped into fewer categories. Age was collapsed from the 10 year increments into three categories: 18 to 34 years, 35 to 55 years, or 55 years and over. Education was collapsed to university and non-university educated, while State to Queensland and non-Queensland based on postcode. Participants who were 'unsure' if they have had genetic or genomic testing were combined with those who answered 'no' to this question. For the gender demographic question, participants who selected 'other' were excluded from gender-based analysis due to the low number (n = 7), but were included in all other analyses. We categorised postcodes into areas of most disadvantaged and most advantaged using Socio-Economic Indexes for Areas (SEIFA) Index of Relative Socio-Economic Advantage and Disadvantage (IRSAD) (23). SEIFA local government postcode list was also used to categorise postcodes to metropolitan and regional categories.

We used a complete case analysis and thus respondents with missing information were excluded. Categorical data was summarised using counts and percentages and compared using a chi-squared test. Statistical significance was set at $p < 0.01$. Questions with multiple response options were dichotomised to compare between groups (Additional File 2). The open text response question (Q16) related to concerns was thematically analysed using a manual process. Analyses were conducted in Stata (version 15.1).

Results

Demographics

A total of 1,658 participants responded to the questionnaire. After exclusion of incomplete responses (n = 422) and those either with international postcodes or without an identifiable Australian postcode (n = 3), there were 1,233 complete responses that qualified for the analysis.

Participants ranged from 18 to over 75 years of age (Table 1). The majority of them were aged 55 years or more (69%, n = 856), female (67%, n = 832), while participants aged 18 to 35 years being under represented (4%, n = 52). Participants were disproportionally from higher socioeconomic status areas (combined ISRAD 3 to 5: 78%, n = 959). While participants from every Australian state were represented in the survey, over half of them were Queensland-based (55%, n = 677).

Table 1
Socio-demographic characteristics of questionnaire participant

Demographic variables (n = 1233)	All participants N (%)
Gender	
Male	394 (31.95)
Female	832 (67.48)
Other	7 (0.57)
Age (years)	
18–24	18 (1.46)
25–34	34 (2.76)
35–44	85 (6.89)
45–54	240 (19.46)
55–64	388 (31.47)
65–74	368 (29.85)
75+	100 (8.11)
Education	
University	772 (62.61)
Non-university	461 (37.39)
Didn't complete year 10	13 (1.05)
Year 10 or equivalent	86 (6.97)
Year 12 or equivalent	124 (10.06)
TAFE/Apprenticeship or equivalent	238 (19.30)
State of residence	
Queensland	677 (54.91)
Non-Queensland	556 (47.09)
New South Wales	206 (16.71)
Victoria	144 (11.68)
South Australia	55 (4.46)
Western Australia	69 (5.60)
Tasmania	31 (2.51)
Australia Capital Territory	44 (3.57)
Northern Territory	7 (0.57)
Worked in life science	
No	1,149 (93.19)
Yes	84 (6.81)
Worked in healthcare	
No	843 (68.37)
Yes	390 (31.63)
Had genetic or genomic testing	
No	747 (60.58)
Yes	421 (34.14)
Unsure	65 (5.27)
SEIFA (ISRAD)	

Demographic variables (n = 1233)	All participants N (%)
1 (most disadvantages)	110 (8.92)
2	163 (13.22)
3	194 (15.73)
4	342 (27.74)
5 (most advantaged)	423 (34.31)
Location	
Metropolitan	755 (61.23)
Regional	478 (38.77)

The majority of participants were university educated (63%, n = 772) and almost one-third have worked in healthcare (32%, n = 390) (Table 1). One-third of participants reported that they have had a genetic or genomics test in the past (34%, n = 421), which could have been clinical diagnostic test, participation in genomic research, or direct-to-consumer testing (health or recreational testing (24)).

Permission For Genomic Data Sharing

Overall, most agreed (86%, n = 1,063) that QH should ask individual permission before sharing identifiable genomic data with researchers, but only one-third (36%, n = 440) considered individual permission necessary when sharing anonymous genomic data (Table 2).

Table 2
Participant preferences for when permission is sought for sharing genomics data and biological samples with researchers (N = 1233).

	Identifiable genomic data N (%)	Anonymous genomic data N (%)	Identifiable biological samples N (%)	Anonymous biological samples N (%)
Should Queensland Health ask your permission before allowing researchers to access the following from your medical record?				
Strongly agree	727 (58.96)	217 (17.60)	705 (57.18)	213 (17.27)
Agree	336 (27.25)	223 (18.09)	340 (27.58)	219 (17.76)
Undecided	52 (4.22)	132 (10.71)	58 (4.71)	135 (10.95)
Disagree	82 (6.65)	416 (33.74)	87 (7.06)	413 (33.50)
Strongly disagree	36 (2.92)	245 (19.87)	43 (3.49)	253 (20.52)
Overall agreement for asking permission [a]	1,063 (86.21)	440 (35.69)	1045 (84.76)	432 (35.03)
Overall disagreement for asking permission [b]	118 (9.57)	661 (53.61)	130 (10.55)	666 (54.02)
How often should Queensland Health ask for permission to give researchers access to the following from your medical record?				
Every time	807 (65.45)	235 (19.06)	808 (65.53)	239 (19.38)
Sometimes	50 (4.06)	78 (6.33)	46 (3.73)	79 (6.41)
Only once	313 (25.38)	455 (36.90)	310 (25.14)	444 (36.01)
Never	63 (5.11)	465 (37.71)	69 (5.60)	471 (38.20)
a: Overall agreement is calculated as the sum of 'agree' and 'strongly agree'.				
b: Overall agreement is calculated as the sum of 'disagree' and 'strongly disagree'.				

Two-thirds nominated that QH should ask their permission either every time or sometime before their identifiable genomics data (69%, n = 808) would be shared with researchers, with a further quarter preferring to be asked only the first time (25%, n = 313) (Table 2). For anonymous genomics data, preferences for being asked only once was higher(37%, n = 455) than the need to ask at least sometimes (25%, n = 313). There was little difference in the observed preferences for seeking permission for biological samples when compared with genomic data (Table 2).

Under half of all participants stated they would allow another person to give permission on their behalf, once they are no longer able to (44%, n = 538) (Table 3), with family members being the most preferred option (62%, n = 335), followed by nominated legal representative (e.g. power of attorney) (48%, n = 258).

Table 3
Participant preferences for when permission is sought for sharing genomics data when patient is no longer able.

	Total N (%)	Family member N (%)	Nominated legal representative N (%)	Doctor N (%)	HREC N (%)	Data governance N (%)
Do you think someone else should be able to give permission for researchers to access your anonymous genomic data from medical records if you are no longer able? (N = 1233)						
No, only I can give permission	289 (23.44)	-	-	-	-	-
No, data freely available	406 (32.93)	-	-	-	-	-
Yes	538 (43.63)	-	-	-	-	-
If Yes, *who would you prefer to give permission for your anonymous genomic data to be used in research on your behalf? (you can select multiple answers) (N = 538)*						
Total N = 538 (100.00)	-	335 (62.27)	258 (47.96)	109 (20.26)	104 (19.33)	49 (9.11)

Preferences For Genomic Data Sharing

There was substantial variations in participant preferences for the organisations with which they would share their genomic data, ranging from 13–92% for anonymous and from 0.7–73% for identifiable data (Table 4). Overall, participants were between 12% and 32% less likely to share their identifiable than anonymous genomic data.

Table 4
Participant preferences for organisation that they would choose to share their genomics data (N = 1,233).

	Yes n (%)	Yes n (%)	Difference [a] (%)
What organisations would you share your …	*Anonymous genomic data*	*Identifiable genomic data*	
Australian not-for-profit research organisations	1,136 (92.13)	894 (72.51)	19.62
Australian universities and research institutes	1,140 (92.46)	881 (71.45)	21.01
Australian government	615 (49.88)	220 (17.84)	32.04
Overseas not-for-profit research organisations	679 (55.07)	285 (23.11)	31.96
Overseas universities and research institutes	716 (58.07)	337 (27.33)	30.74
Overseas governments	214 (17.50)	30 (2.43)	15.07
Commercial company	209 (15.41)	32 (2.60)	12.81
Publically available	158 (12.81)	9 (0.73)	12.08
What types of research would you share your anonymous genomic data with?			
Research specific to a condition I have	1,177 (95.46)	-	-
Research into other diseases and conditions	1,118 (90.67)	-	-
General population health research	1,072 (86.94)	-	-
Ancestry research	807 (65.45)	-	-
Unspecified future research	590 (47.85)	-	-
a: Difference between participants that would share their anonymous and their identifiable genomic data.			

The majority of participants would share their genomic data with Australian universities and research institutes (Anonymous: 92%, n = 1,140; Identifiable: 71%, n = 881), or not-for-profit organisations (Anonymous: 92%, n = 1,136; Identifiable: 73%, n = 894) (Table 4).

Overseas governments (Anonymous: 18%, n = 214; Identifiable: 2%, n = 30), commercial companies (Anonymous: 15%, n = 209; Identifiable: 3%, n = 32) and data being made publically available (Anonymous: 13%, n = 158; Identifiable: 1%, n = 9) rated the lowest for sharing both anonymous and identifiable genomic data.

Nearly all participants would agree to share anonymous genomic data for research of a disease they have (95%, n = 1,177) or other diseases or conditions (91%, n = 1,118), while slightly fewer would for general public health research (87%, n = 1,072). About half of all participants would be happy to share their anonymous genomic data for unspecified future research (48%, n = 590) (Table 4).

Concerns About Genomic Data Sharing

The majority of participants expressed high concerns about potential insurance discrimination (87%, n = 1,031), marketing companies (82%, n = 1,016), employment based discrimination (80%, n = 992), genomic data being made publically available (70%, n = 869), stigmatisation (63%, n = 774), and ethnic/racial discrimination (62%, n = 774) (Table 5). In comparision, under one-fifth of participants felt the same high concern about family finding out about their health results (20%, n = 242), upsetting genetic relatives (20%, n = 241), or data being used for quality improvement in QH diagnostics (18%, n = 216).

Table 5
Participants levels of concern associated with sharing genomic data for health records with researchers.

	Very concerned	Moderately concerned	Somewhat concerned	Slightly concerned	Not concerned
	N (%)	N (%)	N (%)	N (%)	N (%)
Insurance companies using my genomic data to discriminate against me	1,031 (83.62)	76 (6.16)	51 (4.14)	33 (2.68)	42 (3.41)
Marketing companies targeting me to sell me products	1,016 (82.40)	102 (8.27)	58 (4.70)	29 (2.35)	28 (2.27)
Employers using my genomic data to discriminate against me	992 (80.45)	77 (6.25)	57 (4.62)	26 (2.11)	81 (6.57)
My genomic data being made publicly available	869 (70.48)	127 (10.30)	104 (8.43)	76 (6.16)	57 (4.62)
Being labelled or stigmatised in some way	774 (62.77)	123 (9.98)	100 (8.11)	72 (5.84)	164 (13.30)
Ethnic or racial discrimination	769 (62.37)	98 (7.95)	81 (6.57)	55 (4.46)	230 (18.65)
Privacy of my personal details (e.g. name, date of birth, address)	727 (58.96)	205 (17.36)	136 (11.03)	94 (7.62)	71 (5.76)
My genomic data being used for research without my permission	588 (47.69)	214 (17.36)	149 (12.08)	126 (10.22)	156 (12.65)
Police using genomic databases with my details to investigate crimes	474 (38.44)	141 (11.44)	137 (11.11)	142 (11.52)	339 (27.49)
Receiving information about my future health that has no treatment option	431 (34.96)	194 (15.73)	191 (15.49)	155 (12.57)	262 (21.25)
My family finding out about my health results	242 (19.63)	140 (11.35)	140 (11.35)	142 (11.52)	569 (46.15)
Upsetting my genetic relatives, because my genomic information is similar to theirs	241 (19.55)	138 (11.19)	150 (12.17)	191 (15.49)	513 (41.61)
My genomic data being used by Queensland Health to improve services or diagnostic tests	216 (17.52)	202 (16.38)	185 (15.00)	168 (13.63)	462 (37.47)

The most common themes in the open text responses to concerns (n = 209) related to data security (24%, n = 49), commercial use or gains (13%, n = 28), autonomy in choosing to participate (11%, n = 22), and the use of genomic data without consent (10%, n = 20) (Table 6). Although the open text question intended to identify any other concerns about sharing genomic data from medical records for research, about 14% of respondants conveyed their support for sharing data for research purposes.

Table 6
Summary of identified themes from open text box responses of participant concerns.

Themes of concerns (n = 209)	N (% of those responding) [a]
Data security	49 (23.56)
Commercial use or gains	28 (13.46)
Autonomy of choice	22 (10.58)
Consent	20 (9.62)
Personal ethics on research type	15 (7.21)
Implications for self	15 (7.21)
Privacy	15 (7.21)
Access	10 (4.81)
Family implications	6 (3.85)
Trust	8 (3.85)
Misuse	7 (3.37)
Data management	6 (2.88)
Objection to sharing	4 (1.92)
Interest in area of research	4 (1.92)
Questionnaire comment	3 (1.44)
Positive response to sharing	30 (14.42)
a: 274 identified themes of concern identified from free-text comments of 209 respondents. Percentage calculated from number of participants that provided a text box response to Q14 (n = 209)	

Comparing Data Sharing Preferences Across Groups

Analyses based on demographic cohorts revealed some differences across the cohorts (Additional file 2). Preferences for when and how often permission was required varied across age, gender, education, expereince working in health care, or experience for genetic/genomic testing ($p < 0.01$) (Additional file 2, Tables S1 and S2). There was an observed difference in participants responses for those who were < 55 years old, had worked in health care, or were university-educated. A greater proportion of females than males indicated that permission should be required more than once for both identifiable genomic data and biological samples ($p < 0.01$). When participants had an experience of genetic/genomic testing, they disagreed that permission needed to be asked more than once ($p = 0.008$).

Participants who had previous experience with genetic testing, were ≥ 55 years or under 35 years old, or from Queensland more often agreed to sharing their anonymous genomic data for ancestry research (all $p < 0.01$). Conversely, participants who have worked in health care or have attained university education less often agreeed to sharing genomics data for ancestry research or unspecified future research ($p < 0.01$) (Additional file 2, Table S6). Age was also a factor in participants preference for third party permission, and organisations that they would choose to share identifiable and anonymous genomics data (Additional file 2, Table S3, S4 and S5).

Queensland residents expressed less high or moderate concern compared with non-Queensland residents about sharing their genomic data (Additional file 2, Table S7).

Discussion

Our questionnaire of public opinions of sharing genomic data from medical records found that participants were willing to share their genomic data with researchers. However, our results stongly suggest that this willingness is predicated on several caveats related to; availability of personal details, organisation that will be the recipient, and type of research being undertaken. These results confirmed the findings of previous studies related to health information and genomic data that have been observed in other public (16) and patients studies in Australia (17, 18).

Within QH there is precedence for certain types of health information to be considered sensitive and there are specific policies in place, examples being access to sexual health data (5). Currently, QH does not have any specific policies related to genomic data, thus general health information policies are routinely applied instead. In clinical genetic testing, there has been a debate surrounding the concept of genetic exceptionalism – which proposes that genetic and genomic data have special risks not observed in other types of health information and therefore, needs different considerations in data management and patient consent (25). Participants in a global study (that included Australians) who viewed genomic data as exceptional, were more willing to participate in research than those without genomic exceptionalism views (22), thus indicating that even if QH took a genomic exceptionalism position, people would still participate in research. However, patient education and suitable consent processes would need to be in place.

The health information sharing policies of QH (4) reflect the opinions of the majority of participants when considered in the context of identifiable genomic data. However, participants that want permission sort from a third party preferred family or legally nominated representatives to give consent when they are no longer able to consent, rather than doctors, data governance or HREC.

In contrast, QH's current policies around sharing of anonymous health information (9) do not reflect expectations of the public in relation to genomic data. While one-third of participants would accept for their data to be used without permission for research uses, the majority would require permission to be sought at least once.

Our anlysis did identify age, and educational and work related factors, as associating with participant preferences for genomic data sharing. With 35 to 55 year olds, and those that worked in health care tending towards a reluctance to share genomic data. Age, experience with poor health or genetic testing, and educational attainment have been identified as factors in both international and Australian studies of people's willingness to share health information and genomic data with researchers or to participate in biobanking (10–12, 14, 16, 18, 22). The association with age and educational attainment tend to vary depending on the location of survey participants (10–12, 16), whilst experience with genomics or poor health consistently associates with a willingness to share with research (14, 18, 22). Other studies have identified ethnicity/race and religiosity as influencing willingness to participate in genomic research (12, 26), which were not considered in this study.

The present study demonstrated apprehension for commercial organisations access genomic data for research that could be used for profit. This mistrust of for-profit companies has similarly been observed in studies based in other countries (10, 14). Notably, participants stated they were more likely to give permission to have overseas research organisations (not-for-profit, medical institutes and universities) have access to their genomic data than the Australian government. This is in direct contrast to a study where research by domestic governments was shown to be preferred over international researchers (27).

Based on open text responses to concerns, the desire to be asked permission seemed to stem from participants seeking autonomy over the ability to participate in research, wishing for their participation in research to reflect personal priorities and ethics, and a desire to know about the research in which they would be participating. Concerns over genomic data use outside of intended research did not limit participant's willingness to participate in genomic research, which confirms the findings of other studies (10, 22). Interestingly, some study participants conveyed a positive response to sharing data for research inspite of their concerns. Further exploration of the barriers and motivators to research participation would be useful to inform QH policies on clinical data sharing with research.

Limitations

The main limitation in the current study was there was a potential for bias due to over or under sampling of certain sub-populations of paticipants, such as based on age. While a large number of participants did enable us to consider responses of sub-populations, there is still the potential for bias due recruitment strategy and self-selection. These included the questionnaire only being available online, and recruitment material being primarily directed towards people with previous engagement with a research organisation. Both of which may lead to biased sample of participants that is not representative of the general population. Based on the findings of other research (18, 22), we anticipated that a questionnaire of a patient cohort may produce different views on genomics data sharing for research purposes. Investigation of this cohort's perspective is an essential next step in the genomic data sharing discussion.

Through the open text box questions some participants reported feeling concerns as questions (Q15 & 16) were ambiguous, as participants were not directed to consider an *anonymous* or *identifiable* scenario. As such, the findings related to concerns should be considered with caution. Genomics and its associated issues are technically difficult and multifaceted to explain, as such they do not lend themselves well to a multiple choice based questionnaires as it is hard to convey nuanced opinions (28). In our questionnaire, participants were not given a definition of genomic data; therefore, responses were based on participant pre-existing understanding of genomic data, which we expect to vary greatly within the cohort.

Options For Public Health Policy

The study highlights that current data sharing policies only partially reflect public expectation on clinical genomic data sharing for research purposes. Possible options for managing clinical genomic data sharing include:

Continue without change. Apply current policies for health information on genomic data. This assumes that genomic data is not a special data type.
Change research data sharing policies around genomic data. Clarify the health system's position on genomic data sharing, enabling data managers to make decisions on data access requests that are supported by policy.
Inform patients. Provide patients with information on potential secondary uses of their health information prior to genomic testing, so they can make choices related to what clinical data they are willing to have on their medical record.
Educate the public. Implement education programs to inform the public about how health information is used to improve health outcomes for patients and clinical data usage.
Research consent during clinical consent. During the consent process for clinical genomic testing, provide consent options for the use of genomic data for research.
Separate research consent. Establish programs to request broad consent to research from patients outside of clinical consultations, i.e. QH's previously trialled Giving InFormation To Research (GIFTR) (29).
Sharing research results. Support initiatives for patients and the public to engage with research outcomes. For example, public-facing research website, or supporting researchers to engage in post-study dissemination of findings to the community.

Conclusions

In the coming years, QH will be a repository for a large amount of clinically derived genomic data. With this, QH is likely to receive more requests from researchers to access this data using the existing data access practises and policies. This study indicates that, especially in the context of anonymous data, current policies may not meet the public expectation for autonomy in choosing how their clinical genomic data is shared with researchers.

This study demonstrates a high degree of variability in public willingness and preferences for sharing their genomic data with researchers. Here we identified multiple options available to healthcare systems for managing genomic data sharing. These range from policy based changes, through to patient or public engagement initiatives. What is most important is that there is an active decision on genomic data management rather than a continuation of existing data sharing policies without review. This will assist in aligning public expectations with health policy directives, whilst also including global genomic policy developments, bioethics considerations, and technology advancements.

Abbreviations

GIFTR: Giving InFormation To Research; HREC:Human Research Ethics Committee; IRSAD:Index of Relative Socio-Economic Advantage and Disadvantage; PHA:Public Health Act; QGHA:Queensland Genomics Health Alliance; QH:Queensland Health; SEIFA:Socio-Economic Indexes for Areas.

Declarations

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AUTHORS CONTRIBUTION

MV and NW conceived of the project and developed the manuscript. EM and CH advised on analysis and interpretation of data. MV and EM performed the analyses. SK provided specialist input to the data intreptation and manuscript preparation. All authors edited subsequent drafts. NW obtained funding and supervised the work. All authors read and approved the final manuscript.

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AVAILABILITY OF DATA AND MATERIALS

The datasets generated and analysed during the current study are not publicly available because participants did not give consent to share the data beyond the research team.

ETHICS APPROVAL AND CONSENT TO PARTICPATE

The questionnaire was approved by the QIMR Berghofer Human Research Ethics Committee (P3246). Approval was granted for the collection of anonymous data. Participants were provided with a written information sheet prior to starting the online questionnaire.

CONSENT FOR PUBLICATION

Not applicable

COMPETING INTERESTS

MV, SK, EM&CH have no competing interests to declare. NW is co-founder, minor equity holder and Board member of genomiQa.

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References

1. Stark Z, Boughtwood T, Phillips P, Christodoulou J, Hansen DP, Braithwaite J, et al. Australian Genomics: A Federated Model for Integrating Genomics into Healthcare. The American Journal of Human Genetics. 2019;105(1):7–14.
2. Green light for genomics [Internet]. Queensland Government. 2016 [cited 12 October]. Available from: http://statements.qld.gov.au/Statement/2016/10/12/green-light-for-genomics.
3. Australia. 2030: Prosperity Through Innovation [Internet]. Australian Government. 2017 [cited 2019 October 23]. Available from: https://www.industry.gov.au/sites/default/files/May%202018/document/pdf/australia-2030-prosperity-through-innovation-full-report.pdf.
4. Access to Confidential Health Information [Internet]. Queensland Health. 2019 [cited 2018 December15]. Available from: https://www.health.qld.gov.au/hiiro/html/regu/aces_conf_hth_info.
5. Privacy Plan [Internet]. Queensland Health. 2015 [cited 2019 November 05]. Available from: https://www.health.qld.gov.au/__data/assets/pdf_file/0027/439164/doh-privacy-plan.pdf.
6. (Queensland).
   Public HA. 2005, (Queensland).
7. (Queensland).
   Information PA. 2009, (Queensland).
8. NHMRC ARC. UA. National Statement on Ethical Conduct in Human Research 2007 (Updated 2018). Canberra: Commonwealth of Australia; 2007.
9. Queensland Data Linkage Framework [Internet]. Queensland Health. 2016 [cited 2019 October 28]. Available from: https://www.health.qld.gov.au/__data/assets/pdf_file/0030/150798/qlddatalinkframework.pdf.
10. Trinidad SB, Fullerton SM, Bares JM, Jarvik GP, Larson EB, Burke W. Genomic research and wide data sharing: views of prospective participants. Genetics in medicine: official journal of the American College of Medical Genetics. 2010;12(8):486–95.
11. Page SA, Manhas KP, Muruve DA. A survey of patient perspectives on the research use of health information and biospecimens. BMC Medical Ethics. 2016;17(1):48.
12. Sanderson SC, Brothers KB, Mercaldo ND, Clayton EW, Antommaria AHM, Aufox SA, et al. Public Attitudes toward Consent and Data Sharing in Biobank Research: A Large Multi-site Experimental Survey in the US. The American Journal of Human Genetics. 2017;100(3):414–27.
13. Kaufman DJ, Baker R, Milner LC, Devaney S, Hudson KL. A Survey of U.S Adults' Opinions about Conduct of a Nationwide Precision Medicine Initiative Cohort Study of Genes and Environment. PLoS One. 2016;11(8):e0160461.
14. Goodman D, Johnson CO, Bowen D, Smith M, Wenzel L, Edwards K. De-identified genomic data sharing: the research participant perspective. Journal of Community Genetics. 2017;8(3):173–81.
15. Jamal L, Sapp JC, Lewis K, Yanes T, Facio FM, Biesecker LG, et al. Research participants' attitudes towards the confidentiality of genomic sequence information. Eur J Hum Genet. 2014;22(8):964–8.
16. King T, Brankovic L, Gillard P. Perspectives of Australian adults about protecting the privacy of their health information in statistical databases. Int J Med Informatics. 2012;81(4):279–89.
17. Krahe M, Milligan E, Reilly S. Personal health information in research: Perceived risk, trustworthiness and opinions from patients attending a tertiary healthcare facility. J Biomed Inform. 2019;95:103222.
18. Liddell J, Bain C, Myles PS. Patient and community attitudes toward perioperative biobanking and genomic research. Anaesthesia intensive care. 2017;45(3):384–95.
19. Australian Genomic Health Alliance. CTRL - Managing your consent and participation in Australian Genomics research: Australian Genomic Health Alliance; 2019 [Available from: https://www.australiangenomics.org.au/resources/for-patients/your-personal-platform/.
20. Global Alliance for Genomics and Health. Data Use Ontology approved as a GA4GH technical standard: GA4GH; 2019 [Available from: https://www.ga4gh.org/news/data-use-ontology-approved-as-a-ga4gh-technical-standard/.
21. Middleton A, Niemiec E, Prainsack B, Bobe J, Farley L, Steed C, et al. 'Your DNA, Your Say': global survey gathering attitudes toward genomics: design, delivery and methods. Personalized Medicine. 2018;15(4):311–8.
22. Middleton A, Milne R, Howard H, Niemiec E, Robarts L, Critchley C, et al. Members of the public in the USA, UK, Canada and Australia expressing genetic exceptionalism say they are more willing to donate genomic data. European Journal of Human Genetics. 2019.
23. Technical Paper: Socio-Economic Indexes for Areas (SEIFA) 2016 [Internet]. Australian Bureau of Statistics. 2018. Available from: https://www.ausstats.abs.gov.au/Ausstats/subscriber.nsf/0/756EE3DBEFA869EFCA258259000BA746/$File/SEIFA%202016%20Technical%20Paper.pdf.
24. Savard J, Hickerton C, Tytherleigh R, Terrill B, Turbitt E, Newson AJ, et al. Australians' views and experience of personal genomic testing: survey findings from the Genioz study. Eur J Hum Genet. 2019;27(5):711–20.
25. Johnson SB, Slade I, Giubilini A, Graham M. Rethinking the ethical principles of genomic medicine services. European Journal of Human Genetics. 2019.
26. Middleton A, Milne R, Thorogood A, Kleiderman E, Niemiec E, Prainsack B, et al. Attitudes of publics who are unwilling to donate DNA data for research. European Journal of Medical Genetics. 2019;62(5):316–23.
27. Majumder MA, Cook-Deegan R, McGuire AL. Beyond Our Borders? Public Resistance to Global Genomic Data Sharing. PLoS Biol. 2016;14(11):e2000206.

28. Milne R, Morley KI, Howard H, Niemiec E, Nicol D, Critchley C, et al. Trust in genomic data sharing among members of the general public in the UK. USA: Human Genetics; 2019.
29. Health Informatics Society of Australia. HISA submission to Productivity Commission's Inquiry into Data Availability and Use. 2016 [cited 2019 December 12]. Canberra: Productivity Commission, [cited 2019 December 12]. Available from: https://www.pc.gov.au/__data/assets/pdf_file/0020/206813/sub199-data-access.pdf.

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