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Views of reproductive genetic carrier screening participants regarding screening for genes associated with non-syndromic hearing loss

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Abstract

Objective: Reproductive genetic carrier screening (RGCS) panels often include genes associated with non-syndromic hearing loss (NSHL) despite a lack of evidence of acceptability. Although some couples take steps to avoid having a child who is deaf, there are effective interventions for children who are deaf. There is no consensus whether deafness is considered a disabling condition.

Method: This study explored views of people who had RGCS, without genes for NSHL, about this topic. Online surveys were sent to 2186 people who had a low chance RGCS result and 655 completed the survey (participation rate 30%).

Results: Sixty-three percent ($N = 412$) think deafness is a serious health condition. The majority agreed (60%, $N = 391$) that with support (i.e. hearing aids/cochlear implants) deafness is a minor condition in children. Most (84%, $N = 545$) agreed genes for NSHL should be included in RGCS. Thirty-five percent ($N = 231$) indicated they would make different reproductive decisions if they had an increased chance of having a child born deaf; 31% would not change their reproductive plans and 34% were unsure what they would do.

Conclusion: While the majority support inclusion of genes associated with NSHL in RGCS, there was uncertainty about the severity of deafness as a health condition and there was no consensus on whether it is a health condition that warrants changing reproductive decisions.

Key points

What is already known about this topic?

- Genes for non-syndromic hearing loss are already included in many commercial expanded carrier screening panels, yet little is known on the acceptability or utility of their inclusion. Whether deafness is a disability is continually discussed in the literature and research so far has identified mixed views on the utility of genetic testing for deafness in the reproductive setting.

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What does this study add?

- Individuals who value the results of expanded carrier screening generally support inclusion of genes for non-syndromic hearing loss in population screening, but with hesitation regarding support for termination of pregnancy as a reproductive decision for deafness. This group of screen-interested consumers are also ambivalent whether knowing their chances of having a child born deaf would change their reproductive decision making.

1 | INTRODUCTION

Advances in genomic technology has allowed reproductive genetic carrier screening (RGCS) to expand rapidly and there are currently several studies investigating the implementation of this type of screening at a population level.[1–3] Prospective parents may consider undergoing RGCS without personal experience of or knowledge about the conditions included in the screening.[4]

RGCS has generally been accepted for severe childhood onset conditions, but there is ongoing discussion about the inclusion of treatable conditions and of 'milder' conditions; and, more broadly, what constitutes a 'milder' condition. Some studies have reported that members of the public may wish to have screening for adult onset and/or mild conditions[5] while others highlight the difficulty in deciding what defines a genetic condition 'serious' enough to include in RGCS.[6–9] There are additional considerations if RGCS is to be offered as a population-wide publicly funded program, including the potential that inclusion of a condition in screening may represent a signal to couples about the severity of that condition, or a judgement about the appropriate action to take in response to information about carrier status.[10]

With respect to genes associated with non-syndromic hearing loss (NSHL), many commercially available expanded carrier screening panels include these genes despite continuing debate about whether deafness should be considered a disability.[11–13] Many authors discuss whether deafness is a disability, or alternatively whether being Deaf is more akin to being a member of a culturally and linguistically diverse group.[11–13] Genes associated with NSHL are included for screening in the recent American College Medical Genetics practice resource, which based gene selection on carrier frequency (>1 in 200) for autosomal recessive conditions that are at least moderate in severity.[14] Several groups have attempted to classify disease severity for conditions included in RGCS and have placed NSHL in the 'moderate' classification on a scale of profound/severe/moderate/mild.[15–17] A majority of the published and publicly available commercial carrier screening panels include at least one NSHL gene (*GJB2*).[3] In a recent Australian RGCS pilot study, genes associated with NSHL were omitted from the gene panel following extended discussion which concluded they were not sufficiently disabling to meet the criteria (Kirk et al., 2021). The criteria for inclusion of genes were that the condition should be life-limiting or disabling, with childhood onset, such that the average couple would be likely to take steps to avoid having an affected child; and/or be a condition for which early diagnosis and intervention would substantially change outcome.

One study[18] found that whilst 65% of participants who had a child with NSHL support inclusion of NSHL-linked genes in reproductive carrier screening, only 32% indicated they would use reproductive options to avoid having another child with NSHL. This is significantly lower than is reported for parents of children with more severe conditions, particularly those that may be lethal in childhood. A recent study reported that parents of children with a mild/moderate condition showed less support for inclusion of their child's condition in RGCS than those with more severe conditions.[19] A 2019 study looking at potential uses of non-invasive prenatal screening found that only 5.2% of participants indicated they would terminate a pregnancy for deafness.[20]

In the context of reproductive decision making by couples, the concept of disease severity can mean very different things to different people.[8,9] Disease severity has been found to be a dominant theme in guiding reproductive options following carrier screening: couples at increased chance of having a child affected by a condition classified as moderate are less likely than those affected by severe conditions to try to avoid having a child with that condition.[21,22]

Deafness is not always seen as an unfortunate or limiting condition by hearing people who have some knowledge of it (including children of deaf adults and parents of a deaf child), with some arguing that allowing the use of genetic testing for deafness in the reproductive setting expresses a negative view of deafness.[23–25] There are effective interventions and assistive devices for children with NSHL, including cochlear implants and hearing aids.

There is currently no consistent, evidence-based policy framework on whether deafness should be included in population-wide carrier screening.

In designing population health programs, it is important to include consumer input to ensure that health care services best meet the needs of clients.[26] In response, this study analysed how a carrier screen-accepting group of prospective parents perceives the inclusion of genes associated with NSHL in a population-wide RCS program. These views are contextualised within the healthcare setting in which they are imagined.

2 | METHODS

The data for this study was collected as part of a larger study exploring the feasibility of a population wide RGCS program. This project, the Australian Reproductive Genetic Carrier Screening Project, also known as Mackenzie's Mission, is funded by the Australian Government's Medical Research Future Fund as part of its

Genomics Health Futures Mission. The analysis presented here is from a survey distributed to a sub cohort of couples 3 months after the receipt of a low chance result. This study was approved by the Royal Children's Hospital Human Research Ethics Committee (HREC/53433/RCHM-2019).

2.1 | Study design

A quantitative questionnaire-based study was undertaken to ascertain the views of participants who had undergone expanded carrier screening for a panel of conditions which did not include genes associated with NSHL (the panel of conditions screened has been reported in an earlier paper by this group).[3] The analysis presented here is from attitudinal questions regarding deafness and whether genes associated with NSHL should be included in a population wide RGCS program. An online questionnaire was used for data collection and descriptive statistics were used to present the views of participants.

2.2 | Survey instrument

A core set of questions was developed, informed by the literature and the study team's experience. As participants had already participated in an expanded RGCS study, this set of questions were designed to explore attitudes to a hypothetical future screening program. Participants were asked: (i) whether genes for NSHL should be included in RGCS; (ii) whether they would choose to have RGCS for NSHL; and (iii) their views on whether they would use reproductive options (prenatal diagnosis 'PND', pre-implantation genetic testing - Mendelian 'PGT-M', or donor gametes) to avoid having a child with NSHL. Responses used a 5-point Likert scale, from strongly disagree to strongly agree, to assess respondents' feelings and attitudes (see Supplementary S1 for full questionnaire). The questionnaire was reviewed by clinical geneticists, genetic counsellors and a bioethicist, and also by potential participants meeting eligibility criteria of Mackenzie's Mission.

2.3 | Recruitment

Survey data collection took place between February and May 2021. Participants were included in the study if they were over the age of 18 and planning a pregnancy or in early pregnancy and had received a low chance carrier screening result from Mackenzie's Mission. As Mackenzie's Mission is a couple carrier screening research study, all participants had a partner at the time of enrolment and were either under 12 weeks pregnant or planning a pregnancy. Both members of the couple were invited separately to participate in the survey. All participants in Mackenzie's Mission were invited to participate in the study, and this may have introduced some bias if both the male and female members of a couple completed the survey and shared similar views.

2.4 | Data analysis

Cleaning and analyses of survey data collected via REDCap[27] were performed using IBM SPSS Statistics 27 for Windows. Descriptive analyses were used to describe characteristics and generate frequency data. Partially complete questionnaires were included in the data analysis, hence the number of participants that completed each question varies throughout.

3 | RESULTS

Invitations were sent to 2186 eligible participants and 655 completed the questionnaire, giving a completion rate of 30%. Of these, 71% identified as female (N = 463), all of whom were of reproductive age (defined by the World Health Organisation as being between 15 and 49 years). The majority of respondents were of European (69%, N = 454) ancestry and were highly educated with 79% (N = 522) having a bachelor's degree or above. Demographics of respondents are given in Table 1.

Respondents were asked about several factors that may influence their views towards carrier screening. A little over half of the respondents (57%, N = 373) reported they did not have an affinity with a religion. This was slightly higher than the recent Australian census data that reported 46.5% of 24–39-year-olds do not have an affinity with a religion.[28] A majority of the respondents (61%, N = 402) said religion does not influence the decisions they make in their life at all with 32% (N = 212) indicating it somewhat or moderately influences decision-making and 7% (N = 41) indicating religion completely or very much influences their decision-making.

Participants were also asked about their familiarity with deafness, in case this influenced views about carrier screening. One hundred and thirty-nine participants know someone who is deaf (21%) and nearly half (N = 320, 49%) are aware of interventions such as hearing aids or cochlear implants that are available for children who are born deaf.

3.1 | Views on deafness as a health condition and severity

Participants were asked their views on whether deafness is a serious health condition or a disability, and how they feel about having a child who is deaf (shown in Table 2). The majority (63%, N = 412) agreed that deafness is a serious condition, and 11% (N = 74) disagreed. However, a majority also agreed (60%, N = 391) with the statement that 'with support children who are deaf can live a normal life and it is a minor condition'. Most participants agreed (78%, N = 509) that they would be upset if their child was born deaf whilst a minority (25%, N = 166) agreed that 'having a deaf child is not something that worries [them]'. A majority of respondents disagreed (72%, N = 473) with the statement that deafness is not a disability.

TABLE 1 Demographics of respondents

Demographic	Category	Number	Percentage
Sex			
	Female	463	71
	Male	192	29
Age			
	21–30	172	26
	31–39	444	68
	40 +	39	6
Children			
	Yes, has at least one child	145	22
	No children	510	78
Family origins			
	Africa	1	<1
	Asia	85	13
	European	454	69
	Middle East	11	2
	Oceania	12	2
	North American	1	<1
	Central American	0	0
	Unknown	40	6
	Prefer not to say	1	0
	Mix	50	8
Education			
	Prior to year 12[a]	9	1
	Year 12	29	4
	Certificate	82	13
	Advanced diploma	11	2
	Bachelor	303	46
	Postgraduate	219	33
	Masters	2	0
	Prefer not to say	9	1
Household income (in Australian dollars)			
	Under $50,000	6	1
	$50,001–$100,000	65	10
	$100,001–$$150,000	138	21
	$150,001–$$200,000	160	24
	More than $200,001	222	34
	Prefer not to say	64	10
Religion impacting decision making			
	Not at all	402	61
	Very little	137	21
	Moderately	75	11
	Very much	30	5
	Completely	11	2

[a]Year 12 represents the final year of schooling in Australia.

3.2 | Views on the inclusion of genes associated with NSHL in RGCS and reproductive choices for deafness

A majority of participants ($N = 568$, 86%) agreed that they would like to know their chances of having a deaf child, and agreed ($N = 584$, 89%) that they would choose to undertake screening for deafness if it were offered to them (shown in Table 2 and Table 3). Responses were less consistent on reproductive choices, with 35% ($N = 231$) agreeing they would make different reproductive choices in planning a pregnancy if they were found to have an increased chance of having a child who is deaf. Respondents were asked what reproductive choices for deafness should be available to the general public, and what they would choose themselves if they had an increased chance of having a child who is deaf. A strong majority of respondents agreed that couples in the general population should be able to access IVF with PGT-M (82%, $N = 530$) or PND (89%, $N = 579$) for deafness. However, a minority agreed that couples should be able to access termination of pregnancy (TOP) for deafness (36%, $N = 231$). Similarly, when considering reproductive choices for themselves, a majority agreed they would consider IVF/PGT-M (78%, $N = 504$) or PND (87%, $N = 563$) for deafness but only a minority agreed they would consider TOP (21%, $N = 135$).

3.3 | Views inclusion of genes associated with NSHL in a population wide RGCS program

A majority of respondents (84%, $N = 545$) agreed that screening for deafness should be included in a genetic carrier screening program like the one in which they had participated, and only a minority (32%, $N = 205$) agreed with the statement that deafness is not a condition that fits the criteria for inclusion in a screening program (shown in Table 3 and Table 4). The majority (73%, $N = 475$) think inclusion of genes for deafness should be optional in a screening program like Mackenzie's Mission, that is, only those who choose screening for these genes have this screening.

4 | DISCUSSION

Although the offer of RGCS is becoming part of routine antenatal care, little is yet known about prospective parent's attitudes towards what conditions should be included in the screen. We therefore undertook a survey of prospective parents who elected to have RGCS as part of a

TABLE 2 Participant views regarding clinical impact and severity of deafness

	Strongly disagree % (n)			Disagree % (n)			Neither agree nor disagree % (n)			Agree % (n)			Strongly agree % (n)		
	M	F	Tot	M	F	Tot	M	F	Tot	M	F	Tot	M	F	Tot
Deafness is a serious health condition.	2%	1%	1%	9%	11%	10%	29%	28%	26%	50%	41%	44%	19%	19%	19%
	3	3	(6)	17	51	(68)	39	127	(166)	96	191	(287)	36	89	(125)
Deafness is a minor condition and with the support a deaf child can lead a normal life.	4%	3%	3%	15%	17%	16%	27%	18%	21%	42%	50%	48%	12%	13%	12%
	8	12	(20)	28	77	(105)	52	84	(136)	80	230	(310)	23	58	(81)
I Would be upset if my child was born deaf.	2%	2%	2%	8%	5%	6%	15%	15%	15%	53%	53%	53%	23%	25%	24%
	4	9	(13)	15	24	(39)	28	67	(95)	101	245	(346)	43	116	(159)
I Would find it hard to have my child fitted with a hearing-aid or implant.	30%	16%	20%	37%	38%	38%	23%	19%	20%	8%	22%	18%	2%	5%	4%
	57	75	(132)	71	175	(246)	44	89	(133)	15	99	(114)	4	23	(27)
Having a child born deaf is not something that worries me.	6%	9%	8%	36%	39%	38%	30%	28%	29%	22%	19%	20%	6%	5%	5%
	12	41	(53)	69	178	(247)	57	129	(186)	42	89	(131)	11	24	(35)
There is little impact on a child's growth or development if they are born deaf.	14%	11%	12%	43%	49%	47%	30%	24%	26%	11%	13%	13%	3%	3%	3%
	26	52	(78)	82	225	(307)	57	110	(167)	20	62	(82)	6	12	(18)
Deafness is not a disability	21%	17%	18%	49%	57%	55%	22%	19%	19%	6%	6%	6%	3%	2%	2%
	40	77	(117)	93	263	(356)	41	86	(127)	12	26	(38)	5	9	(14)

Abbreviations: F, Female; M, male; Tot, total participants.

research study. We found high levels of support for the inclusion of genes associated with NSHL in a population wide RGCS program. There was however ambivalence about whether this information would change reproductive decisions (for example, to seek PGT-M or PND). Only one third of participants agreed they would change their reproductive decisions if found to have an increased chance of having a child born deaf. Participants supported providing couples with choices in their reproductive decision-making for deafness but indicated low levels of support for TOP for deafness. Views on reproductive choices were similar between males and females in this study although males were slightly more likely to support TOP as an option for couples in the general population as well as an option for themselves for deafness.

Whilst a majority of respondents indicated deafness is a serious health condition there was also a majority who feel that given access to support like early intervention/hearing aids/cochlear implants that deafness is a minor health condition. Participants who were aware of interventions available for a child born deaf were less likely to view it as a serious condition. Perceived seriousness of deafness may be impacted by awareness of potential support and suggests a possible need to providing education to prospective parents in the pre-testing information to assist them in making an informed and values-based decision.

This study demonstrates a lack of clarity by this group on the severity of deafness given current treatment and management options.

Respondents' strong support for prospective parents making their own informed decisions on screening for deafness and subsequent reproductive options is in line with other studies from the UK[29] and Australia.[18,20] Our finding that only a minority of respondents indicated they would consider TOP for deafness themselves or support it for other couples is similar to the low levels of interest reported elsewhere in the literature.[18,20,29] However, our findings also indicated high levels of interest in PGT-M or PND for hearing loss. RGCS has historically linked the utility of carrier screening to reproductive decision making,[6,14,30] so if prospective parents do not intend to use the information in this way, the utility of including genes associated with NSHL is unclear. The context of availability of reproductive choices (publicly and/or privately funded options) could potentially influence views. This becomes even more important if screening programs are publicly funded and therefore indicate endorsement by the state. While RGCS has been endorsed by medical colleges and professional societies,[6,14,31–33] few jurisdictions to date have established a formal publicly funded screening program. As evidenced in this study, some couples may choose to use RGCS for information to plan and prepare without taking further measures to avoid having a child born deaf. Facilitating reproductive autonomy is an important goal in any offer of a population wide RGCS. In our view, undertaking RGCS to plan and prepare is a valid reproductive choice.

The survey responses do not give a clear indication of what it would mean to this group of respondents to be deaf or have a child born deaf. Overall, participants agreed that deafness is a serious health condition and that they would be upset if they were to have a child born deaf. However, they also agreed that with appropriate support, deafness is a minor condition and a child born deaf can lead a normal life. Deafness is an example of a condition that some of

TABLE 3 Participant views regarding screening for deafness in RGCS and reproductive choices for an increased chance of having a child born deaf

	Strongly disagree % (*n*)			Disagree % (*n*)			Neither agree nor disagree % (*n*)			Agree % (*n*)			Strongly agree % (*n*)		
	M	F	Tot	M	F	Tot	M	F	Tot	M	F	Tot	M	F	Tot
If screening for deafness was included in RGCS…															
We would want to know our chances of having a child born deaf	-	1%	1%	2%	4%	3%	8%	9%	9%	53%	43%	46%	38%	43%	41%
		4	4	3	19	22	15	43	58	101	197	298	72	198	270
We would choose to undertake it	-	1%	1%	-	4%	3%	6%	7%	7%	49%	43%	45%	45%	45%	45%
		4	4		19	20	12	32	44	93	200	293	85	206	291
We would make different choices in planning a pregnancy if we had an increased chance of having a child born deaf.	7%	9%	8%	25%	23%	24%	31%	33%	33%	21%	22%	22%	15%	13%	14%
	14	40	54	48	105	153	60	153	213	40	101	141	29	61	90
Reproductive options for couples in general found to have an increased chance of having a child born deaf should be able to access…															
Access IVF and PGT for deafness	-	1%	1%	2%	3%	3%	15%	16%	15%	45%	45%	45%	39%	35%	36%
		4	4	3	13	16	29	71	100	85	208	293	74	163	237
Access PND for deafness.	-	1%	1%	1%	2%	2%	1%	8%	8%	52%	51%	52%	38%	38%	38%
		4	5	2	10	12	2	37	53	99	235	334	72	173	245
Access TOP for deafness	11%	11%	11%	17%	19%	18%	17%	37%	35%	27%	21%	23%	16%	11%	13%
	20	53	73	32	86	118	32	171	228	52	96	148	30	53	83
Reproductive choices for me personally if I had an increased chance of having a child born deaf, I would consider …															
IVF and PGT for deafness	-	2%	2%	7%	10%	9%	11%	12%	12%	46%	43%	43%	36%	33%	34%
		12	13	13	44	57	20	56	76	88	195	283	69	152	221
PND for deafness.	-	1%	1%	3%	6%	5%	9%	7%	7%	49%	48%	48%	39%	38%	39%
		7	7	6	27	33	17	30	47	94	219	313	74	176	250
TOP for deafness	20%	24%	23%	25%	28%	27%	26%	30%	29%	17%	12%	13%	12%	6%	7%
	38	112	150	48	128	176	50	139	189	33	54	87	22	26	48

Abbreviations: F, female; IVF, invitro fertilisation; M, male; PGT, preimplantation genetic testing; PND, Prenatal diagnosis; TOP, termination of pregnancy; Tot, otal participants.

TABLE 4 Participant views regarding including genes for non-syndromic hearing loss in a population wide carrier screening program

	Strongly disagree % (*n*)			Disagree % (*n*)			Neither agree nor disagree % (*n*)			Agree % (*n*)			Strongly agree % (*n*)		
	M	F	Tot	M	F	Tot	M	F	Tot	M	F	Tot	M	F	Tot
Screening for deafness should be included in a genetic carrier screening program	-	1%	0.5%	-	2%	2%	13%	14%	14%	52%	44%	46%	36%	38%	38%
		3	3		11	11	24	66	90	99	202	301	68	176	244
Deafness is not a condition that fits the criteria for carrier screening in a program	5%	5%	5%	29%	31%	30%	33%	34%	34%	26%	26%	26%	7%	5%	6%
	10	21	31	55	140	195	63	155	218	49	118	167	14	24	38
Deafness should be optional in a RGCS program for those who want it	3%	2%	2%	3%	8%	7%	22%	17%	18%	53%	49%	50%	20%	24%	23%
	6	8	14	5	37	42	41	77	118	101	226	327	38	110	148

Abbreviations: F, female; M, male; RGCS, reproductive genetic carrier screening; Tot, total participants.

those affected consider neither a disability nor a pathology in need of correction, but rather that Deaf[1] people are part of a distinct sociolinguistic community.[11,12] A recent review by our group found that those who know most about the lived experience of deafness (either deaf themselves or the child of a deaf adult) do not see it as a disabling condition.[34] The responsible implementation of a publicly funded RGCS program requires careful evaluation to decide which genes should be included, for example, because inclusion can stigmatise people living with the associated genetic condition. This is one reason why screening only for genes associated with severe childhood-onset conditions is considered the most ethically defensible approach.[35–37] However, debate on what constitutes a severe or serious condition is ongoing, and it is possible that no consensus will be reached.

The study shows that, while respondents tended to agree that deafness is a serious condition, there is no consensus on being prepared to act on the information provided through RGCS by changing reproductive decisions. Participants in this study seemed to value information provision as an outcome of RGCS. This finding is important when considering whether the goal of RGCS is solely to support informed decision making about reproductive choices. The idea of screening 'purely for information' has been explored in non-invasive prenatal testing and was argued to be unacceptable by the authors for non-serious traits.[38] An alternative view however is that providing reproductive risk information upholds reproductive autonomy, which many commentators consider as the fundamental goal of RGCS.

Other considerations for population wide RGCS programs include whether to include all genes in a single offering or have options of gene panels available to prospective parents, and how best to present information to promote informed decision-making. In this study, we found that most participants supported screening for genes associated with NSHL as an optional part of a population-wide screening program. Nearly a third of participants believe that deafness is not a condition that fits the criteria for a population wide RGCS program of the type in which they had participated, that is, severe childhood onset conditions such as spinal muscular atrophy and cystic fibrosis.

Information on living with deafness should always be provided post-test to those found to have an increased chance of having a child with hearing loss. The question of pre-test counselling is more difficult. Providing pre-test information to cover a screening test that involves hundreds of clinically diverse conditions is a challenge that can probably not be overcome in most cases of individuals accessing screening. Instead, general information about groups of conditions is more likely to be provided and understood. In this context, a focus on deafness and the genes associated with NSHL in pre-test counselling may seem counterintuitive. However, if it is offered as an option in testing, targeted information provision is important.

This study has highlighted implementation challenges concerning possible inclusion of genes associated with NSHL including the complex needs of potential parents making decisions together or separately, depending on the approach to RGCS. The differences between couples are likely to be complex as they may have different information and counselling needs which may be indirectly connected to income, trust in healthcare system and possibly educational level.

4.1 | Future directions

In our study, respondents had to imagine a hypothetical situation, as the study in which they participated did not include genes associated with NSHL in its RGCS panel. Such exploratory research on the relationship between stated intentions and real-life behaviour has the potential to further inform future policy decisions regarding implementation of RGCS. We have recently started an implementation study in which participants who have undergone RGCS are offered a subsequent screening for genes associated with NSHL. There may be a gap between intention and actual behaviour in the uptake of RGCS for genes associated with NSHL, and subsequent uptake of reproductive options for NSHL in couples with an increased chance of having a child born deaf.

4.2 | Limitations

A potential limitation of this study is its inclusion of participants who were research participants in a pilot population-wide RGCS research study, who had therefore already experienced RGCS and received a low chance result, which may have influenced their responses. Female participants were overrepresented, which may reflect a greater willingness by women to discuss reproductive attitudes and behaviours. Women are often positioned as more heavily involved in reproductive care in general than men, which may have impacted perceived relevance of the study to women's lives. All participants in Mackenzie's Mission were invited to participate in the study, and this may have introduced some bias if both the male and female member of a couple completed the survey and shared similar views.

Our sample is also unrepresentative, since most respondents were highly educated and from households with a high level of income. Consequently, they may have had greater health literacy and their views may not be generalisable to the broader population. Prospective parents with lower levels of education and household income may have different opinions.

4.3 | Conclusion

The decision to include conditions in RGCS has historically been based on the advice of medical experts.[14] However, the opinions of stakeholders, such as prospective parents who are the end users of RGCS, are increasingly being considered in policy development. Many commercial offers of expanded RGCS already include genes associated with NSHL but offering a publicly funded population RGCS program

[1]Deaf with a capital D refers to the signing community

requires concordance with government policies and must be evidence-based. Whilst the majority of participants agreed that genes associated with NSHL should be included in an offer of RGCS to the population, there was no consensus that this information would influence actual reproductive decision-making. Policy makers should consider this ambiguity in the context of the major aim of RGCS: to inform reproductive decision-making for prospective parents.

These findings provide the first empirical evidence of strong support for the inclusion of genes associated with NSHL in RGCS by those who have themselves previously chosen to make use of RGCS. The findings also supported the uncertainty about the severity of deafness as a health condition, previously reported in the literature.

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CONFLICT OF INTEREST

The authors declare no conflict of interests.

DATA AVAILABILITY STATEMENT

The data are not publicly available due to privacy restrictions associated with the Mackenzie's Mission pilot study. Further information is available in the supplementary material or by contacting the corresponding author.

ETHICS STATEMENT

This research study was approved by the Royal Children's Hospital Human Research Ethics Committee (HREC HREC/53433/RCHM-2019).

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SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

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