Human Genomics

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A consensus model for implementation of genomics in Queensland Health – a Delphi study

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Abstract

Background As genomic technologies in healthcare expand, health systems face integration challenges. This study aimed to consolidate collective expertise, experiences, and existing knowledge from local, statewide, and national genomics projects to inform a model of care for integrating and delivering genomic services in the Queensland public health system.

Methods We used a modified Delphi methodology with three survey rounds to achieve expert consensus on the model of care. We analysed quantitative data using descriptive statistics and qualitative data using content analysis.

Results From 112 invited experts, 43 participants took part. The model of care comprised 30 actions across three phases: pre-test, testing, and post-test. Consensus (≥ 75% agreement) was achieved on 25 of the actions, with 5 included with specific considerations. Ten recommendations were developed, including building sustainability, improving access to specialists through a distributed model, and investing in clinical-pathology partnerships and infrastructures.

Conclusion These findings may inform a comprehensive, stakeholder-informed, system-level implementation effort for mainstreaming genomic medicine that is practical, feasible, and acceptable to key stakeholders across the system. The three-phase model of care provides a structured approach to genomic service delivery and has the potential to significantly improve patient access and healthcare delivery.

Keywords Genomic medicine, Implementation science, Stakeholder consensus, Delphi methodology

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Background

Rapid advances in genomic medicine have seen diagnostic and therapeutic value established in fields such as oncology, pharmacology, and rare and undiagnosed diseases [1]. As healthcare systems prepare to integrate genomic technologies, they face implementation challenges including data integration and interpretation, workforce capacity, and social and ethical considerations [2]. Further, the complex nature of funding and delivery of genomic medicine, often through a mix of private and public health systems, can contribute to information siloing, test duplication and resource waste, inequitable patient access to care, and limited inter-jurisdictional collaboration [3–5]. To counter these challenges, implementing genomic medicine requires a comprehensive, system-wide approach to address equitable implementation [6].

In response, many countries have launched large-scale initiatives to accelerate the implementation of genomic medicine, including Australia [1, 7–10]. One notable example is the public health system of Queensland, Australia. Queensland Genomics, a 5-year program implemented in 2015, aimed to demonstrate the value of genomic medicine in everyday healthcare within its complex public health system [9, 11]. The program facilitated genomics uptake by uniting research, consumers, and the health system to promote system capability and infrastructure for genomic medicine [11]. Presently, clinical genomics in Queensland continues to demonstrate focal successes in translation, implementation, and advancement. These successes include the GenetiQs project to develop guidelines for genomic research with Aboriginal and Torres Strait Islander peoples [12] and the establishment of a collaborative Queensland Renal Genetics Clinic [13]. Conversely, ongoing challenges persist post-Queensland Genomics, including change management around program funding, administration, and expectations, alongside competition from health system priorities [14].

These Queensland experiences highlight that system consensus and coordination are less prominent, and future success will require greater system cohesion and synergy around shared priorities and planning among diverse stakeholders. The implementation of genomic medicine involves input from several stakeholders, including genetic and non-genetic clinicians, laboratory scientists, bioinformaticians, operational genomics staff, and importantly, consumer groups [15–17]. The involvement of stakeholders is crucial to ensure acceptability and feasibility of implementation activities [18]. However, each stakeholder group may have preferred approaches regarding comprehensive care to patients, potentially leading to disagreements and conflicts, and subsequently implementation problems such as lack of engagement and buy-in, risk of disjointed and ineffective investment, and ultimately inequitable access to care for patients [19, 20].

Therefore, it is important to consolidate opinions across the healthcare system to maximise the value-add of genomic medicine. Implementation science, the study of the uptake of evidence-based practice and research into regular use, can address this problem [6]. Using stakeholder-driven research methods such as the Delphi method or expert panels, consensus on implementation priorities that have the greatest benefits and reduce research waste can be identified [21]. Given the importance of stakeholder consensus in genomic medicine implementation, this study aimed to consolidate collective experiences, existing knowledge, and established models of care from statewide, national, and international genomics projects to develop a model of care for integrating and delivering genomic services in the Queensland public health system. The study objectives were to:

i) reach stakeholder consensus on an effective and broadly applicable model of care for genomic in the Queensland public health system,
ii) develop stakeholder-informed recommendations to guide the phased integration of the model of care.

Methods

Research design

To guide the study, a cross-organisational working group was established, with expertise in implementation science (SB, MVB, ZF), health services (SB, AM, MD, SM, SI), policymaking (SB, AM, MD), and medicine (AM). We selected a modified Delphi methodology with iterative survey rounds combining quantitative (e.g. rating scales) and qualitative (e.g. open-ended questions) approaches to reach consensus among stakeholders. The method was chosen for its ability to systematically consolidate expert opinions while allowing for refinement of ideas across rounds [22]. No consulting regarding study methodology took place. The study protocol had not been published previously.

The first survey round was designed based on relevant literature and the working group expertise, particularly focusing on genomics models of care across Australia [9–14]. This approach grounded the project in existing knowledge and aimed to reduce the number of rounds and alleviate participant burden [23]. Results from each round were aggregated and returned to participants, until consensus was achieved. The number of rounds was not decided a priori; instead, survey rounds were concluded when the working group determined that the collected data sufficiently addressed the study aims and objectives.

Ethics approval was granted by the Office of Research Ethics and Integrity at the University of Melbourne (reference number: 2024-30792-60552-4)

Study setting and recruitment

Queensland is a geographically large jurisdiction and home to over five million people. Clinical genomics in Queensland is primarily delivered through the public health system's sixteen hospital and health services, underpinned by statewide services for pathology (Pathology Queensland) and clinical genetics (Genetic Health Queensland). Additional mainstreamed genetic services are established in some health services, such as Children's Health Queensland.

Potential participants were identified and shortlisted by the working group using existing stakeholder lists maintained by the Office of Research and Innovation Genomics Program. These individuals included subject matter expertise, systems users or enablers and policy decision-makers for example, involved in the implementation of genomic medicine in Queensland. The group comprised clinical geneticists, genetic counsellors, clinicians with genomics interests, laboratory and scientific staff, department heads, and health system leaders. Individuals whose work was not primarily within Queensland were excluded. Participants were informed they did not need to have participated in the previous rounds to take part in the next round. Participants could also nominate others whose perspectives might be valuable. To emphasise that participation was voluntary, invitations were sent via email from a working group member with whom the invitee had no prior established relationship. Participants provided consent to participate in the study at the start of each survey. Healthcare consumers (patients, carers, and families) were not included in the Delphi rounds. Instead, their input was sought after each survey round via an established consumer group, the Queensland Genomics Consumer Advisory Group. This group comprised 12 consumer and carer representatives with diverse lived experience or interest in genomics. Membership reflected broad geographic and cultural diversity, including people living in rural and remote areas, people with disability or genetic conditions, First Nations peoples, and individuals from multicultural backgrounds. The group was established in March 2024 to act as an advisory body. Members were recruited through an expression of interest process using the Health Consumers Queensland statewide network recruitment program. They were selected based on their strong community networks and ability to represent broader community perspectives. The consumer group participated in a group discussion after each Delphi round, during which preliminary findings were presented. Their feedback was then used to refine the survey questions and response options for the subsequent round.

Data Collection

We conducted three online Delphi survey rounds between December 2024 and May 2025 (Fig. 1). A maximum of three rounds was deemed adequate to meet the study objectives within the available resources. The Action, Actor, Context, Target, Time (AACTT) framework guided the construction of one section of the surveys [24], generated with Qualtrics software. Given the complexity of integrating genomic medicine into the healthcare system, the AACTT framework is useful in providing a structured approach to map a model of care and analyse the specific actions performed by all individuals involved. The surveys were pilot tested by the working group and four additional team members at Queensland Health with an understanding of genomics. They were assessed for clarity, comprehension, average completion time, and ease of navigation, confirming suitability for the expert panel prior to distribution. The survey functioned as a structured consensus method to elicit expert consensus, not as a psychometric scale. Consequently, formal validity and reliability testing of the survey was not needed.

Round 1. Comprised of demographic questions and 37 actions specific to a genomic medicine model of care. Actions were derived from previous research [25–27], initiatives [7–10], and the working group's expert knowledge on genomic medicine. For each action, participants were asked to select (a) whether the action should be included in the genomics model of care, (b) from a pre-defined list, who 'the actor' (e.g., medical specialist) should perform the action (e.g., patient consent), and where 'the context' (e.g., public health setting) and when 'the timing' (e.g., immediately) the action should take place [24]. Additionally, participants were asked to, (c) suggest changes to the action's wording, and (d) specify necessary changes to current practices for the actions to occur. Participants were also given the option to skip to next action if they wished.

Round 2. The Round 2 survey included the same demographic questions and the actions that had reached consensus or had undetermined consensus for inclusion in the model of care from Round 1. For each action, participants reviewed a pre-determined list of actors, contexts, and timelines for further consideration and refinement (see Data Analysis). They were informed that all actions in the survey had either achieved consensus or had undetermined consensus in Round 1. The feedback was aggregated across all expert groups. Additionally, the Round 2 survey included participant-derived recommendations on necessary changes to current practices to establish the model of care within the Queensland public

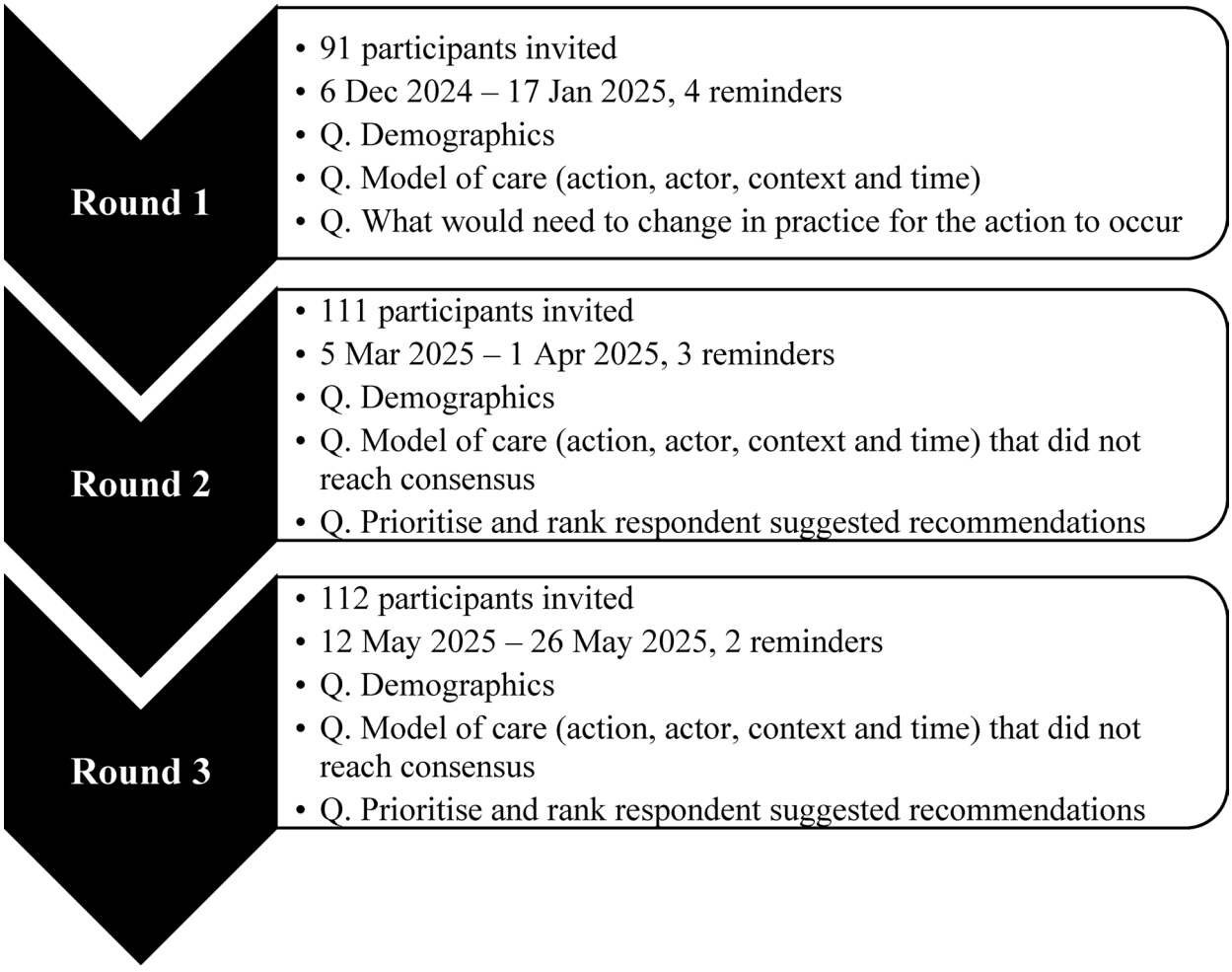

Fig. 1 Summary of the Delphi rounds

health system. Participants were asked to prioritise five recommendations they deemed needs to happen immediately and then rank the remaining recommendations according to what needs to happen in the near future, in the very far future, and what did not need to happen at all, with the option to provide a comment or justifications for their selection.

Round 3. Round 3 survey began with the same demographic questions, and actions with the refined pre-determined list of actor, context, and time, and recommendations on necessary changes to current practices. Similarly, the feedback was aggregated across all expert groups, and participants were informed that all actions in the survey had either achieved consensus or had undetermined consensus in Round 2. Participants then prioritised three recommendations they deemed needs to happen immediately, then the remaining recommendations according to what needs to happen in the near future, in the very far future, and what did not need to happen at all.

Data analysis

Quantitative data

Participant characteristics were analysed and presented descriptively. For an action to be included in the model of care, the threshold for consensus was set a priori at 75% [28], meaning that at least three-quarters of participants had to vote 'yes' for inclusion. Actions that received less than 50% were excluded. The remaining actions between the 50–75% threshold were included with explanations drawn from qualitative findings.

For the pre-defined list of options regarding the actor, context, and time of each action, the threshold for consensus was defined as the mean number of votes across all options. For example, if an action had three possible actors (nurse, medical specialist, genetic counsellor) and the votes were 3, 7, and 5 respectively, the consensus threshold would be the mean number of votes per option, which is 5. An option was considered to have reached consensus if it received votes equal to or greater than this mean value. In this example, the medical specialist and genetic counsellor options for this action were deemed

Table 1 Survey distribution

Surveys distributed	Round 1	Round 2	Round 3
	91	111	112
Surveys received (response rate)	35 (38.5%)	37 (33.3%)	27 (24.1%)
Complete surveys (completion rate)	21 (23%)	32 (28.8%)	25 (22.3%)
Incomplete surveys	14 (15.5%)	5 (4.5%)	2 (1.7%)

to have reached consensus and would be presented to participants in the next round. This approach was used instead of the conventional fixed percentage agreement because each action had multiple possible options for actor, context, and time from which participants could select.

Prioritisation of recommendations on essential changes to current practices was categorised based on the number of votes for each action to happen immediately, in the near future, in the very far future, and not needed to happen. Recommendations were classified as 'immediate or near-term' if over 75% of participants voted for them to be actioned either immediately or in the near future. They were classified as 'medium term' if 50–75% of participants voted for immediately or near-future actioning, with the remaining 25%–50% voting for 'very far future' or 'not needed'. Recommendations were classified as 'long-term or not needed' if more than 50% voted for 'very far future' or 'not needed' (i.e., less than 50% voted for 'immediately' or 'near future').

Qualitative data

Open-ended qualitative data in Round 1 were analysed using inductive content analysis to generate the participant-derived recommendations for necessary changes to current practice. This analysis also informed the refinement of the actions and their associated actors, time, and context options for Round 2 [29]. Data were sorted according to the action they corresponded to and coded. Codes were then grouped into categories based on similarity and continually revised and refined as more codes were added through the analysis process. The same process was applied following Round 2 and Round 3 to further refine the participant-derived recommendations. Between rounds, analysis was conducted by MVB and verified by ZF. Preliminary findings were discussed within the working group and subsequently presented to the consumer advisory group for additional feedback. After considering this feedback, the working group finalised the findings and incorporated them into subsequent surveys, allowing participants of those rounds to review and validate them.

Results

Details of the survey distribution, including response rate and completion rate, are summarised in Table 1. Participant characteristics (Table 2) were consistent across the three rounds. The number of laboratory stakeholders invited and participated in the study, although lower than other groups, was proportional to the number of staff in these posts. In round 3, there was a decrease in

Table 2 Demographics

		Round 1 (*n* = 21)		Round 2 (*n* = 32)		Round 3 (*n* = 25)	
		n	%	n	%	n	%
Role	Clinical staff	11	52.4%	23	71.9%	18	72%
	Laboratory	3	15.3%	4	12.5%	4	16%
	Department head & strategic decision-makers	7	33.3%	5	15.6%	3	12%
Gender	Female	15	71.4%	25	78.1%	18	72%
	Male	6	28.6%	7	21.9%	7	28%
	Others	0	0%	0	0%	0	0%
Age range	20–30	1	4.8%	3	9.4%	3	12%
	31–40	2	9.5%	8	25%	7	28%
	41–50	7	33.3%	11	34.4%	9	36%
	51–60	9	42.9%	8	25%	5	20%
	61–70	2	9.5%	2	6.3%	1	4%
Years of experience in own field	0–5	1	4.8%	2	6.3%	2	8%
	6–10	2	9.5%	7	21.9%	6	24%
	11–20	8	38.1%	12	37.5%	9	36%
	21+	10	47.6%	11	34.4%	8	32%
Years of experience in genomics	0–5	5	23.8%	9	28.1%	5	20%
	6–10	6	28.6%	8	25%	8	32%
	11–20	5	23.8%	9	28.1%	6	24%
	21+	5	23.8%	6	18.8%	6	24%

the number of participating department heads and strategic decision-makers. Participants demonstrated a range of experience levels, both in their own field and in genomics.

Proposed genomics model of care

The actions and their percentage agreements across the three survey rounds are presented in Table 3. Figure 2 shows the final model of care, comprising 30 actions across pre-test, testing, and post-test phases, covering the patient's journey from initial assessment to patient consent, sample collection and sequencing, to post-test follow-up and long-term management. Twenty-five actions reached consensus for inclusion, five had undetermined consensus and are included with caveats, and one was excluded.

The actors, contexts, and time for each action are detailed in Supplementary Material 1. Actors may include medical specialists and nurse specialists with genomic expertise, primary care clinicians, clinical and laboratory geneticists, genetic counsellors, and the genomic multidisciplinary team. Participants indicated that multiple actors may perform each action, subject to training, scope of practice, and expertise. Contexts may include both public and private health settings, primary care clinics, genetic clinics, or pathology services. Timing may include immediate action, action when convenient for patient, action at a specific timepoint (for example, within one month or within six months), or timing dependent on clinical indications. Several participants suggested that ideally, actions should be immediate; however, due to resources and staffing constraints, timing may largely depend on clinical indications.

Phase 1: pre-test

Phase 1 encompasses 10 actions for initial assessment, referral, and counselling for patients presenting with suspected genetic conditions. Actors involved in this phase include medical specialists and nurse specialists with genomic expertise, primary care clinicians, clinical geneticists, and genetic counsellors. Most actions are suggested to take place in both public and private health settings, genetic clinics, or specialist clinics. Urgency largely depends on clinical indications, with some actions suggested to require immediate action.

Participants cautioned that '*funding request*', while necessary for tests where public funding is not available, can be time-consuming, and can create a bottleneck that interferes with timely care, especially as genomic testing grows in volume. Additionally, some participants suggested directing more funding to treating clinicians or clinical services often responsible for securing test funding. For the '*selection of laboratory and tests*', participants stated guidance from a genetics team (i.e., geneticists and genetic counsellors) or a genetic testing stewardship team is crucial to ensure optimal test utility. Participants also recommended upskilling primary care clinicians, medical specialists, and nurses to expand their genomic expertise and scope of practice, to enable confident and appropriate patient management with additional support from genetic services as needed, reducing default referrals to geneticists.

Phase 2: testing

Phase 2 encompasses seven actions for testing and result reporting. Actors involved in this phase include the patient, nurse, and phlebotomist (for sample collection), laboratory geneticists, medical scientists, bioinformaticians, and the genomic multidisciplinary team. The actions are suggested to be situated within public and private health settings and pathology services. Some actions are non-urgent, while others depend on clinical indications.

For the action '*sample collection and storage*,' the actor (e.g., patient) depends on sample type (e.g., saliva). The action '*sequencing & automated bioinformatic pipeline triggering*' lacked consensus due to participants' unfamiliarity with laboratory processes, as most were clinical staff. One participant stated that laboratories may follow their own processes, provided these align with national accreditation requirements. The action '*sharing of raw test data upon request*' was proposed for exclusion because it was a research activity. Participants noted that while research is crucial in genomics, sharing patient data for research purposes should be case-specific, subject to patient consent, appropriate ethics approval, and clinician discretion.

Phase 3a: post-test care

This phase covers seven actions related to post-test follow-up of patients and their families, as clinically indicated. Similar to the pre-test phase, actors involved in this phase mainly include medical specialists, primary care clinicians, clinical geneticists, and genetic counsellors. Most actions are suggested to take place in either public and private health settings or genetic clinics. Urgency largely depends on clinical indications, with some actions suggested to require immediate action.

Some participants noted that geneticists may lack resources for long-term support and follow-up. They suggested that medical specialists and primary-care clinicians, with guidance from clinical geneticists, are better positioned to lead ongoing patient care and re-referral. Clinical indications for re-referral may include paediatric patients approaching adulthood, changes in patient phenotype, or reproductive planning.

Table 3. Actions and percentage agreements across the three survey rounds

Phase	Actions presented to participants in Round 1	Percentage for including in the model of care	Actions presented to participants in Round 2	Percentage for including in the model of care	Actions presented to participants in Round 3	Percentage for including in the model of care
Pre-test	Referral screening & triage	90.5%	Referral screening & triage	NA	Referral screening & triage	NA
Pre-test	Conducting assessment - taking history & pedigree, generating phenotypes & differential diagnoses	90.5%	Conducting assessment - taking history & pedigree, generating phenotypes & differential diagnoses	NA	Conducting assessment - taking history & pedigree, generating phenotypes & differential diagnoses	NA
Pre-test	Discussing eligibility & test selection	85.7%	Discussing eligibility & test selection	NA	Discussing eligibility & test selection (test's utility, benefits, risks, limitations, implications)	NA
Pre-test	Discussing test's utility, benefits, risks, limitations, implications	85.7%	Discussing test's utility, benefits, risks, limitations, implications	NA		
Testing	Returning of results to patient	85.7%	Returning of results to patient	NA	Returning of results to patient and entering results into EMR	NA
	Entering results into Electronic Medical Record	66.7%	Entering results into EMR	78.1%		
Post-test	Post-test counselling	85.7%	Post-test counselling	NA	Post-test counselling	NA
Post-test	Family referral	85.7%	Family referral (with consent)	NA	Family referral (with consent)	NA
Pre-test	Patient consent for genetic testing	81%	Patient consent for genetic testing, long-term storage, and future analysis	84%	Patient consent for genetic testing, long-term storage, and future analysis	NA
	Patient consent for storage of sample (short and long term)	71.4%				
	Patient consent for future analysis of data	76.2%				
Post-test	Genetic counselling for family	81%	Genetic counselling for family	NA	Genetic counselling for family	NA
Pre-test	Patient assessment	76.2%	Patient assessment	NA	Patient assessment for suspected genetics conditions	NA
Pre-test	Selection of test(s)	76.2%	Selection of test(s)	NA	Selection of test(s)	NA
Testing	Sample collection	76.2%	Sample collection and storage	75%	Sample collection and storage	NA
	Sample storage (short term)	57.1%				
	Sample storage (long term)	61.9%				
Testing	Curation & interpretation	76.2%	Curation & interpretation of variants	NA	Curation & interpretation of variants	NA
Testing	Returning of results to clinician	76.2%	Returning of results to clinician	NA	Returning of results to clinician	NA
Pre-test	Patient referral to genomic specialist	71.4%	Patient referral to genomic specialist	84%	Patient referral to genomic specialist	NA
Pre-test	Filling out test request form	71.4%	Filling out test request form (Ordering test)	78%	Ordering test	NA
Post-test	Re-refer to genomic specialist if indicated	71.4%	Re-refer to Genetic Health Queensland if indicated	71.9%	Re-refer to the appropriate genetics service (eg Genetic Health Queensland) following discharge, when clinically indicated.	80%
Post-test	Cascade testing for family members	71.4%	Cascade testing for family members	84.4%	Cascade testing for family members (if clinically indicated)	NA
Post-test	Returning of results to clinician	66.7%	Returning of results to clinician	78.1%	Returning of results to the ordering clinician (if new findings present)	NA
Post-test	Returning of results to	66.7%	Returning of results to	78.1%	Returning of results to	NA

Table 3 (continued)

	patient		patient		patient and entering results into EMR	
	Entering results into Electronic Medical Record	61.9%	Entering results into EMR	65.6%		
Post-test	Long-term support, management & surveillance	66.7%	Long-term support, management & surveillance	71.9%	Long-term support, management & surveillance (if clinically indicated eg increased risk of cancer)	68%
Pre-test	Selection of lab	61.9%	Selection of lab	71.9%	Selection of lab if applicable	60%
Post-test	Re-referral to genomics service	57.1%	Re-referral to Genetic Health Queensland	56.3%	Re-refer to the appropriate genetics service (eg Genetic Health Queensland) when clinically indicated	84%
Pre-test	Funding request for testing	57.1%	Funding request for testing	65.6%	Funding request for testing and reanalysis when applicable (eg not under Medicare)	60%
Post-test	Curation & interpretation	57.1%	Curation & interpretation of variants	62.5%	Curation & interpretation of variants	72%
Post-test	Funding request for re-analysis	57.1%	Funding request for re-analysis	46.9%		
Testing	Storage of test data	52.4%	Storage of test data and result reports	81.3%	Storage of test data and result reports	NA
Testing	Automated bioinformatic pipeline triggering	52.4%	Sequencing & automated bioinformatic pipeline triggering	65.6%	Sequencing & automated bioinformatic pipeline triggering	52%
Testing	Sharing of test data	52.4%	Sharing of raw test data	56.3%	Sharing of raw test data upon request (eg by researchers and clinicians), when appropriate (eg ethics approved) and if patient consents	48%
Post-test	Automated bioinformatic pipeline triggering	52.4%	Sequencing and automated bioinformatic pipeline triggering	46.9%		
Post-test	Routine medical examination	33.3%				

>75% consensus 50-75% consensus <50% consensus

Phase 3b: re-analysis

The re-analysis phase comprises six actions. It was initially included as part of long-term management but became a separate phase due to several complex issues included funding availability, timely consent, infrastructure, and patient loss to follow-up. Actors involved in this phase are similar to those in the Testing phase, including laboratory and clinical geneticists, medical scientists and specialists, genetic counsellors, and primary care clinicians. The settings include both public and private settings and pathology services. Urgency depends on clinical indications and workload.

Several participants expressed concerns about changes in patients' preferences and consent for re-analysis and follow-up due to test fatigue and anxiety from inconclusive results. One questioned the utility of re-analysing older data when newer technologies might yield better results. Overall, participants agreed that geneticists or medical specialists with genomics expertise should lead re-analysis discussions when appropriate and clinically indicated. These discussions should address consent, available funding, and implications of potential new findings. Notably, one participant emphasised that not re-analysing readily available genetic data would be wasteful.

Changes required to implement the model of care

Eighteen recommendations for required changes were generated from participants' Round 1 free-text responses. They were subsequently refined to 10 recommendations in Round 2 (see Fig. 3). Following Round 3 rankings based on urgency, the recommendations were categorised as immediate or near-term, medium-term, and long-term or not needed.

Building sustainability

Four recommendations (1, 2, 3, and 5 in Fig. 3) focused on building system sustainability for growing genetic

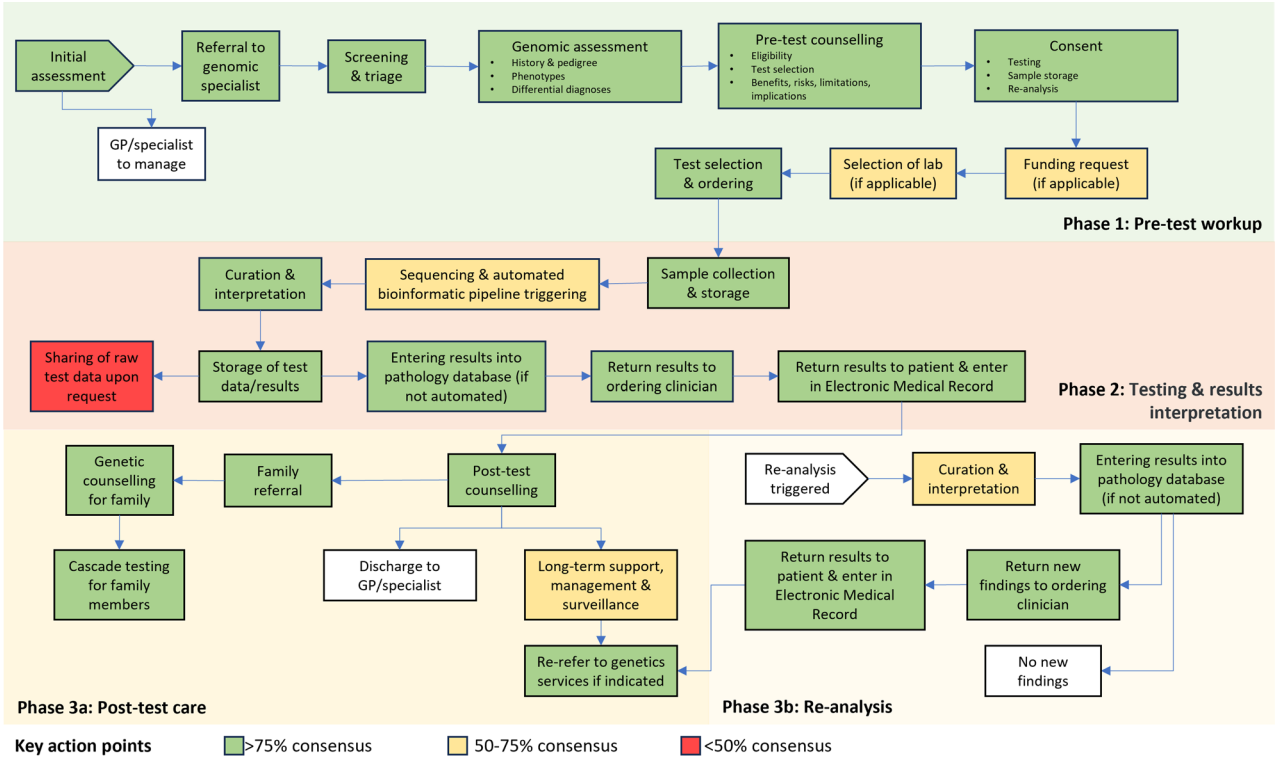

Fig. 2 Genomics model of care

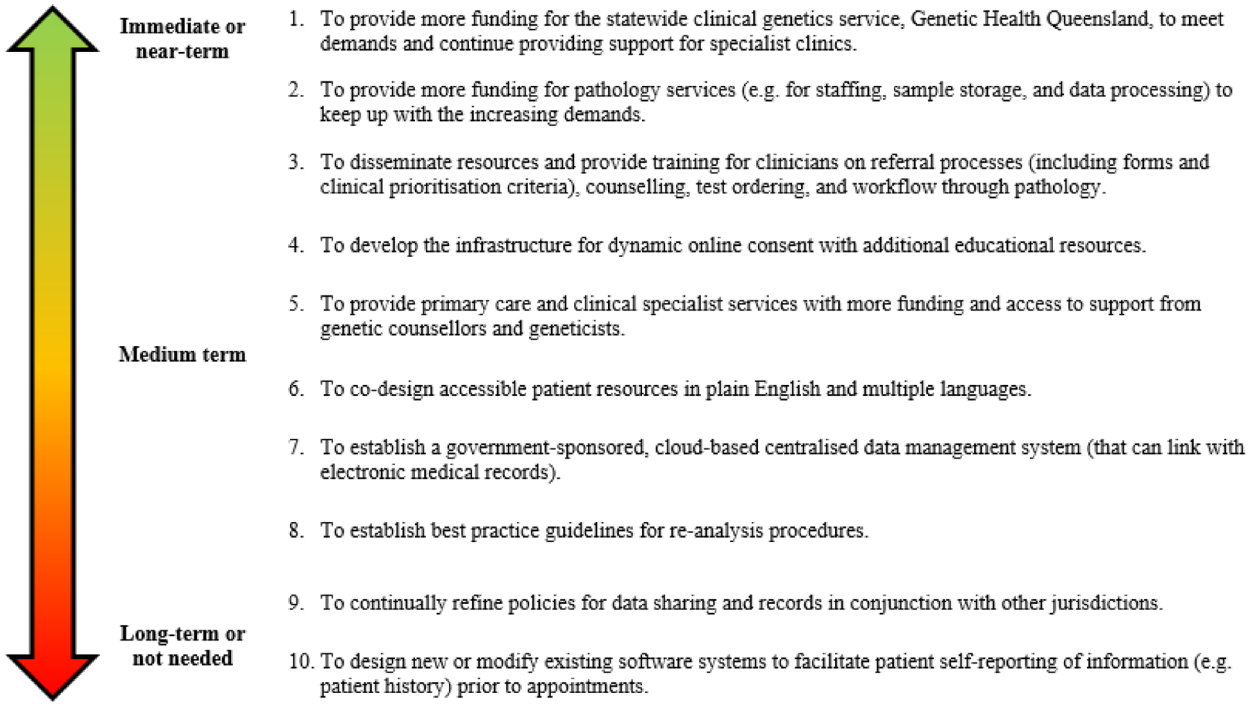

Fig. 3 Stakeholder-informed recommendations for integrating genomic medicine

testing demands across clinical specialties and services. All were deemed immediate/near-term. Participants suggested that additional funding for clinical services will help meet demands through adequate staffing, increased clinical volume, and reduced waitlists. It will also facilitate integrating genetic expertise into multidisciplinary teams to support medical specialists.

Participants further proposed that building sustainability also extends to expanding the workforce and enhancing pathology services' infrastructure and resources. They noted that as genomic medicine mainstreams, clinicians such as medical specialists, nurse specialists, and nurse practitioners may undertake responsibilities traditionally performed by geneticists or genetic counsellors. These tasks include pre- and post-test counselling, test selection, consenting, and results returning. Comprehensive upskilling programs and clinical resources (e.g., online training modules) are therefore essential to broaden their scopes of practice. Furthermore, pathology services require funding to expand staffing, enhance infrastructure, improve data processing capabilities, and increase variant curation capacity.

Building infrastructural capacity and innovation

Four recommendations (4, 6, 7, and 10) are associated with building infrastructural capacity and innovation, with priorities ranging from immediate/near-term to longer-term/not needed. Participants viewed these newer patient-facing technologies and co-designed resources as desirable but secondary to addressing increasing clinical demands. They also noted current infrastructure limitations for these technologies and potential accessibility barriers.

Many participants viewed a government-sponsored, cloud-based centralised data management system integrated with Electronic Medical Records as an ultimate goal, albeit one requiring adequate infrastructure. They suggested that when implemented, such a system could contribute to mainstreaming genomics by addressing information silos and preventing duplicate testing due to fragmented care. Notably, participants preferred contributing towards national data federation efforts to individual, uncoordinated approaches. In the interim, a statewide genetics database was suggested to address these issues.

Policy development

The final two recommendations (8 and 9 in Fig. 3) address data sharing and re-analysis guidelines and policies. Both are considered longer-term goals due to the competing clinical demands. While data sharing policies exist across states, they may require review for jurisdictional alignment. Re-analysis practice guidelines may need national consistency. Some participants suggested both recommendations could fall under Genomics Australia's governance, a national body established on 01 July 2025 to provide leadership and coordination to integrate genomics into the health system.

Centralisation and mainstreaming of genomic medicine

In response to the most immediate priority (i.e. recommendation 1) regarding the centralised statewide service (i.e. Genetic Health Queensland), diverse viewpoints were identified. Several supporters of centralisation argued that the hub-and-spoke model is more cost-effective, given the current low demands for genetic testing at primary care and specialist services. They contended that a central service could provide logistical support (e.g. cover for planned leave) and training for clinicians across these services, enhancing their genomic expertise and confidence to keep up with the growing demands.

Participants also highlighted that Genetic Health Queensland, as a 'well-established' (Participant B6) statewide service with 'appropriate clinical governance' (Participant C21) and infrastructure, is best positioned to deliver statewide education and training. This service supports the genetic testing stewardship program and the variant prioritisation and curation process in pathology services. A central service could reduce information siloing, minimise care fragmentation and test duplication, and provide more holistic and equitable care, for instance through telehealth.

In contrast, some opponents of centralisation argued that as genetic testing becomes mainstream, it will transition from a 'low-volume, high-cost healthcare tool' (Participant B4) appropriately managed by a statewide service to a 'higher-volume, lower-cost' (Participant B4) general healthcare tool better led by specialist services. They contended that patients can be managed appropriately in this distributed model if medical specialists are supported by genetic counsellors and/or geneticists. This approach could improve accessibility to genetic testing for more patients by eliminating the burden of multiple specialist appointments. Consequently, the central service's remit may shift to managing only highly complex cases.

Some participants suggested that primary care and specialist services should retain governance over seeking genomic support, including the ability to 'employ their own clinical geneticist' (Participant C4) or choose genetic services for referral. Participants expressed concerns that this autonomy might be limited under a centralised model. Consequently, it was proposed that funding and genetic support for these services should be delivered collaboratively.

Discussion

This study aimed to consolidate collective experiences and existing knowledge from local health services, statewide, and national genomics projects to establish a streamlined model of care for integrating and delivering genomic services in the Queensland public health system. The findings include a proposed model of care and 10 recommendations to guide the phased implementation of genomic services. Participants prioritised building sustainability, improving access to specialists through a distributed model, and investing in clinical-pathology partnerships and infrastructures. These recommendations can help address the information siloing and unify health systems. They align with system-level challenges identified in the literature, including data storage and sharing, infrastructure, skilled workforce availability, and disparities in patient access to genetic testing [30–33].

The centralisation of genomic medicine was contentious among participants due to concerns about access disparities. Genomic equity remains a focal point in health policies across many settings and health care systems [34]. A centralised model, like those in the Genomics England program [35], may concentrate expertise and be more cost-effective but potentially reduce available genetic services and limit care accessibility [4]. This is particularly relevant in Australian settings for groups such as First Nations peoples, where geographical remoteness, non-medical costs, and mistrust towards the healthcare system can impact access to care [34, 36].

Our findings indicate a delicate balance between service sustainability (addressing clinical demands and equitable care access) and service growth (adapting to the anticipated workload increase) as genomic medicine in Queensland moves towards a distributed (i.e. mainstreamed) model. The longstanding statewide Genetic Health Queensland and Pathology Queensland services are currently well-positioned to provide equitable patient access to care, given their established infrastructure, resources, and expertise. However, as genomic medicine integration progresses, primary care and specialist services will assume responsibility for some patient cases, while the management of complex patients would fall within the remit of these central services. Previous surveys of Australian non-genomic specialists showed strong support for a service model integrating clinical specialties with support from genetic specialists and counsellors pre- and post-test, or by referral [37]. This genetic support is crucial to maintain care standards as genomic medicine is mainstreamed [38].

Implementation science research has the potential to support the integration of genomic medicine via a structured approach. Implementation science research can assess system readiness for mainstreaming, co-design effective implementation strategies with key stakeholders, identify low-value care and re-allocating resources to key health services, and focus on patient workflow, accessibility, and equity [6]. Implementation science frameworks and expertise can address barriers and enablers, guide integration, and evaluate implementation [30]. To overcome the barriers, findings could be used to generate theory-informed strategies (e.g., identify and prepare champions) that can be evaluated for feasibility, acceptability, and fidelity [39].

To our knowledge, our study is the first to combine the AACTT framework with the modified Delphi method. The Delphi method's anonymity and iterative sense-checking allowed thorough exploration of diverse stakeholder perspectives on the topic. The AACTT framework as a process mapping tool has previously been used to successfully design a clinical workflow for electronic patient-reported outcome symptom monitoring and to describe implementation barriers and enablers to the delivery of genomic newborn screening [40, 41]. In our study, it is applied to develop a genomics model of care specifying behaviours across three phases.

By combining the AACTT framework with the modified Delphi method, we identified differences in stakeholder perspectives on who needs to do what, differently, when and where. This approach allowed us to identify gaps in the current system (e.g. workforce insufficiency) that may be targets for implementation interventions (e.g. staff training). Further, it unpacks the complexity of integrating genomic medicine into a complex adaptive healthcare system and helps reduce some of the inherent unpredictability [6]. Moreover, recommendations generated from 'bottom-up' local stakeholders through the iterative Delphi process might be more acceptable and adoptable than 'top-down' approaches [42].

The model of care was developed through consensus among multiple stakeholder groups, including clinical professionals, laboratory staff, and leadership and policymakers. The inclusion of laboratory-based stakeholders in the Delphi rounds and laboratory processes in the model of care highlights the importance of integrating clinical and laboratory aspects in mainstreaming genomic medicine. This integration is crucial for reducing resource waste due to suboptimal panel selection or repeated testing secondary to information siloing. It also prevents bottlenecks from inadequate pathology infrastructure.

However, the study has some limitations. The model of care and recommendations are specific to the Queensland public health context and might not be directly translatable to other settings. Nonetheless, the consensus-building process may be applicable elsewhere. Moreover, some components of the process (such as pre-test and post-test counselling) are common across many genomic programs, suggesting that the insights gained

may still have relevance beyond Queensland Health. The complexity of genomic medicine may not be fully captured in a simplified linear workflow. Stakeholders such as patients, regulatory and funding bodies, and ethics experts were not involved in the Delphi study, as the primary focus was on service providers. We consulted with consumers after each round to capture their perspectives. Future research should involve patients and funders to incorporate their views. The low response rate, while acceptable for a study of this type, may have contributed to nonresponse bias. Round 3 saw reduced participation from department heads and strategic decision-makers. A post-study report was subsequently distributed to participating health services and their executive members and staff, eliciting feedback that helped mitigate this issue. Participants' geographical regions (metro, regional, or rural) were not recorded but may have informed their responses on the model of care.

Conclusion

Using a modified Delphi methodology, we achieved stakeholder consensus on an effective and broadly applicable model of care for genomic medicine and developed stakeholder-informed recommendations to guide its phased integration. These findings have significant implications for implementing genomic medicine in public healthcare systems. They may inform a comprehensive, system-level implementation effort for mainstreaming genomic medicine by assessing system readiness, co-designing implementation strategies, and reallocating resources according to priorities. This stakeholder-driven approach ensures that the implementation of genomic medicine is practical, feasible, and acceptable to key stakeholders across the system.

Supplementary Information

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Supplementary Material 1.

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Data availability

All data generated or analysed during this study are included in this published article and its supplementary information file.

Declarations

Ethics approval and consent to participate

Ethics approval was granted by the Office of Research Ethics and Integrity at the University of Melbourne, Victoria, Australia (reference number: 2024-30792-60552-4). Informed consent was obtained from all participants as required.

Consent for publication

Not applicable.

Competing interests

The authors declare no competing interests.

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