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Levers and constraints on general practitioners offering and managing reproductive genetic carrier screening in Australian primary care: a rich graphic of the complex context

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Abstract

Reproductive genetic carrier screening (RGCS) identifies people who have an increased chance of having a child with a serious genetic condition. In Australia, since November 2022, RGCS for three conditions, cystic fibrosis, spinal muscular atrophy, and fragile X syndrome, has been publicly funded via Medicare (Australia's universal health insurance scheme). Therefore, tests are offered at no cost to eligible people and are accessible through general practitioners (GPs), making primary care a key point of access. This study aimed to develop a rich graphic illustrating the ecosystem of offering RGCS in Australian primary care. The three-stage, exploratory, qualitative approach involved: (1) constructing the first draft of the rich graphic using literature, policy documents, and team expertise; (2) conducting semi-structured interviews with eleven key informants representing diverse stakeholders; and (3) conducting a survey with key informants to review the penultimate version and produce the final version. The final rich graphic positioned GPs at the centre of a network of six stakeholder groups: consumers, government, laboratories, professional bodies, healthcare organisations, and consumer support groups. Inductive thematic analysis identified three overarching themes: (1) reinforcing patterns and feedback cycles; (2) structural misalignments and systemic tensions; and (3) emergent adaptations and system responses. Our analysis revealed that several factors influencing GPs' ability to offer RGCS are not isolated but parts of dynamic patterns of linkages, relationships, interactions, interdependencies, feedback loops, and behaviours that generate complex, sometimes unintended and contradictory, system-level effects, collectively shaping the complex system. Using rich graphic methodology revealed chains of interdependencies that would have remained obscured by using traditional linear implementation frameworks.

Keywords Reproductive genetic carrier screening, Rich graphic, Genetic Testing, Genomics, Complexity, Implementation science

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Introduction

Reproductive genetic carrier screening (RGCS) is a public health strategy, involving testing individuals and couples to determine the likelihood that they could have a child with a serious genetic condition [1]. Targeted RGCS has been available since the 1970s for populations with a high prevalence of specific genetic conditions, such as thalassaemia in communities of Mediterranean descent and Tay-Sachs disease in Ashkenazi Jewish communities [2]. Currently, there is considerable international variability in the design and delivery of RGCS programs. This variability can be explained by various factors, including differences in carrier frequency of genetic conditions, healthcare system structures, and financial, religious and cultural differences across populations [3]. While in most countries, RGCS is available through user-pays offerings, some have government-funded screening programs. In Israel, for example, the whole population is eligible for a three-gene carrier screening program. In the Netherlands, RGCS is offered on a fee-for-service basis to interested couples, with partial reimbursement provided by health insurance for high-risk groups, such as consanguineous couples [3].

In Australia, the Government funded $20 million Mackenzie's Mission—the Australian Reproductive Genetic Carrier Screening Project, which assessed the acceptability, feasibility and outcomes of a nationwide, couple-based genetic carrier screening program, provided free of charge to the participant [1]. The project was conducted between 2018 and 2022 and tested at least 1281 genes in 9,107 reproductive couples in preconception or early pregnancy [4]. Findings showed that 175 couples (1.9%) were identified as having an increased chance of having a child with a genetic condition for which they were screened. Three months after receiving the results, 76.6% of the couples with an increased chance had used or planned to use reproductive interventions to avoid having an affected child. The findings also showed that 98.9% of participants perceived screening as acceptable. Anxiety was higher among those with an increased chance result than among those with a low chance result and the median level of decisional regret was low across all result groups [4]. The delivery of screening to a diverse and geographically dispersed population was also feasible [5]. Following this project, the federal government expanded rebates provided under Medicare (Australia's universal health insurance scheme) to include RGCS for three genes causing cystic fibrosis, spinal muscular atrophy, and fragile X syndrome [6]. RGCS is now offered at no cost to those of reproductive age in preconception or early pregnancy, regardless of family history [7]. GPs in Australia are also eligible to order these tests, making primary care a key point of access for prospective parents [8]. Large panel screening (testing for hundreds of autosomal recessive and X-linked conditions) on a user-pays basis continues to be an option [9].

Evidence from Mackenzie's Mission showed that GPs have concerns about interpreting genetic screening results, offering referral pathways for increased chance results, and discussing the clinical and psychosocial implications of carrier identification for individuals and family members [10]. GPs receive limited support to deliver pre- and post-test counselling, as well as health promotion and education around RGCS [11]. Compounding these challenges, Medicare coverage of RGCS only funds the testing component, not the essential follow-up services such as genetic counselling. Australian clinical genetics services (including both clinical geneticists and genetic counsellors) are in high demand, with long waitlists and are under-resourced to manage the surge of increased chance results [12].

Additionally, the diverse range of stakeholders and organisations with multiple regulatory, advocacy and governance agendas and guidelines create a complex context for the practice of genomic medicine in Australia [13]. Lack of clarity of roles leads to fragmented interactions on multiple levels, with cascading effects across the landscape of RGCS delivery [14]. With these interwoven, interlinked challenges, the implementation of RGCS into routine healthcare requires a whole-of-system perspective [13, 15]. Understanding the ways in which interacting elements of the environment are interdependent, and how actions in one area inevitably shape the functioning of others elsewhere, is therefore essential [14].

Rich graphics inspired by the soft systems methodology of rich pictures have been used in healthcare to better understand the challenges and complexity of implementation and scale-up of interventions and healthcare delivery [13]. They are co-designed visual representations that provide complex maps to explore linkages, relationships, interactions, interdependencies, cascade effects, feedback loops, and behaviours [13, 16]. Rich graphics evolve through iterative data collection. They integrate qualitative data (e.g. interviews), documentary and policy sources (e.g., guidelines, reports, websites), and quantitative or administrative data (e.g., service activity, uptake figures), synthesising stakeholder perspectives depending on the purpose and setting [17]. Accordingly, this study aimed to develop a rich graphic through the collection and analysis of documentary sources and interviews with key informants to provide a holistic picture of the factors influencing GPs' ability to offer and manage RGCS in Australian primary care.

Methods

Study design and overview

This study employed a three-stage, exploratory, multimethod approach. An overview of the methods is

provided in Fig. 1. In "Stage 1" section, the initial graphic was drafted using team expertise and literature reviews. In "Stage 2" section, we conducted key informant interviews to iteratively refine the graphic. "Stage 3" section, the final stage, involved validation of the penultimate draft of the graphic and production of the final version.

Stage 1: Literature and document review

The first author (MV) drafted the initial iteration of the rich graphic using data from empirical and grey literature, as well as a review of relevant documents and websites, assisted by expertise held within the research team. This expertise included four clinicians; two of whom had clinical experience of RGCS, plus two authors with lived experience of genetic conditions and testing. The search was conducted in October 2024. Peer-reviewed literature was identified through searches in MEDLINE/PubMed using combinations of keywords, including "reproductive genetic carrier screening" OR "reproductive carrier screening" OR "carrier screening" OR "population screening" OR "genetic screening" OR "genetic testing" OR "preconception screening" OR "cystic fibrosis screening" OR "fragile x screening" OR "spinal muscular atrophy screening". Grey literature and documents were identified via targeted searches of websites of governments (e.g., NSW Health), professional and regulatory bodies (e.g., Royal Australian College of General Practitioners), clinics (e.g., Monash IVF), government documents (e.g., policies and reports), and a review of evidence-based literature on RGCS using the same keywords, with an emphasis on identifying factors influencing its implementation at multiple levels. The search was limited to the Australian context. Microsoft PowerPoint, version 2506, along with a combination of symbols, shapes and words, were used to draw the graphic, illustrating the system and how it operates. An accompanying narrative was also developed (and refined throughout the project) by MV, to articulate understanding of the ecosystem developed through the project.

Stage 2: Semi-structured interviews

Recognising the complexity of implementing RGCS during stage 1, we identified diverse key informants across the micro- (i.e., individual level, including perspectives of consumers and GPs) meso- (i.e., institutional level, including perspectives from peak bodies, managers at Health District level) and macro-systems level (Australian-State Government funding structures, genetic workforce, laboratory industry as a whole). Key informants were eligible to participate in the study if they (1) had a

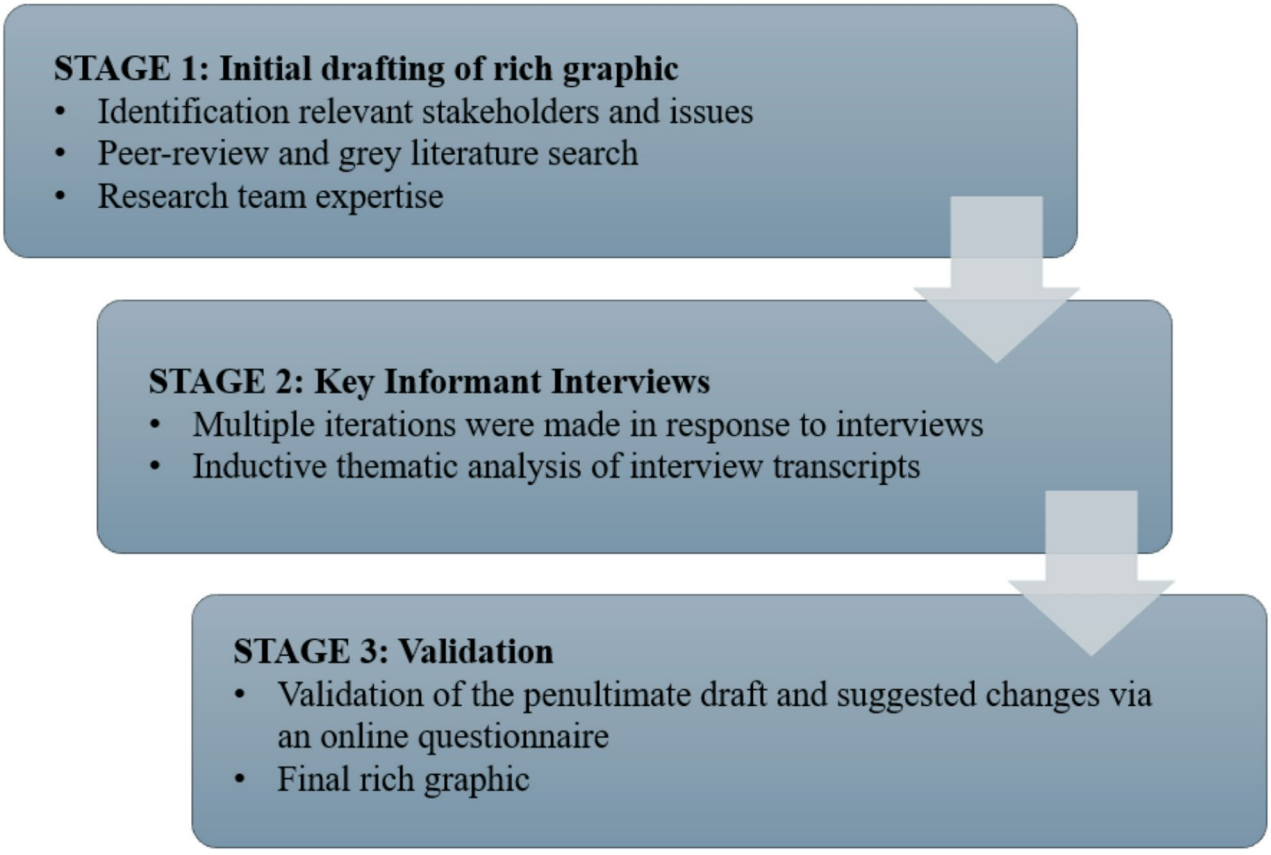

Fig. 1 Overview of the procedure used to develop and ratify a rich graphic of factors influencing offering RGCS by GPs

relevant professional role related to RGCS (e.g., health services, clinical care, laboratories, education, national level program management, policy, funding, or consumer decision-making), or had lived experience of RGCS; (2) were based in Australia; and (3) provided informed consent, including consent to audio-recording.

The first round of participants was drawn from investigators on Mackenzie's Mission research project, representing a range of backgrounds and geographic locations across Australia. Subsequent recruitment used snowball sampling based on interviewee recommendations. Our aim was to engage key informants with diverse viewpoints who were directly involved in or affected by the focal situation but brought different perspectives to the study that together would built a multifaceted picture [18]. Invitation emails, including detailed information about the study and participation information and consent forms, were sent to 40 potential key informants, inviting them to participate.

Online, semi-structured interviews were conducted by two authors (JL, MV). All participants were required to give written consent and return their consent form to the researchers before the interview. At the beginning of each interview, verbal consent was also sought. All interviews were conducted using Zoom, Version 6.5.11, which enabled video recording and automated transcript generation. Each interview started with the participants explaining their role and previous experience (if any) with RGCS. After explaining the aim of the interview and orienting the participant to the graphic, they were asked to "think out loud" as they explored the graphic. A series of prompts were used if needed. Specific questions were asked to assess whether the graphic accurately represented the current landscape of RGCS in Australia. These included:

1. "Do you think this is an accurate representation of the interaction of factors influencing GPs' ability to offer RGCS?"
2. "Have we missed any components or stakeholders?"
3. "Have we missed any connections, interactions or influences?"
4. "Do you wish to make any additions, subtractions, or changes?"

Participants were also asked how the graphic could be improved to be more accurate or nuanced. As interview data were collected, they were read through, and changes or interactions suggested by participants were incorporated into the rich graphic. The graphic, therefore, evolved through multiple changes. Each automatically generated interview transcript was subsequently reviewed and proofread for accuracy by MV. Prior to commencing any data analysis, all transcripts were de-identified to maintain participant confidentiality and originals deleted. Each interview lasted approximately 30–40 min.

Stage 3: Survey

Once all the interviews were completed in Stage 2, the penultimate version of the graphic was created by checking back over the interview transcripts to ensure all feedback from interviewees had been incorporated. Where participant opinions differed on a part of the graphic, MV and JL met to discuss how to accommodate the two views (i.e., showing both points of view on the graphic, acknowledging both in the commentary, or favouring one point of view over another). For any unresolved issues, the options were presented to the broader research team to reach an agreement.

Key informants who were interviewed in Stage 2 were re-contacted to ask them to review the penultimate version of the graphic accompanied by the narrative—the structured, linear account that linked each numbered bubble in the rich graphic to the corresponding stakeholder's roles, responsibilities, challenges, and facilitators (See Supplementary File 1). Participants were asked to view the graphic and read the narrative, then respond to questions via a REDCap survey hosted by Macquarie University. Survey questions included:

1. "Considering the whole graphic now, are there any other stakeholders, organisations, processes, or issues that you think need to be added?"
2. "Has the graphic given you any new insights about interconnections, dependencies, or relationships that you had not considered before?"

Sampling and data collection continued until thematic saturation was reached, defined as the point at which no new codes or themes were identified, and data began to repeat so that further data collection was redundant, signifying that an adequate sample size was reached [19]. In parallel, iterations continued back and forth until graphic saturation occurred, defined as the point at which new interviews and survey no longer generated new issues, insights, elements or relationships in the graphic. At this point, the participants indicated that the graphic was representative of the situation, realistic, and useful [20]. Data collection ceased when both thematic and graphic saturation had been achieved.

Data Analysis

Interviews, inductive thematic analysis, and graphic refinement proceeded concurrently and iteratively. All audio-recorded interviews were transcribed verbatim by MV. Transcripts were deidentified and checked for accuracy against the recorded audio. Inductive thematic

analysis was applied to identify, analyse, and report emerging themes and sub-themes within the data in relation to the research questions [21]. Initial coding was undertaken by MV, and then the data within each code were discussed, refined, and validated with JL, KL and KC. Themes and sub-themes were triangulated with quantitative data and empirical research findings through discussions between team members. Analyses were used iteratively to refine the rich graphic and accompanying narrative across successive versions. Data were managed in NVivo 12.

Results

Nine iterations of the rich graphic were generated. Iteration #1 is shown in Fig. 2, and the final version in Fig. 3. Eleven out of the 40 invited key informants agreed to participate in interviews. Table 1 presents the main characteristics of participants. All participants were from NSW and Victoria states.

All eleven participants also took part in the validation survey, providing feedback on various aspects of the graphic. In addition, one of the 40 invited key informants completed the survey only (KI 12) and did not participate in an interview. All feedback received in this stage was incorporated into the final graphic (Fig. 3).

Rich Graphic: Overview

Figure 3 depicts a rich graphic of the ecosystem for offering and managing RGCS in Australian primary care. It illustrates multiple agents across different system levels, both individuals and organisations, and a web of complex interactions and interdependencies. The final rich graphic is structured with GPs in the centre of the graphic, surrounded by six major stakeholder groups, including other health services, consumers, government and regulatory bodies, laboratories offering tests, professional bodies, and consumer support agencies.

Thematic examination

Data analysis identified three overarching themes that characterised this ecosystem: (1) Reinforcing patterns and feedback cycles; (2) Structural misalignments and systemic tensions; (3) Emergent adaptations and system responses (Table 2). Our analysis showed that factors influencing GPs' ability to offer and manage RGCS operate within interconnected patterns that create complex and sometimes unintended or contradictory outcomes. Table 2 provides supporting evidence, including details of subthemes and quotes.

Theme 1: Reinforcing patterns and feedback cycles

There was broad consistency across key informants, indicating that GPs are often under-prepared to receive increased chance results because these results trigger a sequence of demands—delivering results, providing counselling, and arranging referrals—that many GPs feel poorly equipped to manage. Referring patients to genetic counsellors imposes more pressure on the

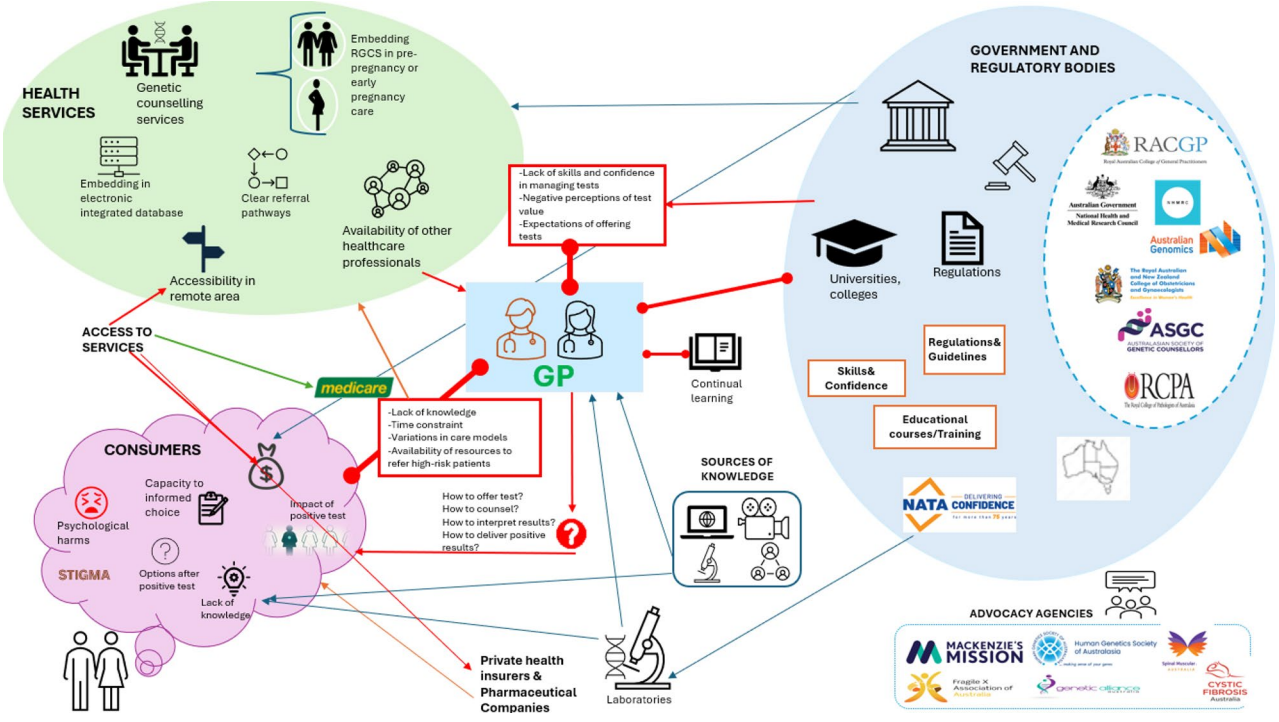

Fig. 2 First iteration of the rich graphic

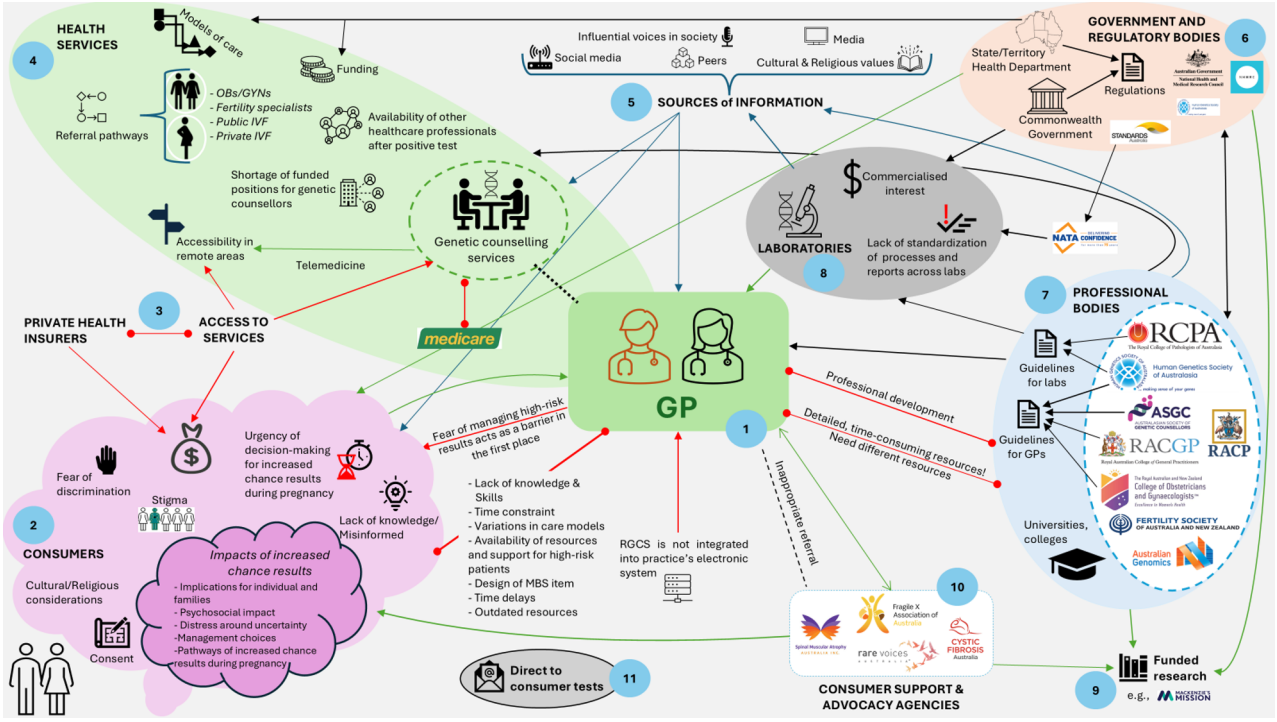

Fig. 3 Final iteration of the rich graphic illustrating factors influencing GPs ability in offering RGCS in Australian primary care. GPs in the middle of the graphic are surrounded by bubbles representing six major stakeholder groups. The placement of the stakeholders is pragmatic for the visual clarity of the graphic and does not imply a hierarchy. The text describes contextual influences. Lines indicate a relationship of some sort (interdependencies, interactions, relationships, sequential pathways, determinants, etc) between stakeholders or factors. Black lines indicate influence or dependencies (e.g., the regulatory body's influence on laboratories). Red lines indicate potential obstacles or barriers to high-quality RGCS management (e.g., impacts of increased-chance results), and green lines indicate facilitators (e.g., Medicare coverage of RGCS). Blue lines show the flow of information between several stakeholders (e.g. information provided by professional bodies). Darker lines represent stronger influences. Solid lines represent established or routine pathways, whereas dotted lines represent possible or emerging relationships. Numbered blue circles indicate elements that are further described in Supplementary File 1; the corresponding number refers to the relevant explanatory note

Table 1 Characteristics of key informants involved in interviews and feedback on rich picture

Key Informant ID	Primary role	Years of experience
KI 1	Genetic Counsellor	~ 20
KI 2	Consumer Support Agency member	> 20
KI 3	Manager Peak professional body	19
KI 4	Consumer with lived experience/Health Services researcher	9
KI 5	Consumer with lived experience/Bioethicist	8
KI 6	Genetic counsellor/Peak professional body	16
KI 7	Clinical geneticist	30
KI 8	Genetic Counsellor/Peak professional body	> 30
KI 9	Genetic counsellor	~ 20
KI 10	General Practitioner	22
KI 11	General Practitioner	7
KI 12	General Practitioner	19

already overstretched system, with long waitlists and limited resources. Interviewees highlighted that, without the Medicare rebate for genetic counselling follow-up, patients had to join public health service waiting lists, where they were typically not a priority, unless already pregnant or having another high-risk feature. This might shift patients to private services, leading to out-of-pocket costs for consumers, thus creating another barrier. Given these barriers, informants suggested some GPs come to perceive that timely, appropriate follow-up care is unlikely or difficult to secure for patients with increased chance results. They indicated that in anticipation of these downstream difficulties, some GPs hesitate to order RGCS—especially for patients who present in early pregnancy—to avoid complex time pressured scenarios they feel unable to manage. Informants reported that the more GPs encounter these situations without adequate support, the more constrained they feel in future encounters, perpetuating a cycle that affects both service delivery and patient experience (Numbered 4 in the rich graphic, Fig. 3).

Table 2 Thematic analysis of key informant interviews identifying the complex context in which GPs offer and manage RGCS in Australian primary care

Themes	Sub-themes	Example Quotations	Suggested interventions
Reinforcing patterns and feedback cycles	Increased chance results can trigger responses that GPs feel poorly equipped to manage	I think these challenges – increased chance results – might end up with a much more protracted relationship with a couple, including psychological harm. A lot of the patients are providing feedback that they would be sort of deeply shocked with an increased chance result, despite the counselling. All of these considerations are going to be exacerbated for a GP. [KI 7, Clinical geneticist]	Stronger support from genetic services providing real-time advice for GPs
	Uncertainty around follow-up pathways and fear of legal dispute	Sometimes GPs choose not to offer RGCS, because they are concerned that if there's an increased chance result, they won't be supported in understanding the pathways and information for their patients. They don't want to do this because they don't want to be sued if they make a mistake. [KI 2, Consumers' support agency]	Clear clinical pathways into genetic services support Easily accessible and credible information on pathways into genetic services and the responsibilities of GPs.
	Cascading consequences of increased chance results	This *[increased chance result]* can have personal health implications as well as reproductive health implications. It has implications for their marriage as well. Another barrier may be the potential for life and income protection insurers to discriminate against people with personal health implications which may deter GPs from offering or consumers from accessing RGCS. [KI 1, Genetic counsellor] GPs do not understand the significance of fragile X carrier finding in a woman. It has potential consequences not only for reproductive decision-making but also for the woman herself. [KI 2, Consumers' support agency]	Point-of-care information and support on specific increased chance results and implications for carriers Having access to high quality consumer-facing resources Decision support for consumers considering RGCS
	Managing incidental findings	The consent that goes with testing can become complicated. For example, a woman undergoing RGCS test discovered a faulty gene for her heart. So, are we doing a good enough job of that in the consent space? [KI 6, Peak professional body]	Skills training or guidance for GPs Decision support and clearly explained expectations for consumers considering RGCS
	Gaps in service delivery because of under-resourced external services	The Medicare item number for RGCS is great, but it does not include genetic counsellors. As a result, when test results indicate an increased chance, GPs must refer patients to genetic counselling services. Due to the lack of coverage and the shortage of funded positions, this often leads to long waitlists and delays in patient care. [KI 3, Peak professional body]	Redesign of Medicare Item number to include genetic counselling Increase funded genetic counsellor positions Consider more telehealth options to increase access to genetic services Consumer-facing resources to provide basic information and alleviate some of the anxiety while waiting for an appointment
	Connected systems support information flow	Having information about the tests readily available in the different HealthPathways *[point of care database of clinical information]* will help GPs to quickly access them. [KI 10, GP]	Point of care information for GPs
Structural misalignments and systemic tensions	Navigating unclear boundaries of their own role and knowledge	As a GP, I think the complexity for me is a big thing in terms of how to bring this up in conversation, and then how much knowledge I have to be able to introduce it. [K11, GP]	Skills training or guidance for GPs Decision support for consumers considering RGCS
	Lack of appropriate point-of-care support for GPs	We have been playing a role in the GPs' education space that really should have been played by the colleges. Not our job to educate GPs about carrier screening, but we do it because we were part of the advocacy, education events around the country. [KI2, Consumers' support agency]	Providing accountable education and support to GPs around RGCS
	Fragmented community-based service provision	A big hurdle is that our genetic counsellors are predominantly based in tertiary hospitals, not in the community setting where GPs are. A lot of the GPs would be referring to their tertiary clinical services, and non-pregnant people are not going to be prioritised on that wait list. They're going to be in your category, which is ideally a 12-month wait at best, but quite often is longer. So, we need more genetic services in the community. [KI 8, Peak professional body]	Increase funded genetic counsellor positions in community Consider more telehealth options to increase access to genetic services Consumer-facing resources to provide basic information and alleviate some of the anxiety while waiting for an appointment
	Inconsistencies in practices driven by the lab's commercial interests	Labs have been developing their own large panel screens with different genes, reporting and information about their utility. At least partly driven by commercial interests and a need to differentiate their product in a crowded market. [KI 8, Consumers' support group]	Guidelines for GPs, pathology labs, obstetricians and genetic professionals should be aligned and consistent, with clearly delineated roles and responsibilities

Table 2 (continued)

Themes	Sub-themes	Example Quotations	Suggested interventions
	Guidelines are not standardised	I hope that the standardisation is improved with the guidelines around offering RGCS tests. Now, labs have their own guidelines, and separate guidelines exist for GPs. We need consistency in messaging across multiple stakeholder groups. [KI 8, Peak professional body]	Guidelines for GPs, pathology labs, obstetricians and genetic professionals should be aligned and consistent, with clearly delineated roles and responsibilities
	Navigating ethical dilemmas in the offer of RGCS	You can be discriminated against for doing it *[offering RGCS to patients]* and discriminated against for not doing it. [KI 7, Clinical geneticist]	Promote clear guidelines and supportive policies that protect healthcare providers and patients from discrimination related to RGCS
Emergent adaptations and system responses	Private labs support GPs to improve service delivery	Some of the private laboratories have in-house genetic counselling services, and that's actually quite a good model in terms of providing that support. [KI 6, Peak professional body]	Consider setting standards for genetic counselling services in private labs to be comparable to genetic clinical services Redesign Medicare item number to include genetic counselling services
	Collaboration between different agents – Communication pathways	We *[genetic professionals]* sometimes get calls from GPs about whether it's appropriate to refer their patient or what testing is appropriate for their patient. [KI 1, Genetic counsellor] I would be very much relying on genetic counselling specialists. [K11, GP]	Increase funded genetic counsellor positions Stronger support from genetic services providing real-time advice for GPs Redesign Medicare item number to include genetic counselling services
	Emergent community advocates	Having some champions like Rachel *[consumer advocate who lobbied for introduction of RGCS]* who promote good quality education is probably really important. [KI 6, Peak professional body]	Increase funded genetic counsellor positions in the community Consider more telehealth options to increase access to genetic services Consumer-facing resources to provide basic information and alleviate some of the anxiety while waiting for an appointment
	The public's expectations are changing	As genomics becomes more mainstream, there are some larger social implications. I think it's likely enough that the public's expectations around it become more realistic. As the public knows about how uncertain it is, and if it was to become more mainstream, knowledge of its uncertainty would become more common, which is in some sense not a bad thing, but also in another sense opens up room to trust it less, or and I guess to misinform. [KI 5, Consumer/Bioethicist]	Provide high-quality information from credible sources across multiple public media outlets
	Evolving public awareness pathways	Having some champions on social media who promote good quality education is really important. [KI 8, Peak professional body]	Consistent messaging from a credible source across many types of media drives further discussion and educates the public
	Proactive efforts are needed to acquire knowledge	General practice is quite an isolating business. They're all their own businesses; they all work in their own ways. You have to be pretty proactive to get it *[knowledge]* out there to general practice. If GPs are interested in it and want to look it up. There's plenty of information out there for them to find, I think, really, and they're perfectly capable of finding it. [KI 11, GP]	Point of care information for GPs Easy to find genetic education courses and skills training for GPs approved by peak bodies
	Emergent models of care to support patients and GPs	Some of the laboratories have their own genetic counselling services… others have separate providers they work really closely with. [KI 1, Genetic counsellor]	Consider setting standards for genetic counselling services in private labs to be comparable to genetic clinical services Redesign the Medicare item number to include genetic counselling services

Survey participants reviewing the final rich graphic confirmed that perceptions around managing increased chance results were a primary driver of a self-reinforcing cycle that shaped GPs' engagement with RGCS. The broader multiple impacts of increased chance results were also noted by interviewees, including implications for couples' relationships and concerns about potential discrimination by life and income protection insurers. These issues were seen as compounding system-related factors in GPs' hesitance to offer RGCS to avoid unintended consequences.

Interviewees also highlighted the complex dynamics underpinning the role of consumer support and advocacy agencies in raising awareness of RGCS among both GPs and consumers. Heightened awareness might stimulate consumer demand for testing, prompting more patients to request tests from their GP and creating pressure for GPs to order tests despite limited training. Consumer

support and advocacy informants, in turn, reported receiving calls from GPs and patients asking for help in interpreting RGCS results when a carrier was identified, stretching their organisations' already limited resources with requests outside their usual responsibilities. This dynamic between GPs and consumer support agencies is thus featured in the rich graphic as a key element in the feedback loop of systemic pressures on RGCS delivery (Numbered 10 in the rich graphic, Fig. 3).

Theme 2: Structural misalignments and systemic tensions

Interviewees noted the positive outcome of the Medicare rebate for RGCS in removing a key financial barrier and expanding consumers' access to screening. Nonetheless, many observed that the unintended consequence of covering testing, but not post-test genetic counselling, was to expose and intensify existing capacity constraints within genetic services, particularly in referral and counselling pathways. Survey participants agreed with the rich picture representation of the Medicare rebate as a structural intervention driving both opportunities and constraints.

System misalignments were also highlighted by key informants in their descriptions of inconsistent, non-standardised processes, report formats and terminology across different laboratories (Numbered 8 in the rich graphic, Fig. 3). These inconsistencies were reported to cause uncertainty for GPs when interpreting results in the absence of consistent standards, contributing to fragmentation and variability in the delivery of RGCS across the system. Several participants also raised issues of persistent misalignment between the usability of support structures and resources available to GPs and the practical demands of point-of-care decision-making. Existing resources, for example, education modules, Clinical Practice Guidelines or Standards, were described as overly detailed, time-consuming to work through, and outdated, requiring GPs to invest extra effort in identifying and accessing appropriate resources. Participants noted that in the absence of coordinated oversight to train GPs, some consumer advocacy organisations had stepped in to fill this gap by running educational events to raise awareness of RGCS among GPs. These ad-hoc practices, while well-intentioned, were regarded as neither sustainable nor appropriate substitutes for systematic, professionally-led support structures. Similarly, interviewees noted that pathology laboratories often provide guidelines and educational resources for both consumers and GPs, but conflicting guidance offered by different companies leads to systemic tensions.

Lack of foundational education and adequate professional development was also highlighted as a structural misalignment between workforce preparation and practice demands. GPs reported not receiving sufficient training because RGCS had so recently been introduced in primary care, an issue that was especially relevant for GPs who had graduated many years ago with no formal education and training on genomics. Genomics is a fast-moving field, and screening is a tricky practice as it gives high or low chance, not definitive results. A GP participant noted a lack of training about how to introduce RGCS into conversations with consumers from culturally and linguistically diverse (CALD) backgrounds. Survey participants confirmed the rich picture view of insufficient training as a system-level misalignment, with several commenting that providing more training is not a solution if GPs still lack time to complete training (service pressures), laboratory reporting remains non-standardised, point-of-care support is unavailable (training cannot cover every scenario), and genetic counselling remains unfunded (GPs perceive an inadequate support system).

Theme 3: Emergent adaptations and system responses

Key informants described how complex interactions among components of the system in which RGCS is offered lead to adaptive strategies to cope with or manage complexity by system agents. One commonly reported adaptation was the establishment of genetic counsellors in pathology laboratories to interpret complex genetic results and communicate findings to GPs and patients. Informants noted that in-house genetic counsellors act as a liaison between GPs and the laboratory to ensure appropriate testing, interpretation, and communication of results. Survey participants supported the rich picture view of this adaptation as an emergent means of bridging the gap between existing genetic service delivery models and the increasing need for genetic services, noting the fragility of these external resources and the unpredictability of emergent adaptations when these supports were unavailable or overstretched.

Participants also observed that heightened public awareness of RGCS, driven by social media and consumer advocacy, increases demand for RGCS, intensifying the pressure on GPs to adapt to the use of the most accessible resources and support. In the absence of coordinated care pathways, GPs navigated to readily available informational materials produced by laboratories, although some participants also raised concerns about the commercial interests of laboratories framing these resources and support. Additionally, this increased demand was said to prompt greater use of evidence-based, web-based knowledge portals, such as HealthPathways [22], an online platform providing GPs with locally tailored guidance on the assessment, management and referral of a range of conditions, including RGCS. HealthPathways was described as facilitating information flow between multiple system actors, leading to more confidence in offering RGCS and supporting the emergence of a learning health system.

Discussion

This study presents the first rich graphic of RGCS dynamics in Australian primary care and offers novel insights into the system, identifying factors influencing the offering and management of RGCS by GPs. The rich graphic illuminates how barriers and facilitators to the delivery of RGCS do not operate in isolation. Their interaction stems from and generates reinforcing patterns and cascading effects, structural tensions, and emergent responses that shape the implementation of RGCS. Using rich graphic methodology enabled the complex system, relationships, dynamics, and associated factors to be seen as a whole.

A key dynamic highlighted in the rich graphic relates to funding for RGCS, which is currently implemented as standalone Medicare Benefits Schedule pathology items rather than being part of a coordinated screening program with a formal structure, such as National Cervical Screening Program with systematic invitations, reminders, and follow-up coordination [23]. Ideally, a screening program would include all necessary elements for delivery, including patient and provider education, adequate clinical support, access to genetic counselling services and patients' follow-up [5]. Nonetheless, there are no provisions in the Medicare rebate for pre- or post-test counselling, highlighting a system-level misalignment.

While GPs are capable of adequately providing pretest counselling for RGCS, there are implicit complexities in counselling for RGCS that make it a potentially time-consuming and resource-intensive process, another likely contributor to RGCS hesitancy among GPs [24]. Providing post-test counselling for couples in the event of an increased chance result will often result in an even higher level of complexity for numerous reasons. These include the implications and potential variability of the condition itself and the reproductive options available; the potential ramifications for family members and a need for a range of specialists, including genetic counselling services to be involved. However, there is neither a Medicare rebate nor sufficient genetic counsellor capacity in Australia to meet increasing demand [25]. Without a coordinated program structure, the system is highly dependent on the interplay between various actors, such as laboratories, which is likely to result in uneven implementation and variable patient experiences.

Several participants raised cultural and religious considerations (as evident in the rich graphic), including the need for greater support for GPs to introduce RGCS to people from CALD and religious backgrounds. GP hesitation to discuss RGCS with people from CALD communities is a healthcare delivery issue highly relevant to the Australian context and vividly evident in the rich graphic. In our study, and as reported elsewhere, this concern stems from several intertwined factors. Explaining genomics can be complex in any language and medical or genetic terminology often does not translate easily across languages [12]. Additionally, other research suggests that GPs are concerned about patient receptiveness due to their religious and ethical values, cultural beliefs and sensitivities [26]. While over 300 languages are spoken in Australian households [27], this diversity was not adequately considered in the Medicare rollout of RGCS with only limited translated or culturally tailored educational resources, highlighting a structural misalignment. The lack of resources reinforces GPs' hesitation to offer RGCS for CALD communities, leading to unmet need and inequitable access amongst different population groups [28]. As a means of addressing this inequity, it has been suggested that at least part of the subsidy to pathology services when RGCS is ordered could be distributed to develop centralised practitioner and patient support [11].

Genetic and genomic testing is inherently complex and requires extensive training to provide appropriate guidance and recommendations [29]. As noted in our findings and represented in the rich graphic, this complexity—combined with the rapid growth in test utilisation and the limited expertise in genetics among the non-genetic health workforce—has led to an emerging system-level response, the introduction of in-house laboratory genetic counsellors in some laboratories [30]. This emergent solution to the lack of resources can be a support for GPs in offering and particularly managing increased chance results; however, this service is only available to patients tested within the associated laboratory not accessible to the public.

Community-based genetic counsellors could also play a key role in supporting GPs in navigating RGCS, but there is currently no funding model to enable this [31]. Given GPs' perception of their own ability to offer RGCS appears to be contingent on the availability of expert genetic counselling and clinical support, increasing funded positions and including genetic counselling in the existing Medicare item is essential [32]. Expanding these services not only ensures that individuals and families receive consistent, accurate information and timely support, but also supports GPs in offering, and particularly, managing increased chance results [25]. The sustainability of the genetic counselling workforce is a major consideration that needs to be addressed by policymakers and health services to support embedding of RGCS in primary care [5].

Implications for policy and practice

The rich graphic identifies levers that could support more consistent and equitable RGCS delivery in primary care, including moving beyond standalone pathology items towards a more coordinated program approach, funding and scaling access to genetic counselling and follow-up,

and standardising laboratory reporting and terminology. It also highlights implementation priorities for primary care, such as accessible point-of-care tools and support for GPs and appropriate resources for the CALD population to address equity gaps. These outputs can be used in stakeholder co-design to prioritise feasible actions to improve the equitable implementation of RGCS in primary care.

Limitations and Strengths

A static, two-dimensional rich picture has limits to what it can depict. Drawing all interactions between all agents was not possible as it would result in a dense, busy graphic, making interpretation difficult. The same challenge applies to feedback loops, which were noted between multiple components of the graphic. Furthermore, the graphic may not capture all perspectives on RGCS implementation in Australian primary care, and additional viewpoints (e.g. from under-represented regions, service types, or professional roles) could identify further nuances or alternative linkages. The rich graphic depicting the RGCS within the Australian primary care context identifies agents and interrelationships that may be useful for international audiences and contexts. In addition, other research groups can create similar rich graphics using the thoroughly outlined methodology mentioned in this manuscript to capture the complexity in similar healthcare settings. Nevertheless, by integrating diverse data sources and a robust validation process involving genomics care experts, this approach provides a powerful investigative framework for various care contexts and settings.

Conclusion

Using the rich graphic methodology, we were able to analyse and understand the dynamic and interdependent nature of offering RGCS and display these complex findings, summarising the relationships, dynamics and associated factors within one diagram. This study showed that GPs' behaviours in offering RGCS are emerging from interactions with consumers, consumer support and advocacy agencies, laboratories, genetic services, policy and professional bodies. These multiple independent actors interact with strong interdependency but without central coordination. Our findings revealed that fragmented, outdated resources reflect independent stakeholder development; inconsistent guidelines arise from multiple standard-setters without coordination; insufficient skills reflect a lack of pre-genomics curricula and time-constrained practice structures; and unclear pathways stem from misalignment between service delivery and federal funding. Effective implementation of RGCS in this complex system requires Medicare coverage for pre- and post-test counselling and genetic counselling services, coordinated guideline development, aligning incentives, resources, and information flows across all parts of the system.

Supplementary Information

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Supplementary Material 1.

Clinical trial number

Not applicable.

Authors' contributions

JL conceived the study and obtained the ethics approval. MV designed and built the graphics. KL critically reviewed the manuscript. RP organised the key informants' recruitment. JL and MV conducted the interviews. SS, MC commented on all versions of the graphic. JL, MV, KL, KC, LE analysed findings against the rich picture complexity overview. MV drafted the initial version of the manuscript. All authors reviewed and approved the final version of manuscript.

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Data availability

The datasets generated and/or analysed during the current study are not publicly available, as all data are included in the manuscript body.

Declarations

Ethics approval and consent to participate

This study received approval from Macquarie University Human Research Ethics Committee (Approval number: 520241746957819). All participants were required to give written consent and return their consent form to the researchers before the interview. At the beginning of each interview, verbal consent was also sought.

Consent for publication

Not applicable.

Competing interests

The authors declare no competing interests.

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