Direct-to-consumer genetic tests and Canadian genetic counsellors: An exploration of professional duties in response to novel biotechnologies

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Abstract [430 words]

Direct-to-consumer genetic tests (DTC tests) for health purposes provide genetic information directly to consumers, usually without the intervention of healthcare professionals. The results of such tests may be used by consumers to make informed health and lifestyle choices. That being said, without mediation by healthcare professionals, there is concern for consumer informed consent and privacy. As well, the raw data and results of tests may be misunderstood and misinterpreted by consumers, who may misconstrue the products as medical-grade, clinically useful, and scientifically valid. This could have serious implications for the healthcare system when important diagnoses are subsequently delayed, or consumers self-refer to healthcare providers. There is consensus in the literature and among the Canadian and American Medical and Genetic Counselling Associations that genetic counsellors should be involved in pre- and post-counselling of DTC tests. However, there is a paucity of research regarding how such services could be provided. Here, a close-ended, 48-question quantitative online survey explores how Canadian genetic counsellors could mediate the use of such disruptive technology, from

participating in public education, to promoting consumer informed consent and interpretation of results. These additional roles challenge the existing duties of genetic counsellors and could combat in part the ethical dilemmas raised by the current un-regulated state of DTC genetic tests in Canada. The survey focuses on three key areas: counsellors' concerns regarding 1) informed consent and 2) privacy, and 3) potential policy actions genetic counsellors/counselling associations could implement. Participants were recruited through the Canadian Association of Genetic Counsellors listserv, and the data were analyzed through descriptive statistics. The data were analyzed in regard to the professional duties of genetic counsellors and updated roles are suggested in light of the limited resources of genetic counsellors, while considering the insensitivity of DTC tests toward people of colour and the challenges such tests pose for Indigenous communities. DTC tests could mark a shift towards preventative models of healthcare, with citizens taking active roles to monitor their health, and pose a powerful knowledge translation tool for increasing public science literacy and empowering patients in the genomic era, but with the current flaws, the tests remain merely an untapped potential. This study adds to the existing literature on the professional duties of genetic counsellors and builds on the calls for genetic counsellors to intervene in DTC tests. Additionally, this study aids genetic counsellors and physicians in better understanding the role of genetic counsellors and how to best utilize their professional skills around DTC tests in order to minimize burdens on the healthcare system, and further the understanding of the support required by DTC test consumers.

Résumé de la Thèse [543 mots]

Les tests génétiques destinés directement aux consommateurs (tests DAC) à des fins de santé fournissent des informations génétiques directement aux consommateurs, généralement sans l'intervention de professionnels de la santé. Les résultats de ces tests peuvent être utilisés par les

consommateurs pour faire des choix éclairés en matière de santé et de mode de vie. Toutefois, sans l'intervention de professionnels de la santé, le consentement éclairé des consommateurs et le respect de leur vie privée suscitent des inquiétudes. En outre, les données brutes et les résultats des tests peuvent être mal compris et mal interprétés par les consommateurs, qui peuvent penser à tort que les produits sont de qualité médicale, cliniquement utiles et scientifiquement valides. Cela pourrait avoir de graves conséquences pour le système de santé lorsque des diagnostics importants sont par la suite retardés ou que les consommateurs s'adressent eux-mêmes à des prestataires de soins. La littérature et les associations canadiennes et américaines de conseil médical et génétique s'accordent à dire que les conseillères en génétique devraient participer au conseil préalable et postérieur aux tests DAC. Cependant, il y a peu de recherches sur la façon dont ces services pourraient être fournis. Dans le présent article, une enquête quantitative en ligne utilisant 48 questions fermées explore la façon dont les conseillères en génétique canadiennes pourraient intervenir dans l'utilisation de cette technologie perturbatrice, de la participation à l'éducation du public à la promotion du consentement éclairé du consommateur et à l'interprétation des résultats. Ces rôles supplémentaires remettent en question les devoirs actuels des conseillères en génétique et pourraient combattre en partie les dilemmes éthiques soulevés par l'état actuel non réglementé des tests génétiques DAC au Canada. L'enquête se concentre sur trois domaines clés : les préoccupations des conseillères concernant 1) le consentement éclairé et 2) la protection de la vie privée et 3) les actions politiques potentielles que les conseillères génétiques/associations de conseil pourraient mettre en œuvre. Les participants ont été recrutés par le biais du serveur de liste de l'Association Canadienne des Conseillers en Génétique et les données ont été analysées au moyen de statistiques descriptives. Les données sont analysées au regard des devoirs professionnels des conseillères génétiques et des rôles actualisés sont suggérés à la lumière des

ressources limitées des conseillères génétiques, tout en tenant compte de l'insensibilité des tests DAC envers les personnes de couleur et des défis que ces tests posent aux communautés autochtones. Les tests DAC pourraient marquer une évolution vers un modèle de soins de santé préventif, les citoyens jouant un rôle actif dans le suivi de leur santé et constituer un puissant outil d'application des connaissances pour accroître la culture scientifique du public et responsabiliser les patients à l'ère de la génomique, mais compte tenu des lacunes actuelles, les tests ne représentent qu'un potentiel inexploité. Cette étude s'ajoute à la littérature existante sur les devoirs professionnels des conseillères génétiques et s'appuie sur les appels à l'intervention des conseillères génétiques dans les tests DAC. En outre, cette étude aide les conseillères génétiques et les médecins à mieux comprendre le rôle des conseillères génétiques et la meilleure façon d'utiliser leurs compétences professionnelles dans le cadre des tests DAC afin de minimiser les charges sur le système de santé et de mieux comprendre les besoins des consommateurs de tests DAC.

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Contribution of Authors

Cassandra Haley is the sole author of this M.Sc. thesis. Cassandra completed this work under the supervision and financial support of Prof. Ma'n H. Zawati.

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List of Abbreviations

AMA – American Medical Association

CAGC – Canadian Association of Genetic Counsellors

CBGC – Canadian Board of Genetic Counselling

CMA – Canadian Medical Association

FDA – Food and Drug Administration (USA)

GC (GCs, GCing) – Genetic counsellor/genetic counsellors/genetic counselling

NSGC – National Society of Genetic Counselors (USA)

PSS – Professional Status Survey

Chapter 1: Introduction

1.1 Background

Direct-to-consumer genetic testing (DTC tests) for health purposes is one of the latest innovations of commercialized precision medicine. These genetic tests are available to Canadians to purchase without the intervention of a healthcare professional, with the implicit assumption that knowledge of disease risk or susceptibility could lead to changed lifestyle and health behaviours among consumers[1]. While DTC tests are available for a much wider range of possibilities (such as ancestry testing, physical characteristics, prenatal assessments, and pharmacogenetics, etc.) this thesis will focus on DTC tests related to disease risk assessments and health profiles generated from consumers' genetic information. Compared to DTC tests that only provide physical descriptions or ancestry information, health-related DTC tests may cause consumers to seek care from healthcare practitioners, and of particular concern to this research, genetic counsellors (GCs). Given the limitations of an M.Sc., and the vast size of the industry, this project will concentrate on health-related DTC tests. Notably, pharmacogenomics tests are highly contentious and merit exclusive study, which is beyond the scope of this thesis.

DTC tests present a looming challenge to Canada's GCs and the Canadian healthcare system as the DTC testing industry continues to grow in size and popularity; it is projected to be worth USD $4.2 billion by 2028[2], and 11% of Canadians have reported taking a DTC test[3].

DTC tests grant consumers unprecedented access to their health data, through providing information directly to consumers without primary care physicians' or other healthcare professionals' involvement. The removal of healthcare professionals from discussions of genetic health by DTC tests is particularly concerning and raises questions regarding informed consent and interpretation of results which will be discussed at length herein. However, access to genetic

health data could promote healthy behaviours and lifestyle choices among consumers (indeed, many marketing materials from DTC companies rely on this strategy) although this assumption remains contentious in the literature[1]. DTC tests also raise normative questions around knowledge translation and public science literacy, as tests may increase the public's interaction with genetics and potentially increase public understanding of this dynamic scientific discipline. These tests would undoubtedly benefit public awareness of the scientific research enterprise, although such studies are scarce within the literature and would likely prove methodologically challenging. DTC tests hold great potential to increase public engagement with the field of genetics through the direct access approach to health information.

However, the beneficence of DTC tests remains hotly debated within the literature. While DTC companies usually clarify in the terms of service documents that their proprietary tests are not medical diagnoses, this statement is a poor contender against the distressing and often highly emotional responses invoked by test results. Many DTC tests are perceived by the public as diagnostic tools, warning of associations between one's genetic profile and risk scores for genetic diseases, and when combined with upsetting results, the information these tests provide may be misunderstood and/or misinterpreted by the average DTC test consumer[4–6], who may lack understanding of the possible risks[7]. Consumers may even misunderstand the products as being medical-grade tests[6,8–10], clinically valid[4,11] or scientifically accurate[10,12–14], creating a false sense of security for patients who receive false negative tests, which could have serious implications for the healthcare system when important diagnoses are subsequently delayed. Consumers may delay important prophylactic cancer surgeries after receiving false-negative results, as demonstrated in the case of Victoria Boer, who, after the frustration of months long delays in genetic testing, received a negative BRCA1/2 test from 23andMe, only to receive a startling positive test result

from her health centre weeks later[15]. Victoria discusses how, if she had only relied on the 23andMe results, she would not have pursued prophylactic surgery and would likely have been at a much higher risk of the cancer that caused the death of her own mother[15]. False positives, resulting from wrongly interpreting results, over-interpreting variants of unknown significance, or other methodological faults[14] may also cause consumer anxiety and push them to pursue clinical validation[16]. One study identified a false-positive rate of 40% in genotyping data returned to consumers of genes with potential clinical impact, demonstrating the need for accredited laboratory confirmation and intervention by healthcare professionals to interpret the results[4]. However, clinically validating privately generated genetic data may place additional burdens on healthcare systems when concerned patients self-refer to GCs or other healthcare providers[7,10,17,18].

The clinical utility of DTC tests is a source of controversy within the literature as well. DTC tests fall into two categories: narrow tests and broad tests[19]. The former are often limited in terms of the number of variants they cover (thousands of variants and hundreds of other genes may be absent from the test), while broad tests often include genes that have not yet been associated with an established risk estimate or assigned medical treatment guidelines[19]. Thus, narrow tests fail to provide a complete overview of a consumers' disease risk, while broad tests may lead to inconclusive results[19]. Furthermore, there are often environmental factors such as lifestyle and health behaviours that could contribute to the onset or prevention of disease, which may not be reflected in the test results. Consumers may easily misunderstand that the tests only detect a genetic susceptibility to disease, which may be impacted by a variety of environmental factors. Therefore, the disease results are far from comprehensive and are not necessarily clinically valid[4,11,19].

DTC tests make use of single-nucleotide polymorphisms (SNPs), single base-pair sequence variations, to profile as biomarkers for disease or genetic health, and such tests do not usually

involve sequencing the entire genome[20]. More sophisticated and expensive DTC tests may make use of next-generation sequencing, but most DTC tests on the market only examine a small percentage of genes and loci associated with health risks[21] and thus fail to interrogate all clinically relevant genes and mutations. Medical grade genetic tests, on the other hand, are held to established quality standards set by the Standards Council of Canada[22], and involve more thorough testing beyond SNP-based arrays, including next-generation sequencing, chromosome copy number analysis and even methylation analysis[21]. Test results are interpreted according to medical guidelines by healthcare professionals and draw from patients' personal and family histories to personalize results[21]. The use of next-generation sequencing entails massively parallel sequencing of any or all of the 22,000 coding genes of the human genome to provide a basis for high depth bioinformatical analysis of interrogated loci[23], and medical tests are thus able to present a much more detailed characterization of the genome for health risk analysis.

DTC tests are currently unregulated in Canada, but American medical devices (including DTC tests) imported to Canada must meet the applicable Food and Drug Administration (FDA) requirements[24]. However, samples are often analyzed in the USA, and Canadians must accept the privacy risks that come with shipping genetic samples across the border. The Office of the Privacy Commissioner, the Canadian Medical Association (CMA), and the Canadian Association of Genetic Counsellors (CAGC) all have informational materials regarding DTC tests[25–27].

Using accessible language, The Privacy Commissioner's website is an excellent resource for would-be DTC consumers. The website offers Canadians points to consider before purchasing a test, such as the implications of sharing genetic data, how genetic data can be used or sold for research purposes by DTC companies, and how even once anonymized, genetic data could be connected back to a person[25]. It provides an excellent resource for Canadians to understand the

scope of what DTC tests can test for, how the lack of regulation in the industry could lead to inaccurate tests, and the appropriate internal regulations that companies should have in place to safeguard sensitive health, genetic, and personal information before one considers purchasing a DTC test[25]. The Office of the Privacy Commissioner's website also highlights that Canadians are protected under the *Genetic Non-Discrimination Act* (Bill S-201, 2017)[28], wherein organizations are forbidden from collecting, using, or disclosing personal genetic test results without the consumers' explicit consent[25,28]. However, once Canadians agree to share their genetic information for research purposes, they are no longer covered by the *Act*[25,28]. The *Act* forbids companies and individuals from: forcing other individuals to take or disclose the results of a genetic test, or using or disclosing the results of an individual's genetic test without their written consent, with the exception of healthcare practitioners and researchers[28]. Alongside safeguarding the privacy of DTC test results, the *Act* broadly protects Canadians from being forced to disclose the results to insurance providers or employers and is the only federal legislation currently enacted to protect Canadian's genetic information.

The CMA and CAGC statements on DTC tests bear a strong thematic resemblance. The CMA has expressed concern regarding the patient and societal harms that may arise from DTC testing, given the wide range of clinical validity and reliability of the unregulated tests, and is cognizant of the toll DTC tests may take on Canadian healthcare providers who could be asked to translate results and provide counselling for their patients, which may be outside of their area of expertise[26]. The CMA recommends pre- and post-DTC test GCing, advocates for patient privacy and informed consent, and calls for the regulation of the DTC industry in Canada[26]. The CAGC similarly stresses the importance of informed consent in DTC testing, and advocates for responsible marketing by DTC companies. The CAGC notes that GCs would be an effective

intervention in pre- and post-GCing, but caution that there are limited counselling services available in Canada[27].

Within the United States, the FDA is the sole regulator of DTC tests by categorizing them as medical devices[7], a subsection of the *in vitro* diagnostics (IVDs) they are mandated to regulate. The FDA has adopted a tiered system for regulating DTC tests: those that cover "non-medical, general wellness, or low risk medical purposes" are not regulated, whereas those that test for "moderate to high risk [*sic.*] medical purposes, which may have a higher impact on medical care" are reviewed before they can be sold[29]. On this second tier, the FDA evaluates DTC tests based on reliability, analytical validity, clinical validity, and what claims the company makes about how well the test works[29]. The FDA does not regulate tests that are proprietary to research labs and that are recommended by physicians[29]. The reasoning for this regulation of industry was founded on the concept of the citizens' 'right to genetic knowledge', with the FDA acting as a guarantor of the veracity of the information generated by DTC testing and thus enabling citizens to make informed health choices[7].

Once introduced to their genetic health profile through taking a DTC test, Canadian consumers are often left without support and must grapple with new genetic information that can have profound impacts on their perception of self and their genetic health predispositions[10,12,30,31]. The unfiltered and newfound knowledge DTC tests provide can destabilize consumers' extant self-identification[13,32,33], causing damage to a person's identity by revealing hidden parentage or other familial information previously unknown to the test-taker. Additionally, due to the nature of the tests, which continue to update results as new data is collected and proprietary algorithms are updated, consumers may receive new information that is contradictory to previous results[34]. These results may be extremely distressing to members of the public, who may struggle to dissociate

genetics from identity, and may even reflect technical errors in the tests as previously incorrect information is updated[13]. The intensity of emotional harm DTC tests may cause to some consumers cannot be understated.

More broadly, DTC tests also create gaps between patient and healthcare provider where important knowledge translation is lost. As well, consumers may incorrectly believe doctor-patient confidentiality applies, or that there are standards regulating how DTC companies collect and store samples or personal information, contributing to a false sense of security with purchase, and presenting a number of privacy concerns for Canadians' sensitive health information[11,20,35].

Crucially, DTC tests also raise questions of scientific validity, as companies design and maintain proprietary algorithms trained on their own data sets[36] – often white and homogenous datasets[37] – which often results in vague or inaccurate genetic health results delivered to people of colour (POC)[12,13]. The built-in racial bias of DTC tests perpetuates existing racial disparities in access to healthcare through providing vague or inaccurate data to POC, which could undermine the consumer's ability to make informed health and lifestyle choices based on the results of DTC tests[9,10]. This lack of clinical and scientific validity can have serious implications for the healthcare system when important diagnoses are subsequently delayed.

DTC tests also raise several questions as to the delivery of results and nature of informed consent of consumers. Studies have noted that DTC companies lack pre-counselling, fail to include a comprehensive overview of risks and benefits, present challenges to healthcare data privacy, and lack transparent policies regarding the return of incidental findings[11,20,35,38], all of which present a serious concern for ensuring informed consent at purchase. Consumers may struggle to achieve informed consent according to the accepted model in the literature (capacity, understanding, voluntariness[39]) as they may not have the tools to understand the privacy implications or the

scientific methodology of the tests, leading to a misunderstanding of results. Here, GCs are often consulted to provide interpretation of results in the context of the patients' family history and the current scientific literature. GCs often provide counselling before and after taking medical genetic tests in the clinic (pre- and post-test counselling) to ensure informed consent is achieved and to counsel patients on their results.

A crucial point to consider in this plan, however, is that GCs in Canada are not considered professionals. That is, GCs do not have title protection or delegated acts, as would accompany licencing. Anyone may thus present themself as a Canadian GC without any legal repercussions, although this is unlikely to occur in the healthcare settings as the vast majority of employers require the M.Sc. in GCing and certification by the Canadian Board of Genetic Counselling (CBGC)[40]. This lack of professional status creates issues of standardization within the profession, as the role of the GC is not legally clarified and GCs may hold varying amounts of autonomy in their practice with physicians, ranging from complete supervision to ordering tests and conducting counselling entirely autonomously under the physician's license[41]. The range of possible roles for GCs affects what roles they could adopt in the DTC consumer counselling scenario and merits attention. As well, the lack of professionalization of GCs can create geographic, financial, and psychological barriers to their access[42]. This holds especially true for patients in rural areas, where getting access to GCing services is challenging and results in inequities in access and availability of GCs[43]. GCs historically operate in tertiary care settings, requiring referral[42]. DTC consumers may go to a hospital or genetic clinic and request a consultation with a physician, but the route to accessing GCing services is much more convoluted since they are not professionals. Elsewhere, researchers have suggested a stepwise process to integrate GCs into primary care, in hopes to reduce these

accessibility barriers[44], but the lack of professionalization of the GCing profession is an important factor to consider in efforts to utilize their skills for patients who pursue DTC counselling.

There is consensus in the literature that GCs ought to be involved in pre- and post-counselling of DTC tests to mediate the public's direct access to these technologies, to encourage informed consent at purchase, and to guide consumers through understanding the results of tests[5,6,9,11,45–47]. As well, the American College of Medical Genetics and Genomics recommends that DTC tests be conducted in an accredited laboratory, stressing the need for consumer informed consent and the possible privacy concerns, and states that a board-certified GC or medical geneticist be should be available for pre- and post-test counselling[48]. The CMA, American Medical Association (AMA), National Society of Genetic Counselors (NSGC), and the CAGC similarly have policy statements recommending pre- and post-counselling for DTC tests[26,27,49,50]. However, there is a paucity of research regarding how the services of GCs ought to be best utilized. A 2018 survey of Canadian and American GCs found that a majority (89.7%) of respondents agreed or strongly agreed that DTC companies should provide in-house counselling, while 41% of surveyed counsellors reported feeling uncomfortable providing counselling to DTC consumers due to a lack of knowledge needed to counsel on results, uncertainty around the accuracy of DTC tests, and such counselling being a misuse of clinical time[47]. The lack of GCs or health professionals in consumer access to DTC tests is problematic because it can lead to serious impacts on the healthcare system, as important diagnoses can be delayed because of the false sense of security tests give consumers[6,8–10].

According to the Code of Ethics for Canadian GCs, GCs ascribe to the principles of justice, fairness, honesty and integrity, and so value diversity, pluralism and equality of access to medicine, while supporting patient autonomy and the non-judgemental provision of services[51]. They ought

to stay well-informed of the latest developments in the field, and be prepared to communicate with others in the medical profession as cases require[51]. GCs have a Duty to Inform, as well as extensive obligations associated with their patients and patient secrecy[52], with three core competencies: counselling and communication, genetic expertise, and professional and ethical practice[53,54]. DTC tests present a new challenge to these Duties, because in the era of commercialized genetic tests, where accuracy, validity and utility are uncertain, GCs must grapple with new uncertainties as they seek to provide their professional care. Currently, there are approximately 500 GCs in Canada who could provide counselling on DTC test results, yet there exists no framework outlining how the GCs could respond to this wave of commercialized genetic tests.

For the purposes of this research, the secondary interaction between DTC companies and GCs (Fig. 1) will be examined in detail, whereas the antecedent interaction between the consumer and DTC company will only be examined in the literature review insofar as such inquiry benefits discussion of the GC-DTC company interaction.

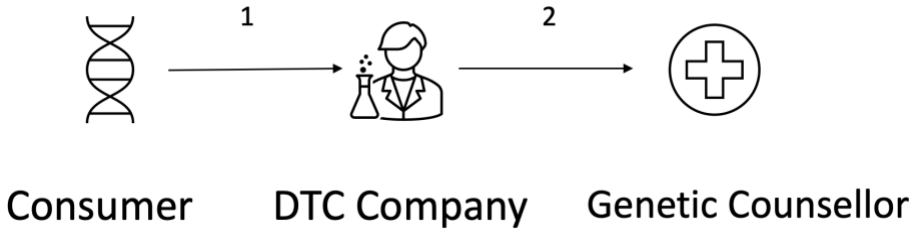

Figure 1. Interactions between consumer, DTC company, and GC through the DTC test odyssey.

DTC tests could pose a potential new route for a preventative model of healthcare, with citizens actively participating in health monitoring through these commercialized personalized medicine products. This model could save the public health system innumerable costs if the tests

are scientifically accurate, clinically valid, and function in diverse populations. DTC tests could pose a powerful knowledge translation tool for increasing science literacy and empowering patients in the genomics era, but without increased intervention by healthcare professionals, the tests remain unregulated, and an untapped potential.

1.2 Research question

For my M.Sc. thesis, I will be examining the following research question: as academic discourse moves toward requiring pre-and-post counselling for DTC tests, is there a role beyond the clinic for GCs and how are their traditional duties challenged? I will explore how GCs could mediate the use of such a disruptive technology, from participating in public education, to promoting consumer informed consent and offering interpretation of results. These additional roles may challenge the existing duties of GCs and could combat in part the ethical dilemmas raised by the current un-regulated state of DTC genetic tests in Canada. The objectives will be to (1) evaluate the current practices within Canadian DTC companies regarding the communication and delivery of results, (2) examine the ethical and policy issues DTC tests present to the traditional duties of GCs, and how they might be updated to reflect the challenges posed by DTC tests, and (3) explore how practicing Canadian GCs view DTC tests in Canada.

With the findings of the survey, I will suggest points to consider for Canadian GCs who are interested in engaging with DTC genetic tests and patients who pursue DTC testing. The survey results will also help to identify ways the traditional duties of GCs could be updated to reflect the challenges posed by DTC tests. These findings will be particularly relevant to GCs to understand how their role may change given the rise of disruptive biotechnologies as the field of genetics continues to innovate.

1.3 Implications

The survey will add to the existing literature on the professional duties of GCs and builds on the calls for GCs to intervene in DTC tests[5,6,9,11,45–47]. The study will help GCs, GCing students, physicians, and policy makers to better understand the role of GCs and how to best utilize their skills in the area of DTC genetic tests to minimize the burden of DTC tests on the healthcare system. This project will also help to inform future standards and guidelines of DTC testing communication standards in Canada, and further the understanding of the support that Canadians require when taking DTC genetic tests. As well, the involvement of GCs with the DTC test delivery and regulation in Canada strengthens the argument for the legal recognition of the GCing profession by augmenting their role in healthcare.

Chapter 2: Literature and normative reviews

2.1 Literature and normative review search terms: Genetic counselling

The objective was to identify all potentially relevant normative documents pertaining to the traditional duties and roles of Canadian GCs. As discussion of the role of GCs is mainly grounded in the CAGC Code of Ethics[51] and Practice-Based Competencies[53], and the CBGC Knowledge Competencies[55], these documents formed the core of the following review. Relevant documents in this qualitative normative review were collected through a series of searches on scientific databases (Google Scholar and SCOPUS, AMA's Policy Finder and CMA's PolicyBase), basic Google searches, and on government or Canadian GCing websites (Government of Canada, CAGC, CBGC), and research was stopped on June 15, 2022. Search terms for the Normative and Literature Review (sections 2.2 and 2.3) included "(Canadian genetic counsel*) AND (role OR competencies OR code of ethics)", "practice of genetic counselling", "genetic counsellor duties", "genetic counselling legal", "genetic counselling ethics", "genetic

counselling accessibility", and "duty AND genetic AND counsel*". Inclusion criteria were documents from GCing organizations or government webpages; advertisements or information about GCing M.Sc. programs were excluded as were documents discussing legal obligations of GCs in countries other than Canada. Literature relating to the duties of GCs in the United States were excluded as being beyond the scope of this thesis.

2.2 Practice of genetic counselling

Currently, there are between 450–500 practising GCs within Canada. There are five M.Sc. GCing programs across the country, graduating approximately 25 students per year in sum total[56]. Canadian GCs must pass a certification exam after graduation, offered by the CBGC, maintaining certification through either continuing education and continuing practice credits, or re-examination every 10 years[40]. Certification from the CBGC or American Board of Genetic Counselling[57] is required by most employers in Canada to practice GCing. The CAGC regulates the vocation of GCing in Canada, and maintains the Code of Ethics[51], Code of Conduct, position statements, and Practice Based Competencies[53], supplemented by the CBGC Knowledge Based Competencies[55]. The CAGC exists to promote standards of practice, support professional growth, and increase public awareness of GCing[58], but the CAGC is not a government agency.

While GCs may practice without direct supervision, the role is not autonomous and there is no existing licensure, registration, or legal recognition of the GC profession in Canada[56,59], with the exception of the province of Manitoba, where GCs are legally delegated by physicians[59]. GCs working within Canada in provinces or territories outside of Manitoba are therefore not subject to provincial or territorial legislation that is designed to ensure public protection through safe, competent, and ethical standards of practice[58]. With the growth of genetics and genomics in healthcare, GCs' specialized training in medical genetics and counselling has led to expanded

clinical roles, and others have argued elsewhere for legal recognition of the profession in Canada[59,60].

According to the CAGC, GCs are "non-physician health professionals with specialized training and expertise in the areas of medical genetics and counselling" and may work on multidisciplinary healthcare teams. The mandate of GCing is to provide "individuals and families with information on the nature, inheritance, and implications of genetic disorders to help them make informed medical and personal decisions and promote an understanding and adaptation of the associated medical, physiological, and familial implications"[53]. The process of GCing involves interpreting family medical histories, educating patients about the inheritance, testing and managing genetic diseases, and counselling patients to promote informed health choices[61]. GCs additionally provide training to GC students and to other healthcare providers to help reduce the burden of genetic risk assessment and counselling on healthcare systems.

The three core competencies of the Canadian GC are to excel at counselling and communication with patients, maintain genetic expertise, and strive for professional and ethical practice in their role[53]. GCs must apply principles of trust, respect, beneficence, honesty and empathy to patient relationships while responding appropriately to patients' emotional states to convey genetic health information to patients in a manner that "meets their needs and levels of understanding"[53]. They must integrate counselling skills with their understanding of medical genetics to effectively manage clinical cases, and be prepared to collaborate with other healthcare professionals, respectful of their own role and that of their teammates while embodying the established ethical values of the profession[53].

With the incorporation of genetic technologies into healthcare practice (whole genome sequencing, exome sequencing, transcriptome sequencing etc.), the role of GCs has extended

beyond the bounds of traditional practice[59] to include work in non-genetic clinical collaborations, laboratories, public health, research, and even private industry. In this dynamic environment, GCs are forced to contend with vast increases in the number of conditions tested and variants considered, expanding their role to include counselling on complex genomic data and health risks, interpretation of variants of unknown significance, and even discussions of pharmaco-genomic implications of test results[60].

The recent introduction of DTC tests to the Canadian market has provided the public with unprecedented access to their personal genetic health data. As discussed in the Introduction, this novel biotechnology has been associated with many consumers accessing additional healthcare after receiving results[7,10,17,18]. Consumers could seek counselling relating to the interpretation of results or possible implications for family planning or other family members[62], or potentially for questions around test methodology, privacy concerns, or to promote informed consent before taking a test. Additionally, consumers are likely to seek counselling if they have previously accessed GCing services, used a DTC test for health reasons, report that they are in fair or poor health, or are confused with the results[63].

There is consensus in the literature that DTC companies should offer GCing pre- and post-DTC test, since the results of these health-related tests could cause psychological distress to consumers[5,6,9,11,45–47]. DTC companies typically either do not mention GCing, direct consumers toward external GCing services, offer in-house advice from a 'health professional', or only offer post-test counselling upon consumer request[5]. For GCs who provide counselling to DTC consumers, elements of a successful consultation include establishing clear contracting at the beginning of the clinical relationship (i.e., motivations for seeking GCing, results interpretation), managing consumer expectations from counselling (including justification of why not all results

may be reviewed with the GC), explaining the limitations of DTC testing (including differences from clinical testing and why additional clinical verification may be unnecessary), and perhaps most importantly, listening to and grounding counselling in the patient's concern for their results[62].

While offering written or video materials can be helpful for educating consumers, authentic, two-way communication through GCing is irreplaceable in the health-related DTC context[5]. DTC companies that do offer GCing must clearly present the professional qualifications and certifications of GCs and the definition of the services, and demarcate of the geographical locations such services may be offered[5]. There are also questions around possible negligence by DTC companies that direct consumers toward a local GC without any provided mechanism for contact, and by DTC companies that implicitly rely on healthcare systems to offer follow-up care after offering distressing results to consumers[5].

The influx of non-medical DTC patients has placed additional stress on the GCing profession, already struggling to cope with long waitlists and high demand for services. When providing counselling to DTC patients, GCs will need to adapt their role to consider the special needs of DTC patients compared to clinical patients (for further discussion see Chapter 5.2). Elsewhere, researchers have described four types of GCing found on DTC test websites: integrated counselling, discretionary counselling, independent counselling, and product advice[45]. GCing then either follows the traditional clinical model, is offered at the consumers' request, is a service offered by a third-party GCing company (e.g., InformedDNA), or is restricted to information about the test being sold (often similar to pre-test counselling)[45]. GCs thus adopt new roles beyond their traditional scope as genetics educators and risk interpreters, to include roles as entrepreneurs, mediators, and lifestyle advisors[45]. GCs have founded telegenetics services, become mediators between consumers and their physicians (GCs may counsel on what to discuss with physicians or

even contact them directly), and may offer DTC consumers lifestyle or health behaviour advice given the test results[45]. These expanded roles may shed light on how disruptive biotechnologies will necessitate evolution in the role of the GC.

While the theoretical toll of DTC consumers has been well described in the literature, there is a paucity of recent empirical studies directly consulting GCs on the subject. One keystone study of American GCs has found that a majority of practicing American GCs feel negatively or very negatively toward DTC tests, and while a vast majority believe GC counselling would benefit DTC testing, few actually feel comfortable providing counselling to patients who have pursued DTC tests[47]. GCs who had counselled patients who had pursued DTC testing on results interpretation held more critical perspectives of DTC testing and were less likely to see value in DTC testing, and more likely to perceive harm therein[47]. GCs perceived positive value for DTC patients in helping patients understand the limitations of DTC testing and explaining the medical significance of results, but cited a lack of knowledge on DTC tests, misuse of clinical time, and questions about test accuracy as reasons they would not feel comfortable providing counselling. Instead, the majority of respondents viewed it as the responsibility of DTC companies to provide in-house GCing to consumers, rather than clinical GCs[47].

DTC consumers evidently present a challenge to the role and traditional duties of Canadian GCs, which has not yet been described in detail. DTC tests pose unique challenges to the communication, patient relationship, and nature of the counselling session and while the sentiments of American GCs have been well described, there exists no parallel in the Canadian context.

2.3 Traditional duties of genetic counsellors

To preface, in the context of DTC tests there are two levels of interaction; the relations DTC consumers have with DTC companies, and the relations between those DTC companies and GCs (Fig. 1). The primary interaction, when the consumer purchases a DTC test directly from the DTC company, involves the informed consent and potentially pre-counselling of the consumer. This interaction is covered extensively in the literature and is not the main focus of this research. The second interaction occurs when the DTC consumer (mediated through the DTC company or of their own volition) contacts a GC. The GC could be employed in-house at the DTC company[45] or external to the DTC company, and it is this interaction that is the focus of this research. The literature review focuses on the interaction between the consumer and DTC company, due to the volume of academic discourse in the area, which will inform the focus of the thesis on the interaction between the DTC company and the GC.

While the profession of GCing is not yet legally recognized in all Canadian provinces, the civil liability of the GC has been explored[52]. Here, role of the GC has been distilled to the traditional duties of the Duty to Inform, Duty to maintain Confidentiality, and the Duty to maintain Competence of medical genetics[52]. These draw from the *Civil Code of Quebec*, the CAGC Code of Ethics and Core Competencies documents, and the legislation regarding delegated acts of health professionals which GCs must not perform (in Quebec: *The Professional Code*).

The Duty to Inform draws from the CAGC Code of Ethics statement: "Genetic counsellors translate complex clinical and scientific information so that individuals and families can adjust to new diagnoses, make informed choices, and benefit from advances in the field of medical genetics"[51]. This obligation includes providing non-directive information on the patient's condition, risks associated with it, treatment options, the purposes and nature and consequences of

treatment, risks associated with treatment, the limits and alternatives to treatment, possible risks to the family, and information on patient support groups or organizations that may be available[51]. The Duty to Inform patients is also an ongoing duty and is not limited to the specific time of the GC appointment. If there are relevant changes in treatment options or the patients' condition, GCs are obligated to continue to inform and keep their patients abreast of developments[52]. This Duty draws heavily from the CAGC Practice Based Competencies[53].

The Duty to Inform is differentiated from the duty to advise, and GCs are to avoid directive information for fear of any possible associations with eugenics[52]. However, others have noted how neutrality in counselling is not always attainable, as counsellors must compete with two opposing objectives when offering information: reducing the inheritance of genetic disease while facilitating the patient's freedom of choice[64]. Any counsellor operating under such circumstances could justify decisions on the grounds of non-maleficence (prevention of harm), and as such there is an inherent eugenic bias in counselling[64]. More recently, there has been suggestions that GCs could provide recommendations to patients in some circumstances based on their clinical expertise, informed by the values and emotions of patients and (if applicable) their family members[65]. This new framework of GCing stresses the importance of balancing beneficence and non-maleficence with the traditionally privileged value of patient autonomy, such that GCs could rely on evidence and their expertise to assess whether testing might benefit a patient, and prevent any results from being used harmfully[65].

The ultimate goal of the Duty to Inform is to promote patient autonomy in the GCing context[52]. GCs are to provide all relevant information as this allows patients to make informed health decisions, and to meet this standard, GCs must obtain free and informed consent as outlined

in the CAGC Code of Ethics[51]. However, DTC tests present unique challenges to informed consent, which are discussed further in section 2.5.

The Duty of Competence is also ascribed to GCs[52]. This Duty describes the fundamental skills relevant to the GCing profession, and is separated into two parts: knowledge-based[55] and practice-based[53] competencies[52]. The knowledge-based competencies draw directly from the CBGC Knowledge-Based Competencies document[55], which spans nine sub-disciplines within genetics ranging from prenatal genetics and cancer genetics to clinical and molecular genetics. The CAGC Practice-based Competencies document in turn defines three areas of skills in GCing: counselling and communication, genetic skills, and professionalism and ethical practice to which all GCs must adhere[53]. The CAGC Code of Ethics encourages GCs to act in their patients' best interest, referring to other professionals as necessary, and to take into account and be attentive to their own limits when offering a patient consultation[51]. It is important to note that in the provision of this Duty, GCs must comply with the relevant legislation, particularly as it relates to the delegated acts of healthcare professionals[52].

The ethical practice guidelines in both the Knowledge-based and Practice-based Competencies mention the need for GCs to "understand the legal and ethical issues pertinent to genetic counselling and testing… including … privacy of health information"[55], and to "be familiar with issues surrounding privacy, informed consent, confidentiality, real or potential discrimination, self-determination and other legal/ethical matters related to the collection, use, disclosure and exchange of genetic information"[53]. This emphasis on privacy protection in regard to genetic data is of particular importance to DTC tests as well, as they present novel challenges to consumer privacy. These themes are explored further in section 2.6.

The third duty often ascribed to GCs is the Duty of Confidentiality[52]. Drawing from the *Civil Code of Quebec* (CCQ)[66], the CAGC Code of Ethics[51] and the Practice-based Competencies[53], this obligation does not fall within the obligation of professional secrecy described in the *Quebec Professional Code*, since GCs are not recognized professionals. Articles 35 and 36 of the CCQ in particular discuss the privacy rights of individuals[52,66]. More broadly, the Code of Ethics bides GCs to "respect their patients' confidentiality in accordance with existing regulations in medical and research settings"[51], while the Practice-based competencies document encourages GCs to "maintain appropriate confidentiality and security in the transmission, storage, management and discussion of professional issues and clinical and research information"[53].

A final duty that is currently under debate within the literature is the Duty to Warn, which exists in tension with the Duty of Confidentiality. Here, GCs may be unclear whether the obligation of confidentiality toward patients ought to take precedence over warning at-risk relatives in the event that a patient refuses to provide family members with pertinent genetic information[67]. A recent study showed that only 30% of American GCs believe they have a duty to inform patients' at-risk relatives[67], while elsewhere it has been reported that 69% of surveyed medical geneticists believe they have an obligation to inform at-risk family members[68]. These contrasting results within the medical community highlight the disputed nature of this obligation. However, the CAGC Practice-based competencies and Code of Ethics only require GCs to respect their patients' confidentiality without discussing any possible obligation to at-risk family members[51,53].

2.4 Literature review search terms: DTC tests

The data in this qualitative literature review were collected through a series of web searches on McGill University Library's website, Google Scholar, and PubMed, and research was stopped on May 31, 2022. The magazine articles referenced came from similar searches on Google.

Primary search terms for sections 2.5 and 2.6 were "direct-to-consumer genetic testing informed consent", "direct-to-consumer genetic testing privacy", "direct-to-consumer genetic test privacy best practices" "direct-to-consumer genetic testing ethics". Secondary variations of these search terms were also employed to gather articles, and reference lists of papers gathered in the above search terms were also mined for further resources. Papers discussing ancestry DTC tests were excluded as beyond the scope of this thesis.

2.5 DTC tests & informed consent

Tandy-Connor *et al*. describe how in the United States, the FDA has granted authority to 23andMe to sell DTC tests for 10 multifactorial conditions: Parkinson's disease, late-onset Alzheimer disease, celiac disease, α-1 antitrypsin deficiency, early-onset primary dystonia, factor XI deficiency, Gaucher disease type 1, glucose-6-phosphate dehydrogenase deficiency, hereditary hemochromatosis, and hereditary thrombophilia[4,29]. With this approval, 23andMe is cleared to brief consumers on their genetic health risk for developing these conditions, but consumers may not fully understand that the genetic risk assessments are not comprehensive; they are merely based on a limited set of mutations that are prevalent in affected individuals, but which have not necessarily been established as causal[4]. These mutations might be associated with increased risk, but there are unknown factors such as environment, lifestyle, and health behaviours which might also contribute to the development of the condition[4]. Furthermore, the list of mutations that 23andMe is able to study is far from exhaustive[4,69]; there might be hundreds of mutations to a given gene and the genetic test only interrogates one or two variants, and there may be many more genes involved in increasing one's genetic susceptibility to developing these diseases than is covered by the test. These diseases are all multigenic and extremely complex, and researchers have not established the complete list of genes implicated in the onset of disease. Tandy-Connor *et al*.

highlight the case study of Parkinson's disease: 23andMe is able to test for one mutation in each of the two genes linked to the disease, but it is well known that several other detrimental mutations exist in these two genes, and that many other genes are also linked to Parkinson's disease[4]. In another example from 23andMe, the BRCA1/2 test includes only three specific variants usually found in Ashkenazi Jewish populations, and so the test is uninformative for individuals of other ethnic backgrounds[19,70].

Additionally, it is entirely possible that the health predictive attempts of the tests could lead to false positives and negatives, with corresponding health anxiety or contrarily, a false sense of security[4] if the consumers did not give full and informed consent. While 23andMe has an entire section of their website devoted to consent[71], the form does not explicitly mention the possibility of false positives and negatives, but rather focuses entirely on consent to future research, not on the consent to the genetic health risks the DTC tests will analyze *per se*. As Matthews *et al.* describe, with the absence of a healthcare professional to mediate, authorize, and support consumers throughout the testing process, it is challenging to ensure informed consent, and to ensure the samples submitted indeed originate from the person taking the test and were not taken without consent[35]. Furthermore, DTC consent documents may not be as rigorous as the standards for informed consent in a medical setting[69], further challenging consumer informed consent.

A 2016 exploratory qualitative analysis of consent forms of several DTC companies found that the examined DTC companies lacked pre-counselling, failed to include a comprehensive overview of the risks and benefits of testing, and lacked transparent policies regarding the return of incidental findings, with the authors recommending that additional measures should be taken to ensure DTC companies operate with informed consent[38]. A 2015 systematic review also identified that DTC companies do not offer enough information about genetic testing to adequately inform

consumers, and found that at the time, DTC tests were "not informative, have little predictive power, and do not measure genetic risk appropriately"[72]. Popovsky has argued that the process of informed consent in DTC tests must also not be overwhelming to consumers, decreasing comprehension through so-called "information dumps" that can mask important details, and ideally would facilitate consumers asking questions and being provided with evidence-based answers to promote understanding[73]. Another review of several DTC websites identified that Navigenics and deCODE (both prominent American DTC companies) fail to clearly promote the informed consent policy on the company website[74]. The absence of healthcare professionals in DTC testing may negatively impact consumer informed consent, potentially resulting in harm to the consumer's health and wellbeing[35,75] when risks or implications may cause consumers to be unduly influenced by aggressive DTC test marketing[5,76].

Autonomous consent in the clinical context, as Hawkins and Ho point out, is a challenging endeavour that involves covering technical and scientific information to patients, as well as discussing any psychosocial values and family issues that patients may approach the session holding[76]. Conveying such knowledge to consumers to promote informed consent may take considerable time and is only challenged by the increasing complexity and breadth of DTC tests[76]. Patients additionally may not comprehend the scope of risks of the tests or the possible harms of genetic information disclosure without counselling[7,76]. If DTC consumers do not understand what they are due to receive from the tests, and do not possess the scientific knowledge required to understand the potential for a false negative or false positive, it remains challenging to ensure informed consent without the mediation of a healthcare professional[35]. A 2015 study of consumers of DTC tests found that 79.1% of consumers were able to adequately interpret the health implications of test results, but that demographics such as higher education level, genetic

knowledge, and numeracy were associated with increased comprehension[77]. The authors identified that old age (above 60) was associated with lower comprehension[77].

There is consensus in the literature that GCs could intervene here to educate consumers further on the scope of genetic tests and information of DTC tests[5,6,9,45,46,72], and pre-DTC testing GCing would be an efficient way to ensure that informed consent is maintained in DTC tests. However, given the limited number of GCs in Canada[56], this is a contentious solution, as reflected in the analogous American context[18].

The questions surrounding informed consent and DTC tests can have serious consequences for the healthcare system. When consumers receive alarming and unfiltered news regarding their health, the anxiety this causes can result in DTC consumers flocking to medical providers, such as general practitioners and GCs[10]. One study has found that 59% of patients self-referred to a GC after taking a DTC test, and the downstream costs of follow-up care were as high as USD $20,604[17]. Although this study was only able to analyze the results of the 22 genetics professionals who responded, to our knowledge it marks the only effort to track the financial impacts on the healthcare system of consumers anxiety after receiving DTC test results. Nill and Laczniak have reported how consumers lacking in informed consent may seek unnecessary and potentially expensive follow-up testing, and that in combination with preventable health conditions avoided by false-negatives on DTC tests, DTC tests can lead to a societal misallocation of resources and overburdening of healthcare system resources[78]. Others have reported that 78% of those who would consider taking a test would consult a physician for help interpreting the results, and 61% of those surveyed believe that physicians have a professional obligation to interpret DTC test results[79]. It is worth nothing that physicians themselves may lack sufficient knowledge of

genomics to counsel DTC consumers on their reports; it has been reported that only 74.4% of American primary care providers provided accurate results interpretation to DTC consumers[80].

2.6 DTC tests & privacy

From the onset, DTC companies privacy policies have been heavily criticized in the literature as being difficult to find, and including clauses that bring into question the companies' commitment to consumer privacy protection[18,81]. The policies are notably convoluted and hard to understand, sometimes with key components such as storage retention, access by third parties, and breach of security notification missing, with companies reserving the right to alter the agreements at any time, without notifying consumers[18].

One of the major concerns with DTC testing is the storage and use of consumers' samples for secondary research and sale. As Allyse *et al.* note, hidden in fine print within the agreement forms of the DTC test kits reviewed for their 2018 article, DTC companies include clauses allowing them to store and use the consumer data "with impunity"[12]. With these articles, companies are free to profit from the sale of aggregate data to unknown third parties and can use the consumer's sample for research without their explicit consent[12]. Such relationships present great possibility for drug development, as researchers can identify groups of genes associated with a particular disorder to target for drug intervention. Given the size and statistical power of the largest DTC companies, mining the data for possible interventions presents a tantalizing and lucrative opportunity for drug development.

DTC tests additionally raise a host of privacy concerns in terms of how companies de-identify samples and encrypt sensitive information, how long they store genetic samples, whether they share samples with law enforcement, insurance companies, or public databases, and what happens to the samples in the event the company goes bankrupt or is sold[6,9,18,20,25,35,81–83].

Importantly, de-identification is frequently used refer to a rule-based process that removes identifiers from data sets[84], and originates from the USA HIPPA 45 C.F.R. §164.514(a) and (b), where it is defined as "health information that does not identify an individual and with respect to which there is no reasonable basis to believe that the information can be used to identify an individual" [85]. In contrast, anonymous information is defined in the General Data Protection Regulation (GDPR, European Union), as: "information which does not relate to an identified or identifiable natural person or to personal data rendered anonymous in such a manner that the data subject is not or no longer identifiable"[86], and is irreversible[84]. From the Canadian perspective, the Tri-Council Policy Statement 2 also presents useful definitions: coded information is information from which "direct identifiers [have been] removed… and replaced with a code", where "it may be possible to re-identify specific participants" [87]. Anonymized information is information that has been "irrevocably stripped of direct identifiers", where no code has been maintained to facilitate relinkage. Anonymous information, as the most stringent method of protection, is "information [that] never had identifiers associated with it"[87].

Sharing the contents of genetic databases with law enforcement could have the unintended consequence of subjecting innocent family members of a potential criminal to police surveillance[78], while sharing with insurance companies could raise premiums as a consequence of genetic discrimination[78,88]. DNA samples taken without consent from others could also lead to third parties being privy to some of the most sensitive information on a person and their family, and consumers may uncover distressing family history when surprises are revealed in parentage.

In the event of a data breach, genetic samples could be reidentified[89] with health and financial information, posing a serious privacy and security concern. Data analysts can potentially re-identify personal genetic data that has been aggregated or anonymized since the record is usually

associated with demographic and lifestyle factors[78,90]. Genetic data breaches can impact the consumer as well as the consumers' family (who usually do not give consent), impacting employment opportunities, and personal relationships[78]. Consumers may also incorrectly assume doctor-patient confidentiality applies, or that there are standard practices in place regulating how DTC companies collect and store samples and personal information, which may lead to a false sense of security when purchasing a DTC test[20].

Of note, leaders in the DTC industry have collaborated to produce a list of privacy best practices including emphasis on transparency, consent, sharing only for the purposes of research, sample storage and retention, accountability, security, and consumer education[18,91]. This short document is supported by 23andMe, Ancestry, MyHeritage, Habitat, African Ancestry, and Living DNA, but is non-enforceable and does not provide clear guidelines other than "reasonable practices" for many of its recommendations[91].

Chapter 3: Methodology

3.1 Background

The culmination of this research was a quantitative closed-ended survey distributed to Canadian GCs (Appendix A). In preparation for this survey, preliminary research in the form of an informal deductive thematic review was conducted, which covered the informed consent and privacy policies of DTC tests. The objective was to understand the existing consent and privacy policies of DTC tests available to Canadians in order to identify themes to interrogate within the survey. This review encompassed the current industry practices of DTC test communication of circulating health-related DTC tests in Canada. The review covered the DTC tests available for purchase over Amazon (n=7, from 23andMe [USA], TellmeGen [Spain], Inagene [Canada], Genomind [USA], 24Genetics [Spain], Advanced Genomic Solutions [USA], MyHeritage

[Israel]), as well as test kits from Nebula Genomics [USA], Circle DNA [Hong Kong, other global offices], Sequencing.com [USA], and Invitae [USA]). Amazon, as the leading ecommerce provider in Canada, was selected for ease of test identification, and results were complemented with a Google search. Discontinued health-related tests (such as AncestryHealth and Dante labs) were excluded, as were tests from companies that do not publicly share these documents. Reviewed documents included the terms and conditions and privacy policies, alongside consent forms.

A follow-up informal deductive thematic review was then conducted to gather the policy statements of authoritative governance and medical bodies in the Canadian and American biomedical research landscape. Relevant normative documents were collected through a series of basic Google Searches and government websites (Office of the Privacy Commissioner, National Human Genome Research Institute, and the FDA). The CMA Policybase AMA PolicyFinder databases were also consulted. Search terms included "direct-to-consumer genetic test policy", and "direct-to-consumer genetic test position statement". The CMA, AMA, CAGC, and the NSGC position statements on DTC tests were consulted as the leading medical authorities in Canada and the United States. Inclusion criteria were leading medical authorities in North America, and position statements of individual journals, DTC advertisements, and more obsolete organizations were excluded from this informal research were.

To explore the policy and ethical challenges DTC tests present to the duties of Canadian GCs, the ethical, legal, and policy documents regarding the traditional duties and roles of GCs were reviewed (see section 2.2 and 2.3). Documents were analyzed from a practical perspective using a deductive thematic approach, to better understand the traditional duties and roles of GCs

toward a patient, and the document search helped to inform questions regarding ways these traditional duties could be updated to reflect the challenges posed by DTC tests.

These reviews aided in preparation of the survey questions by identifying themes and points to interrogate, to better understand industry practices and the challenges inherent to DTC testing. Due to practical constraints, the results of these preparatory deductive thematic reviews are not reported as data here.

3.2 Survey methodology

3.2.1 Study design

This study explored how Canadian GCs could mediate the use of DTC tests to combat, in part, the numerous ethical dilemmas embedded in such a disruptive and unregulated novel biotechnology.

The study objectives were to:

1. Describe the current role of GCs with regard to DTC tests.
2. Compare alternate strategies of involvement of GCs with DTC tests.
3. Elucidate the potential implications of GCs engaging with DTC tests in the context of legal/ethical duties of the profession.

In light of the general skepticism of Canadian GCs towards DTC tests[27], an online survey using a quantitative approach was developed to explore the concerns of practicing GCs, their opinions on the various policy directives within the corporate DTC community, and what they envision their role to be in the industry, following the recommendations of the aforementioned professional societies and think tank. A survey was chosen as the preferred method of data collection due to practical considerations and the timeline of an M.Sc. Quantitative surveys are useful tools for collecting data on large populations, which facilitates greater statistical power, and

provide a feasible manner to gather large amounts of information[92]. Surveys are also useful for measuring individuals' beliefs and opinions on a subject matter and are useful for making direct statistical generalizations about those beliefs and opinions (i.e., they facilitate construct/external validity). An email-based survey, as used here, saves on postage and printing costs, gives access to a wide audience irrespective of their geographical location, offers flexibility of layout and structure, and conveniently houses results downloaded into a spreadsheet[93]. A quantitative approach was chosen over qualitative given the time constraints of M.Sc. research.

The survey questions were informed by the aforementioned deductive thematic policy reviews and centered around the feasibility of pre-/post-DTC GCing, what forms this might take, and how the traditional duties of GCs could be affected by DTC genetic tests. The survey consisted of 48 broad close-ended questions designed to take under 30 minutes to complete and was available in English and French to maximize the number of respondents (see Appendix A).

As this project involved interactions with healthcare professionals, study approval was obtained from the McGill Faculty of Medicine Institutional Review Board prior to data collection. Study participants were treated in accordance with the ethical guidelines of the McGill Faculty of Medicine IRB (Appendix B).

Although there were no foreseeable significant risks with study participation, there was a small risk that participants could be re-identified through indirectly identifying information. As indicated above, there was a plan in place to minimize this risk, but the risk could not be eliminated. Therefore, participants were at liberty to withdraw from the study at any time, with no consequences.

Contact information was not solicited through this study. There was a possibility that participants could be re-identified through indirectly identifying information, since the survey

collected information such as province of practice, field of practice, and work setting. To respect confidentiality, responses were be recorded on a password-protected excel file, only accessible by the student investigator (Cassandra Haley) and Prof. Zawati.

To ensure participants' privacy protection and to minimize risk of re-identification, the following protections were implemented:

1) All surveys were assigned an alpha-numeric code. Participants' identifying information was not collected.
2) Publication parameters are being strictly implemented. Any publications related to this study, including supplemental materials, will minimize information regarding each individual participant to reduce the likelihood of re-identification. Demographic data will be either aggregated or will be presented in a manner which minimizes the likelihood of re-identification (for example, if referring to an individual's work setting and field of practice, their province of practice will not be included).
3) After the study's completion, data will remain anonymized and the separate document linking individual surveys with the alpha-numeric code will be destroyed.

The study team is taking these steps to reduce the risk of re-identification, but a small risk of re-identification remains.

The first 10 items of the survey covered GCs' impressions of the informed consent process in DTC tests. They interrogated GCs' opinions regarding the ways informed consent is covered in DTC terms of service documents, what concepts GCs would like to see mentioned in terms of service documents, and whether GCs and the CAGC can do more to promote informed consent through a variety of consumer-oriented approaches. As well, the questions interrogated how GCs

could envision their roles changing with the unique challenges posed by DTC tests. Questions also investigated the role of the GC in the DTC context. This met aims 1 and 2 of the study objectives.

The next six items of the survey covered GCs' opinions of the privacy aspects of DTC tests, interrogating where counsellors perceive privacy issues in DTC tests, what actions the vocation could take, and whether it is within the scope of GCing to respond to the privacy concerns of DTC tests. Questions in this section asked participants to consider what they think the bounds of their roles should be in the DTC context. This also met aims 1 and 2 of the study objectives.

The next 10 items of the survey delved into GCs' potential involvement in policy around DTC tests. Questions centered around whether GCs believe DTC tests are sufficiently regulated, how the CAGC could become involved with DTC tests, and the feasibility of offering pre- and post-DTC test GCing, as well as what forms this may take. The questions also drew from the recent literature to investigate how GCs feel the field should respond to DTC tests. This section met aim 3 of the study objectives.

To minimize attrition, demographic information was collected in the last section of the survey. There were nine survey items which collected information regarding participating GC's experience working with DTC consumers, their province of work, work experience, and current work setting. This section met aim 1.

Participants contact information was not solicited.

3.2.2 Population and recruitment

The survey was designed such that respondents would provide informed consent and be prompted to answer three questions to ensure they meet inclusion criteria before gaining access to the entire list of questions. Inclusion criteria for this study were: 1) Canadian- or American-certified GCs, 2) currently working in Canada, and 3) GCs who work with patients. Participants

were recruited for the study through an email broadcast to the CAGC listserv, accessible for a $50 fee. Three recruitment emails were sent through this listserv: one on March 1st, March 23rd, and April 4th, 2022.

Before entering the survey, participants were promoted to respond to the following questions to ensure compliance with the inclusion criteria:

1. Are you a Canadian or American certified genetic counsellor? (Yes/No)
2. Are you a genetic counsellor currently working in Canada? (Yes/No)
3. In your role as a genetic counsellor, have you worked with patients? (Yes/No)

If the participant responded 'No' to any of these questions, they were identified as ineligible for the study and the survey would close.

Only Canadian GCs were eligible to participate in the study, and so participation rates were estimated from the results of the most recent Professional Status Survey (PSS). At the time of survey development, the 2021 CAGC PSS was inaccessible, so the results of the 2020 NSGC PSS of both American and Canadian GCs was used to estimate the response rate[94]. The survey reported 2,691 respondents, at a 50% response rate, of which 9% were Canadian GCs. Therefore, 242 Canadian GCs responded. Using this data, at 95% confidence level with a 5% confidence interval, and a 50% response rate, a minimum sample size of 184 would be required to achieve statistical significance. Considering that there are close to 500 total GCs in Canada, and with the broad inclusion criteria outlined above, it appeared feasible that 184 responses would be recorded.

3.2.3. Data collection: Online survey

Data were collected through an online survey, which was created in the LimeSurvey program licensed by McGill University. Collected data was stored in the password protected LimeSurvey platform until the collection phase was completed. The survey was launched on

March 1st, 2022, and the collection phase ended on April 15th, 2022. Upon completion of the data collection phase, the data were exported to a password-protected excel file and housed on a secure server. Access to this data was restricted to the student investigator (Cassandra Haley) and the study supervisor (Professor Ma'n Zawati).

3.3.4 Survey data analysis plan

The sample size failed to meet the requirements of statistical power, but all close-ended survey questions were analyzed through descriptive statistics. Descriptive statistics are useful for calculating, describing, and summarizing research data in an efficient and logical manner, with results presented either numerically or graphically[95].

No open-ended questions were included in this survey. At this stage of the study, aims 1 (describe the current role of GCs with regard to DTC tests), 2 (compare alternate strategies of involvement of GCs with DTC tests) and 3 (elucidate the potential implications of GCs engaging with DTC tests in the context of legal/ethical duties of the profession) were consolidated.

3.3 Survey limitations

This survey data, while useful in describing some of the sentiments of Canadian GCs regarding DTC tests, is not statistically significant. The CAGC advertises over 400 members[96], and 25 members responded to this survey, representing a 6.25% response rate. While this low response rate is not ideal, it is worth considering that electronic surveys are known to have a low response rate, and physicians (and by correlation GCs, as healthcare workers) have lower response rates than the general public[92]. Indeed, the CMA's 2018 National Physician Health Survey had an 8.5% response rate, which the authors note is a typical response rate for online surveys[97]. Furthermore, while the 2020 NSGC PSS was used to estimate response rate, GCs may selectively

answer this survey at a much higher rate than others, since the results of the survey directly impact their work and reflect the status of the entire profession.

The low response rate reported here could have been due to a number of factors, the most predominant being the ongoing COVID-19 pandemic. Due to the toll the pandemic has taken on healthcare workers across the field, GCs have been overworked and expected to cope with higher-than-ever demands as the prevalence of genetics in healthcare has grown and waitlists have increased. After over two years of this extremely burdensome work environment, it is quite plausible that Canadian GCs are burnt out and not responding to survey recruitment as readily as before the pandemic. Furthermore, with high patient loads and a growing demand for their services, GCs must decide between competing a 30-minute survey or providing care to patients in their dossier. In the immediacy of their work, it is not improbable that counsellors would prioritize patient care. The timing of the first recruitment email also coincided with the beginning of Spring break, and given that the field of GCing is 95% female[94], often with young families, the initial recruitment email may have been missed. A possible solution to this low participation rate could have been to extend the survey through several more months, but such an endeavour was outside the timeline of this M.Sc. research project.

Institutions that GCs work with may also have policies that artificially limited the response rate; internal policies may be in place preventing GCs from accepting referrals for patients who engage with DTC tests. Therefore, GCs at these institutions may never encounter DTC consumers and so decided not to respond to the survey.

It is also important to note that survey attrition is a common occurrence with quantitative surveys. The themes and topics explored in this survey are broad and respondents may have more nuanced opinions than the format allows. Therefore, respondents may have had difficulty with the

finite closed-ended structures and may have been unable to engage with the topic in such a way. However, since this study was undertaken within the parameters of an M.Sc. degree, a quantitative survey was one of the most feasible methods for interrogating the opinions of Canadian GCs in regard to patients who engage with DTC tests.

Finally, although unlikely, it is possible that there were some practicing Canadian GCs who were missed in recruitment efforts due to not being a member of the CAGC. However, this population would only represent a small number of the practising GCs in Canada and likely would not have significantly influenced the results.

While a 6.25% response rate is not statistically significant, the results of this survey still add value to the literature. This survey represents the first effort to understand Canadian GCs' opinions on DTC tests, specifically interrogating their views on how DTC tests challenge important paradigms of the GCing vocation including informed consent and privacy. To our knowledge, this survey is also the first to examine how this disruptive biotechnology impacts the traditional duties and roles of the Canadian GC, providing initial exploratory evidence of how Canadian GCs may view their changing roles as they adapt to unprecedented public interaction with genetic information.

Chapter 4: Results

4.1 Participation

In total, 42 participants opened the survey. Within this pool, 88.1% (n=37) of respondents responded in the affirmative to the consent document, 7.1% (n=3) respondents responded in the negative, and 4.8% (n= 2) did not complete the consent form. As for the inclusion criteria, 32 participants affirmed they were Canadian- or American-certified GCs (five were not; presumably students), 33 participants affirmed they were currently working in Canada (four were not), and 35

responded that they have worked with patients in their role as a GC (two responded negatively). Two surveys were initially accessed in French, but both these participants did not consent to the survey. All other responses answered the survey in English. Finally, only completed surveys were included in data analysis; one participant failed to complete the entire survey and was eliminated as an outlier. Thus, there were 25 participants who met all the inclusion criteria and provided informed consent, and who therefore entered the survey, representing a 6.25% response rate. Of these 25, most participants responded to every question, with just one question garnering 23 responses.

4.2 Demographic information

16% (*n*=4) of GCs reported no experience working with DTC consumers, while 84% (*n*=21) GCs report having provided counselling to patients who have pursued DTC testing (Fig. 2). Most GCs (56%, *n*=14) have provided counselling to between 0–5 patients who have pursued DTC testing. 20% of GCs (*n*=5) have counselled between 6 and 10 individual patients, 12% (*n*=3) have counselled between 11-20 individual patients, and 8% (*n*=2 have counselled between 21 and 50 individual patients. Only one GC reported counselling more than 50 individual patients who have pursued DTC testing (Fig. 2).

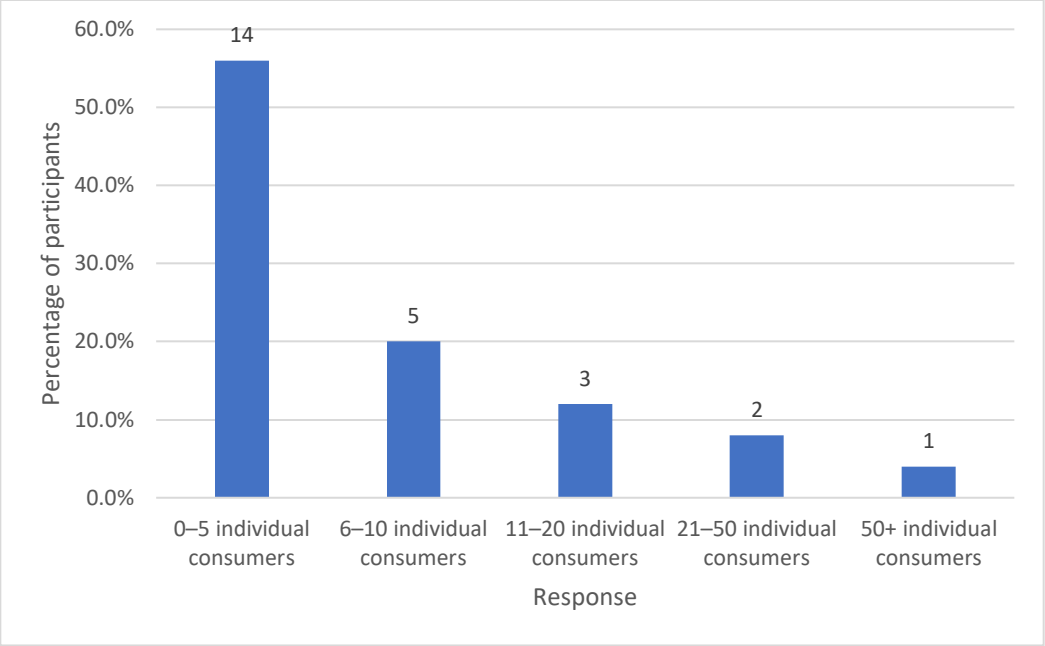

Figure 2: Number of individual DTC consumers to whom GCs have provided counselling. *Most GCs have counselled under 5 individual patients who pursued DTC tests. A minority of GCs have counselled over 20 individual patients who pursued DTC tests. Participant counts also displayed.*

Of the GCs who report having provided counselling to patients who have pursued DTC testing, 68% (n=17) of GCs ranked the interpretation of results as the concern most often raised by clients. 16% (n=4) ranked implications for family members as the other concern most often raised by clients. 60% (n=15) GCs ranked the implications for family members as the second most often raised concern of patients who pursue counselling after DTC tests, with 16% (n=4) ranking the interpretation of results and 4% (n=1) of GCs ranking the lack of informed consent as such. GCs were more divided over ranking the third most concern of their patients who pursued DTC tests; 36% (n=9) of GCs listed privacy concerns as the third greatest concern, 20% (n=5) GCs listed the lack of informed consent, and 4% (n=1) listed the implication for family members as the third greatest concern. As for the fourth ranked concern of patients who have pursued DTC testing,

32% (*n*=8) of GCs listed that the lack of informed consent was their patients' greatest concern, while 24% (*n*=6) listed privacy concerns as their patients' greatest concern. There was attrition between every ranking level, with all 21 GCs who have worked with DTC consumers completing the initial ranking, which dropped to 20 for the second ranking, 15 for the third ranking, and 14 completing the fourth ranking (Fig. 3).

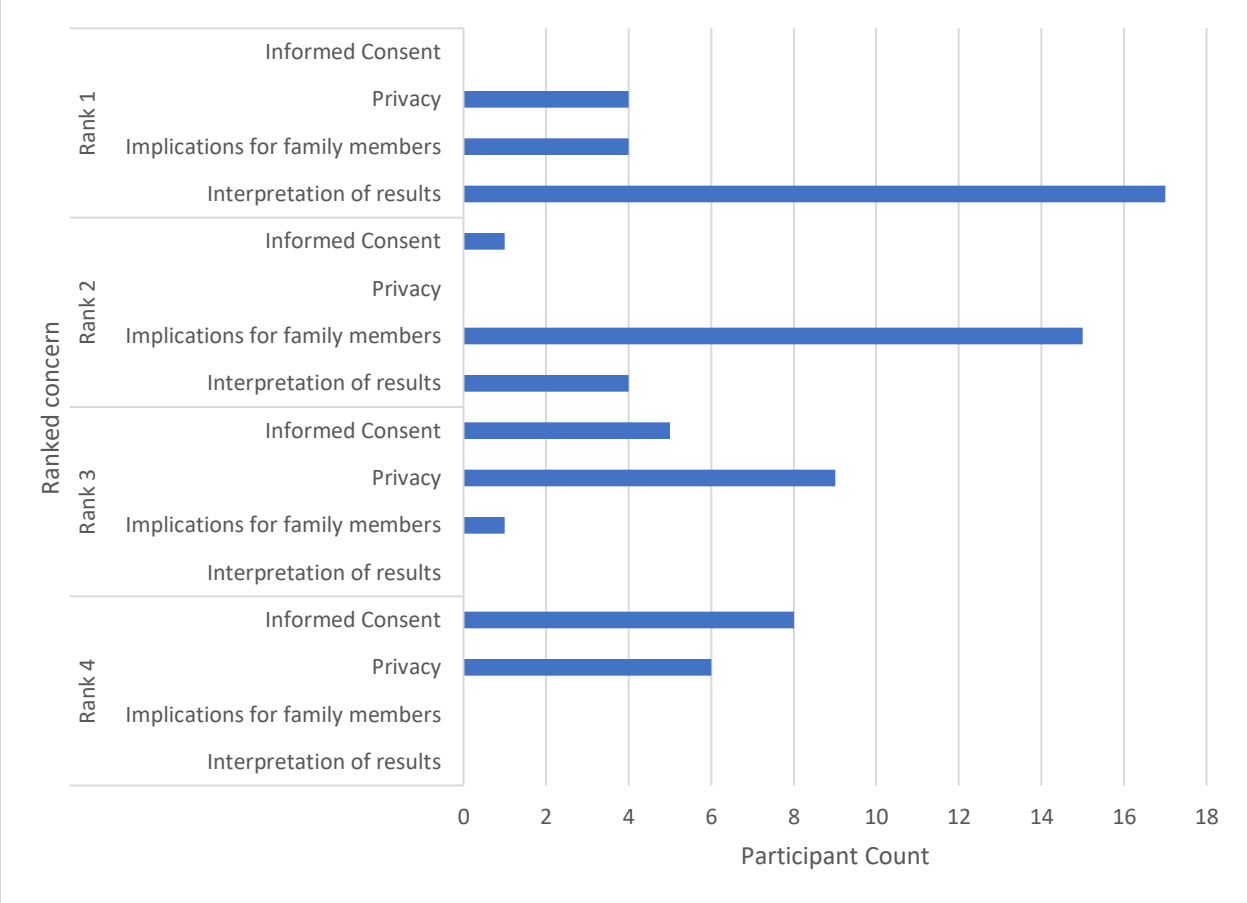

Figure 3: Ranking of patient concerns reported to GCs who have worked with DTC consumers.

Majority of GCs listed interpretation of results as patients' top concern, with implications for family members as the second highest concern, followed by privacy concerns and informed consent as the third and fourth highest concerns, respectively.

Of those who have worked with patients who pursued DTC testing, 36% (*n*=9) of GCs report having worked with other healthcare professionals, while 48% (*n*=12) responded in the negative (Fig. 4).

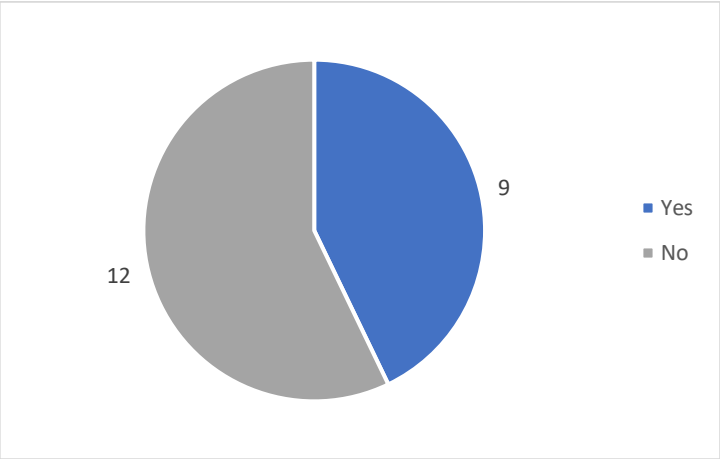

Figure 4: GCs' collaboration with healthcare professionals when counselling DTC consumers.

Of those who have worked with patients who have pursued DTC testing, most GCs report not having collaborated with other healthcare professionals.

Most GCs who responded to the survey were based out of Ontario (48%, *n*=12), with some GCs responding from Alberta and British Columbia (12%, *n*=3 each), Manitoba and Quebec (8%, *n*=2 each), and Newfound & Labrador, Nova Scotia, and Saskatchewan (4%, *n*=1 each) (Fig. 5).

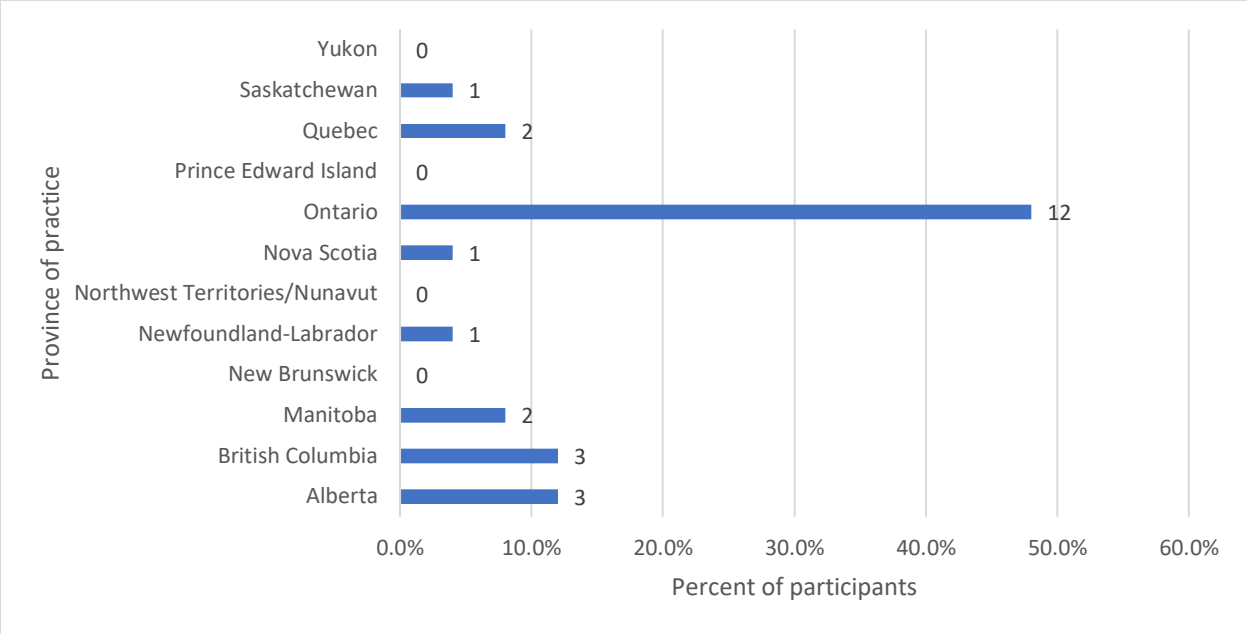

Figure 5: Province of practice of participating GCs.

Most GCs who responded were based out of Ontario, with some answering from Alberta and British Columbia, Quebec and Manitoba, and Saskatchewan, Newfoundland-Labrador, and Nova Scotia GCs also participating.

Participants of this survey all graduated after 1992, with most participating GCs having graduated after 2010 (Fig. 6). The eldest participants were the two GCs who graduated in 1992, while four GCs who graduated in 2021 responded.

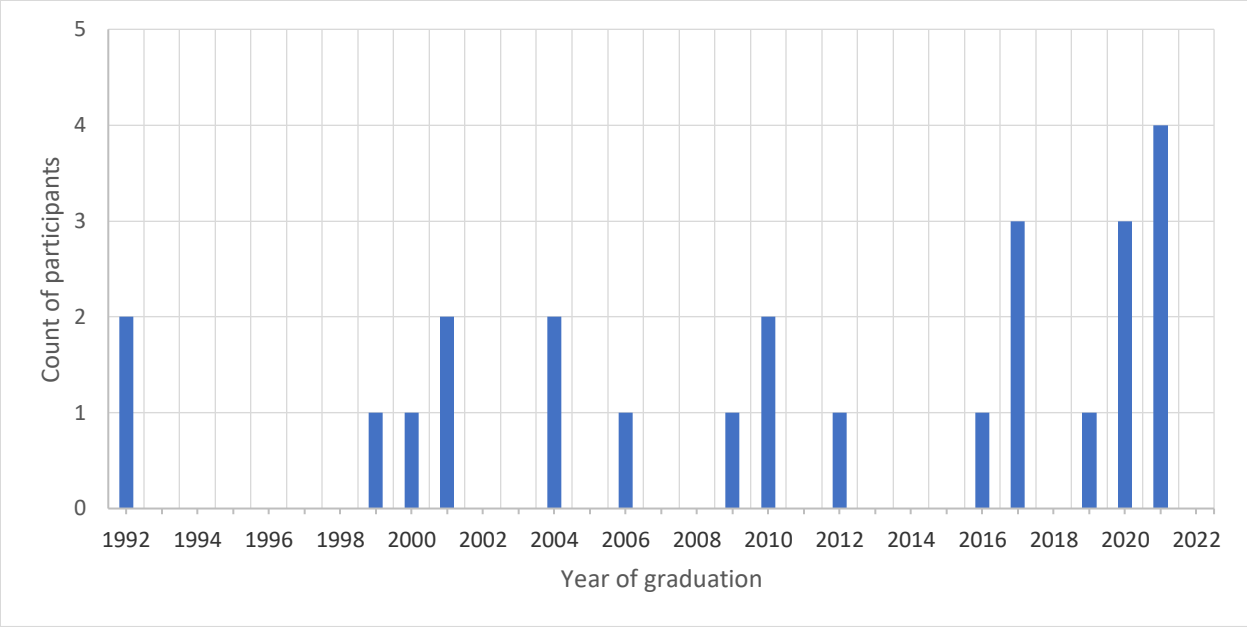

Figure 6: Graduating year of participating GCs.

The majority of GCs who participated graduated after 2010, with 2 GCs who graduated in 1992 responding, and 4 GCs who graduated in 2021 responding.

Most of the GCs who participated in the survey report their current role as clinical (88%, *n*=22), with GCs from laboratory (16%, *n*=4), research (12%, *n*=3), academic (12%, *n*=3), and industry (8%, *n*=2) participating, as well as two GCs who categorized their current role as other (8%, *n*=2, Fig. 7).

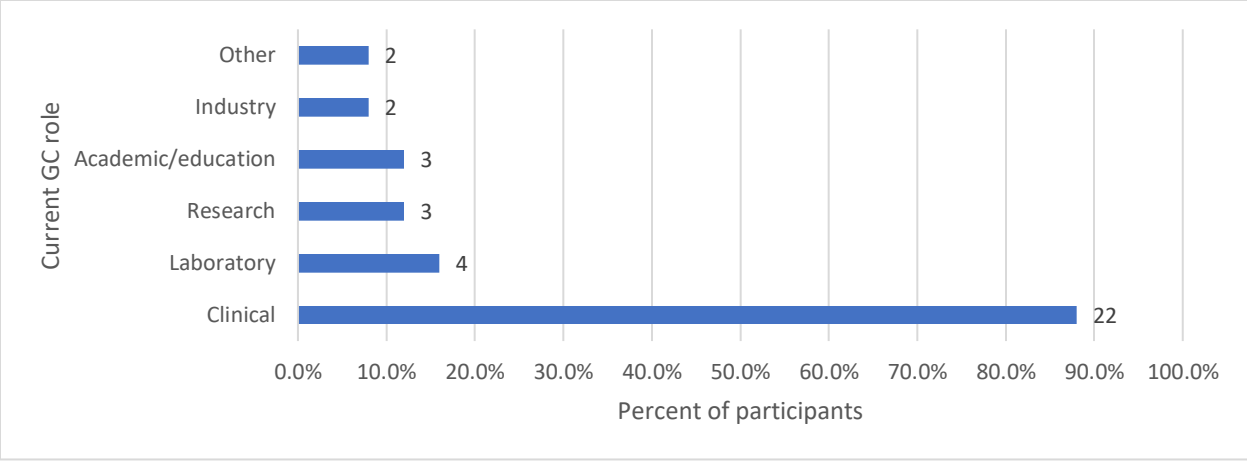

Figure 7: Current role of participating GCs.

Most GCs report their current role as clinical, with representation from GCs who work in a laboratory, in research, academia and industry. Participant counts also shown.

In terms of previous role, most GCs (80%, *n*=20) listed clinical, with (20%, *n*=5) having previous experience in the academic setting, and 12% of GCs (*n*=3) reporting experience in laboratory, research, or other, respectively (Fig. 8).

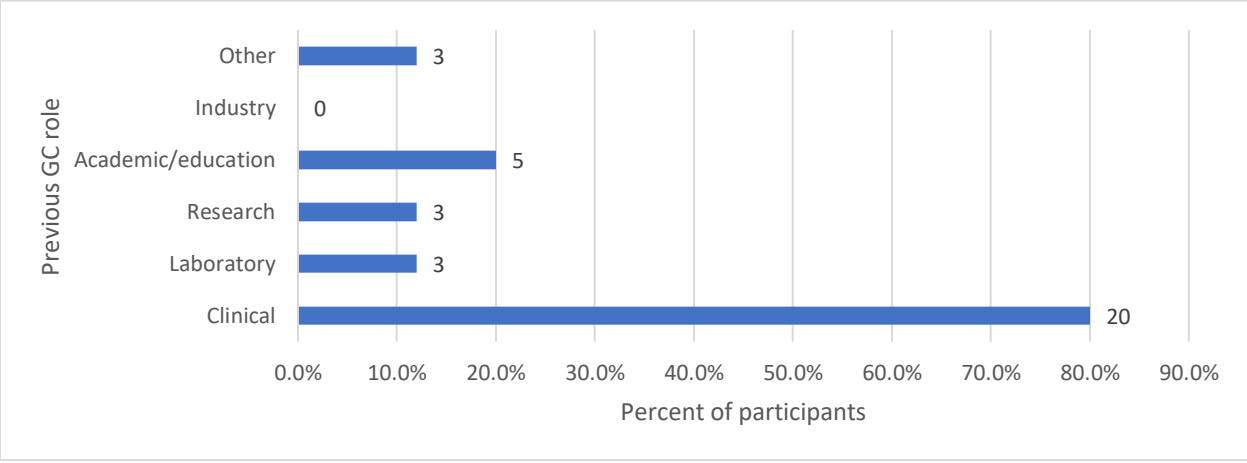

Figure 8: Previous role of participating GCs.

Most GCs reported their previous role as clinical, academic, laboratory, research, or other.

In terms of current work setting, most GCs (68%, *n*=17) answered that they work in a genetic clinic, while GCs also report working in a university (20%, *n*=5), a non-genetic department/division within a healthcare institution (public sector, 12%, *n*=3), in industry (8%, *n*=2), and 4% of GCs (*n*=1) reporting working at a private-pay clinic, in the government, or other, respectively (Fig. 9).

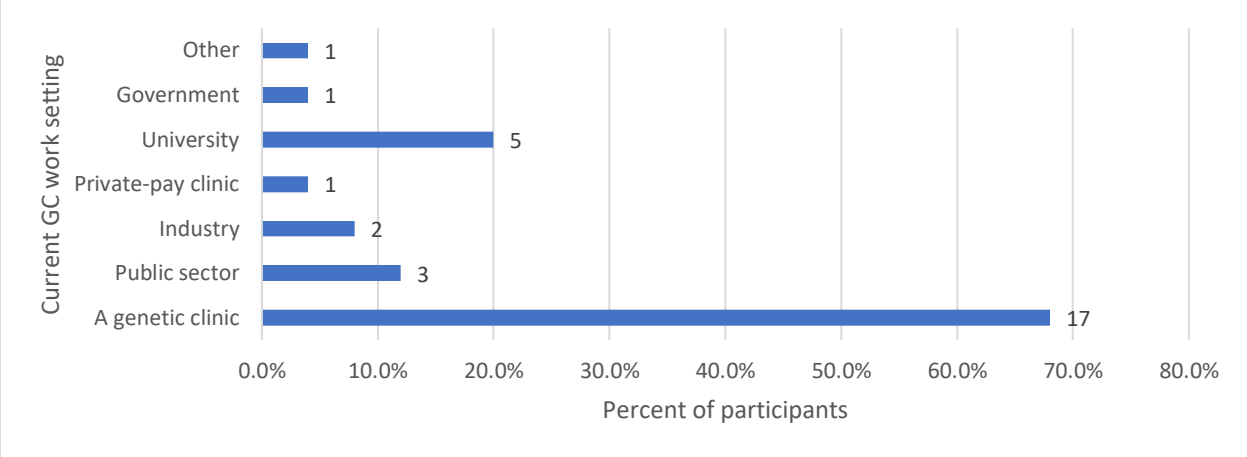

Figure 9: Current work setting of participating GCs.

Most GCs report working in a genetic clinic or a university setting.

Most GCs report previously working in a genetic clinic (72%, *n*=18), with 16% of GCs (*n*=4) reporting previous work at a university, 4% (*n*=1), a non-genetic department/division with a healthcare institution (public sector), while 8% of surveyed GCs (*n*=2) selected other (Fig. 10).

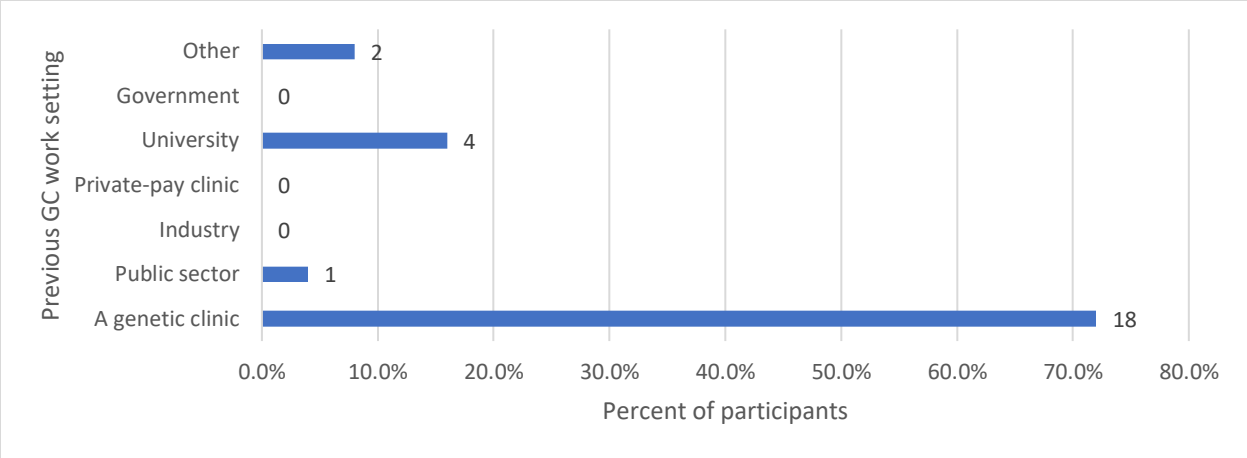

Figure 10: Previous work settings of participating GCs.

Most GCs report working in a genetic clinic or a university setting.

4.3 Informed consent

When asked if they considered working with DTC consumers part of their professional obligations, GCs appeared very divided: 56% (n=14) responded in the affirmative, while 44% (n=11) considered consumers who pursue DTC tests outside the scope of their professional practice (Fig. 11).

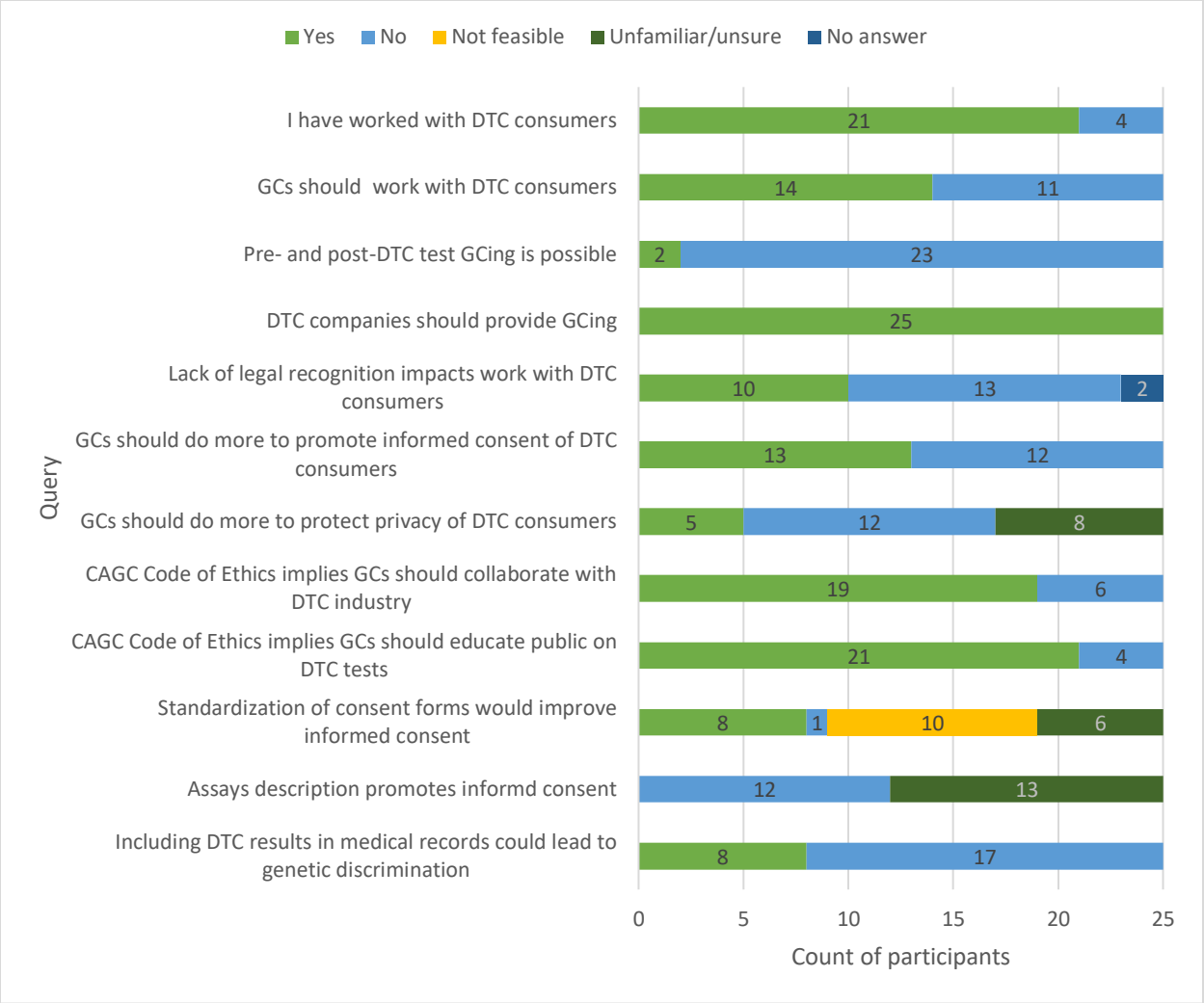

Figure 11: GCs responses to yes/no survey queries.

Participant counts also displayed.

While the majority of GCs (48%, *n*=12) were unfamiliar with the terms of service documents of the DTC industry, 32% of GCs (*n*=8) reported that the terms of service documents poorly described the benefits and limitations of the test. 20% (*n*=5) of participants reported that these documents were too varied across the industry to be compared (Fig. 12).

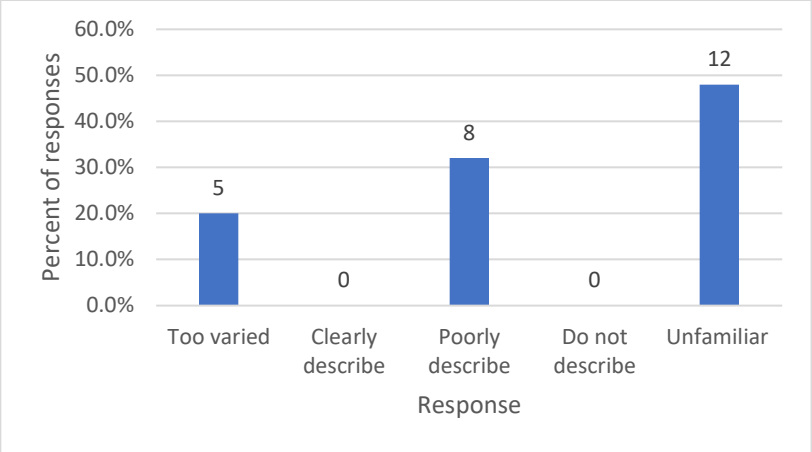

Figure 12: Opinions of GCs on clarity of benefits/limitations in DTC terms of service documents.

While most GCs were unfamiliar with terms of service documents, others feel benefits and limitations are poorly described in terms and service documents or terms and service documents vary too much across industry to comment. Participant counts also displayed.

Participants who reported being unfamiliar with DTC terms of service documents were prompted to select which elements they would prioritize for inclusion (n=12, Fig. 13). They were allowed to select as many responses as they felt appropriate, which resulted in very close results. The most popular element GCs selected was disclaimers: not medical diagnoses/devices, no guarantee of accuracy/security, not liable for damages, will disclose to law enforcement (n=12). 11 counsellors highlighted the need for a layman's description of services, a discussion of the limitations of DTC tests (in terms of comprehensibility, gene/environment interaction), and mention of the retention/use/storage/disposal of genetic sample in DTC terms of service documents. Interestingly, only nine of the GCs selected that DTC terms of service documents should carry the recommendation to speak with a GC.

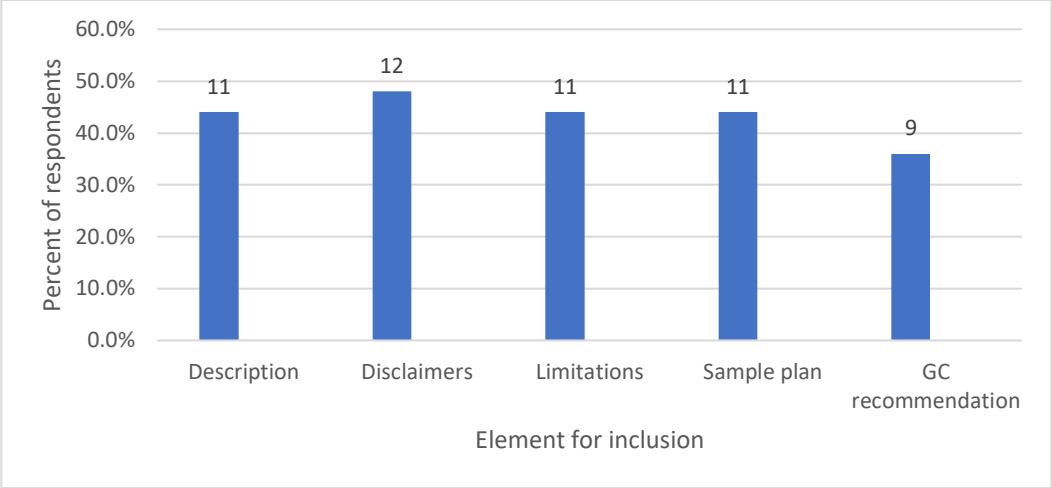

Figure 13: Elements of DTC terms of service documents GCs who are unfamiliar with DTC tests would prioritize for inclusion.

Most GCs wish to see a description of the tests, disclaimers related to diagnoses, accuracy, and liability, as well as the limitations of the tests and a plan of what happens to the genetic sample provided, while fewer GCs want to see a recommendation to speak to a GC. Note that participants were allowed to select multiple reasons, resulting in a higher count than number of participants for both categories. Participant counts also displayed.

Among the GCs who were familiar with DTC terms of service documents, all agreed that the documents failed to use language that adequately describes the sequencing technology and/or assays in the test to promote informed consent (48%, n=12). The remaining participants (52%, n=13) were unfamiliar with DTC terms of service documents (Fig. 11).

Most GCs (52%, n=13) agreed with the statement that DTC tests removing GCs from discussions of genetic health data negatively impacts consumer informed consent, with 8% (n=2) strongly agreeing, 24% (n=6) responding as neutral and 16% (n=4) disagreeing with the statement (Fig. 14).

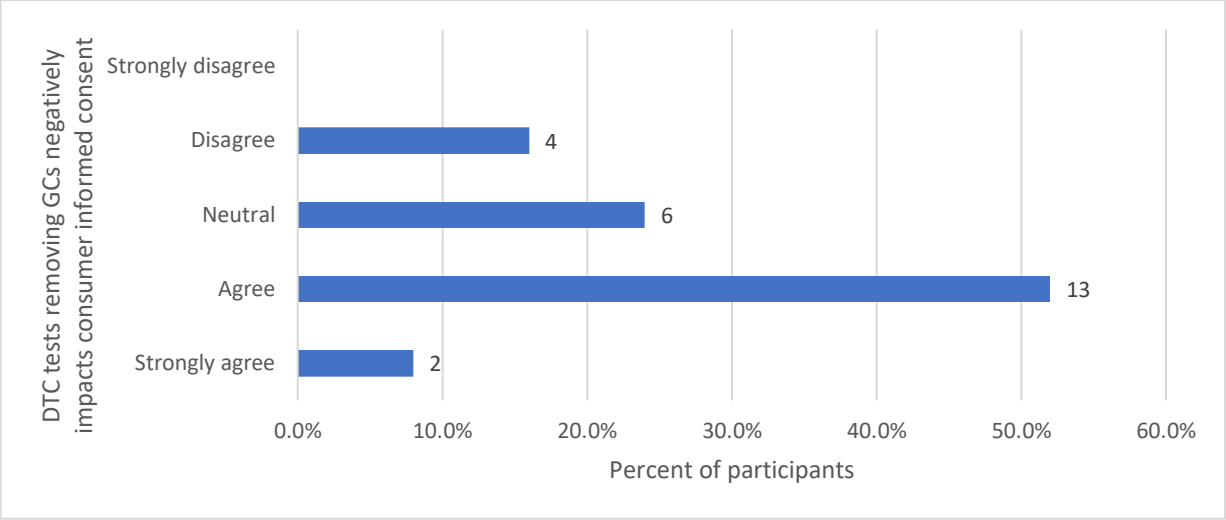

Figure 14: DTC tests' impact on consumer informed consent.

Most GCs agreed that the removal of GCs from discussions of genetic health data by DTC tests negatively impacted consumer informed consent, with 4 dissenting GCs and 6 GCs reporting neutral responses to the statement. Participant counts also displayed.

When asked if standardization of consent forms could contribute to promoting consumer informed consent, 32% (*n*=8) of GCs agreed, while 24% (*n*=6) deemed it unfeasible to enforce, 40% (*n*=10) were unsure, and one respondent did not believe standardization would benefit consumers' informed consent (Fig. 11).

GCs were asked whether the CAGC Code of Ethics directive to "promote awareness of the roles of medical genetics professionals"[51] included collaboration with the DTC test industry, with 76% (*n*=19) GCs agreeing and 24% (*n*=6) GCs disagreeing (Figs. 11, 15). Of the GCs that responded positively, 16 GCs answered this would allow for the evaluation of DTC tests, 14 GCs answered this would allow for standardized informed consent, and five selected that DTC company funding could allow for further research and development in the GCing field. Of the six GCs who responded in the negative, two listed the cause as beyond the scope of the GC practice, while three

listed their reason as being a conflict of interest, and one did not make a selection (Fig. 15). Note that within the sub-question, GCs were allowed to pick more than one reason to support their answer, resulting in percentages over 100 and counts above the number of respondents.

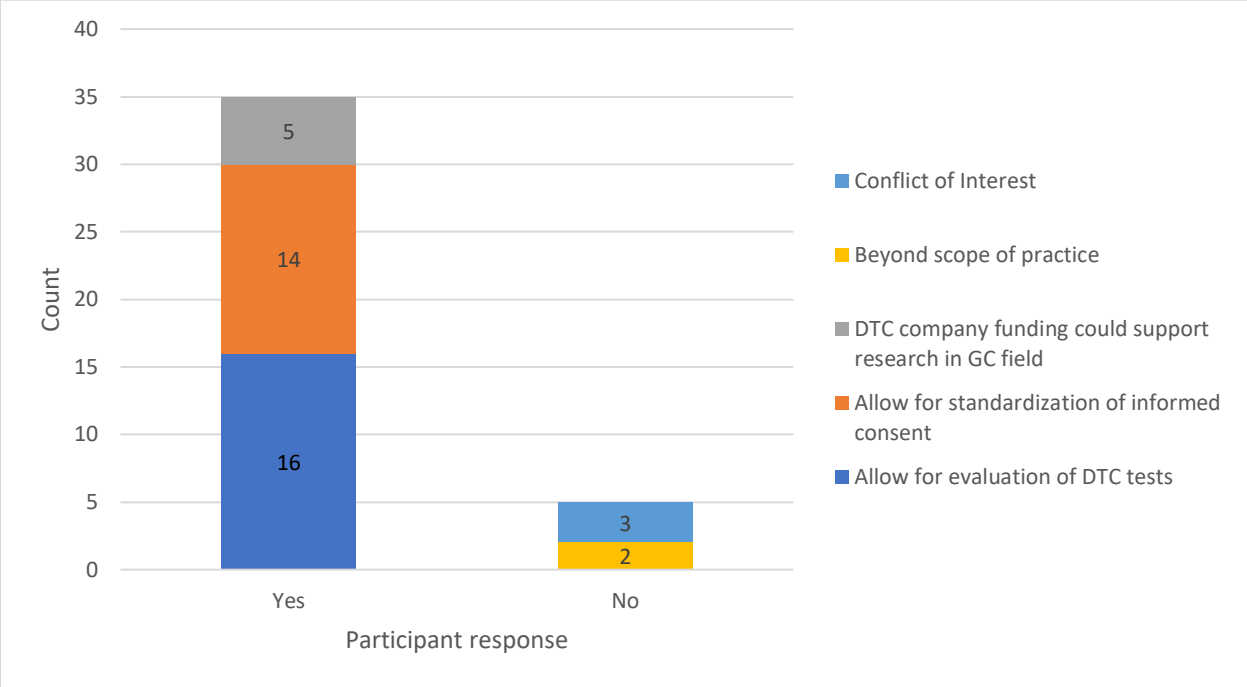

Figure 15: GCs responses to suggestion of collaboration with the DTC test industry, stratified by explanations.

Most GCs believe the CAGC Code of Ethics should include collaboration with the DTC industry, while a minority of GCs disagreed. Note that participants were allowed to select multiple reasons, resulting in a higher count than number of participants for GCs who selected 'yes'. Participant counts also displayed.

GCs were also asked whether the CAGC Code of Ethics section stating GCs should "... promote awareness of the roles of medical genetics professionals through activities such as participation in multi-disciplinary teams, providing public education, contributing to policy-

making and provincial/national consultation" [51] ought to apply to information on DTC tests. Here, 84% (*n*=21) responded yes, while 16% (*n*=4) responded no (Fig. 11).

Participants were split regarding if the field of GCing should do more to protect the informed consent process for DTC consumers (Figs. 11, 16). A slim majority of GCs (52%, *n*=13) believed that the field could take more actions, since the limitations of the tests could be unclear to consumers (44%, *n*=11), the accuracy of the tests could challenge informed consent (32%, n=8), and the absence of healthcare professionals could challenge the process of informed consent (44%, *n*=11). However, 48% (*n*=12) of GCs surveyed responded that the field should not do more work to promote the informed consent of DTC consumers. Of these, 36% (*n*=9) listed that such work would be an inefficient use of clinical time as their reasoning, while 20% (*n*=5) responded that the tests are not medically necessary and therefore do not merit intervention from GCs. No GCs were concerned they lacked the necessary training to provide informed consent to DTC consumers.

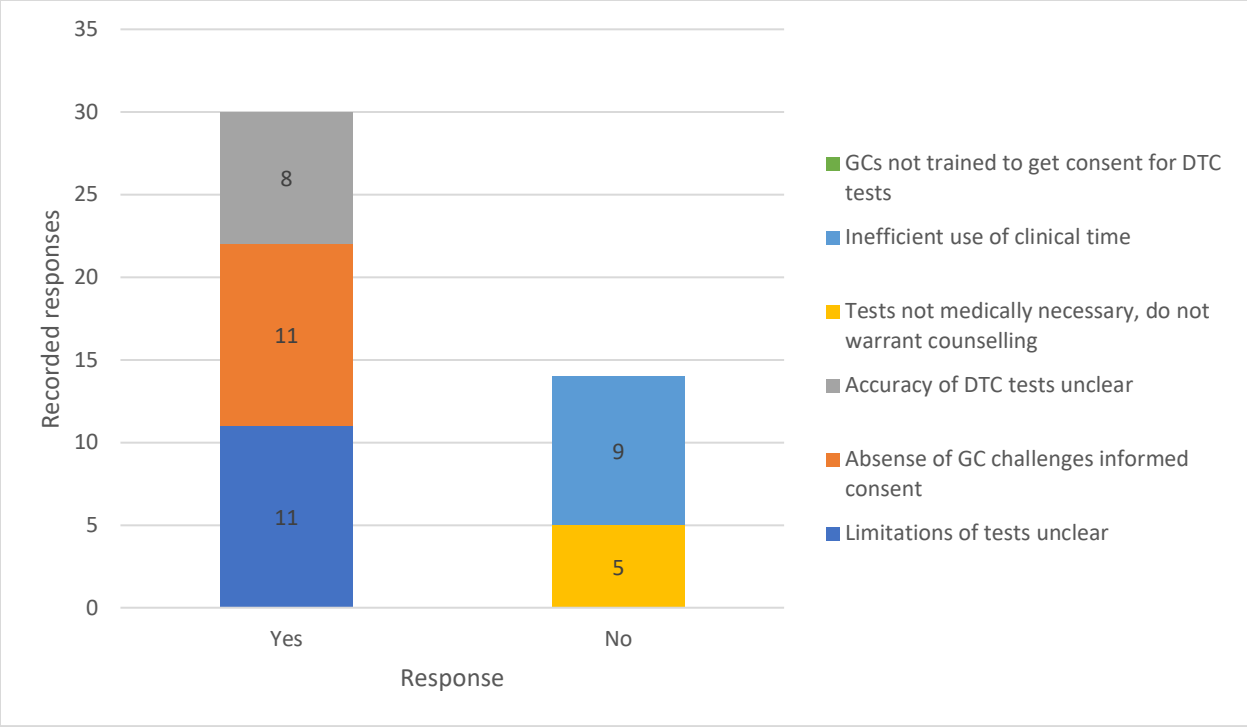

Figure 16: GCs believe the profession needs to protect the informed consent of DTC consumers.

A slim majority of GCs believe the profession needs to do more to protect the informed consent of DTC consumers, while a minority of GCs do not believe the profession should do more to protect the informed consent of DTC consumers. Note that participants were allowed to select multiple reasons, resulting in a higher count than number of participants for both categories. Participant counts also displayed.

When asked how the continually changing nature of DTC tests could challenge the practice of GCing, the majority of participants declared that such changes would not impact GCing, since existing medical genetic tests continue to evolve regardless (56%, n=14). GCs also highlighted how this evolving technology could prolong the relationship with DTC consumers to offer more counselling as results are updated (40%, n=10), but placed less emphasis on working with growing

medical teams in the course of treatment (24%, *n*=10) or that DTC tests would expand the knowledge required of GCs beyond a reasonable understanding of the tests, diverting time and energy from clinical patients (24%, *n*=10) (Fig. 17).

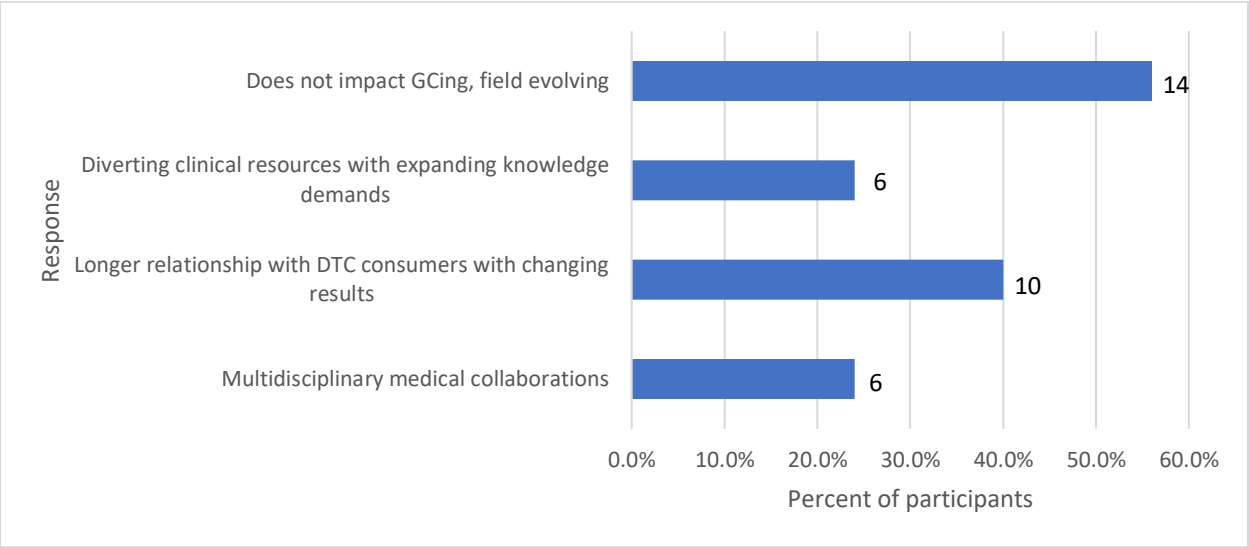

Figure 17: Potential challenges to GCing posed by the ever-evolving nature of DTC tests. *The majority of GCs did not believe the tests would impact counselling, given the rapidly changing nature of the field, although some GCs noted the tests could lengthen the counselling relationship with DTC consumers. Note that participants were allowed to select multiple reasons, resulting in a higher count than number of participants for both categories. Participant counts also displayed.*

4.4 Privacy concerns

52% (*n*=13) of GCs are concerned with the privacy polices of DTC companies operating in Canada. However, on a scale of 1 to 5 (1 being unconcerned, 5 being strongly concerned), 40% selected 3 or less (Fig. 18). The average answer was 3.52, corresponding with concerned (standard deviation 0.92).

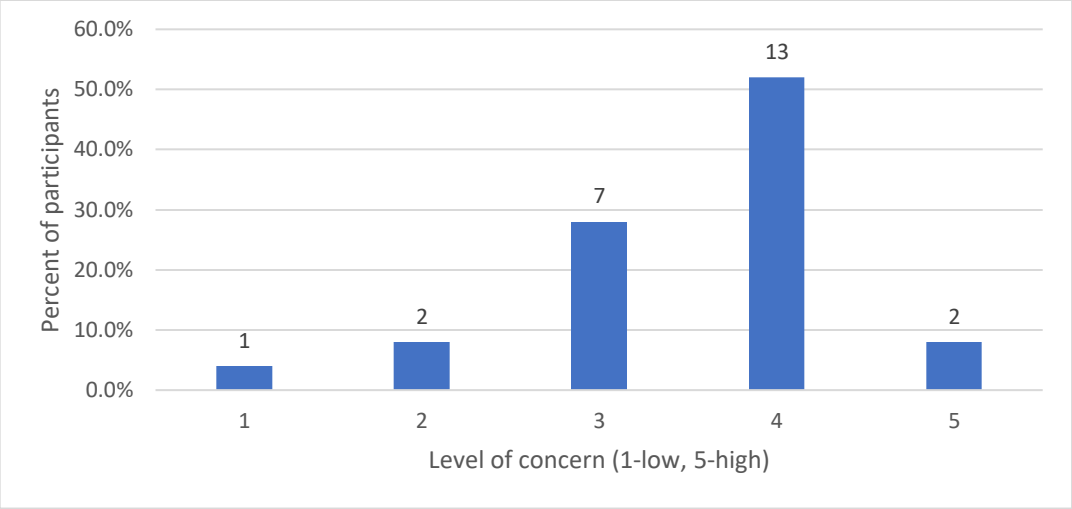

Figure 18: GCs' concern for the privacy policies of DTC tests.

On a scale of 1 (unconcerned) to 5 (highly concerned), the majority of GCs rated their concern as a 4, average score was 3.52, standard deviation was 0.92. Participant counts also displayed.

When asked where they perceive privacy issues in DTC tests, 84% (*n*=21) of GCs selected the sale of aggregated data, 72% (*n*=18) GCs selected the retention of samples, and 40% (*n*=10) selected the anonymization of genetic data. Only 12% (*n*=3) of GCs believed that DTC tests adequately safeguard personal genetic security, and one GC (4%) agreed that the federal *Genetic Non-discrimination Act*[28] and *Personal Information Protection and Electronic Documents Act*[98] ensure that DTC consumers' privacy is protected (Fig. 19).

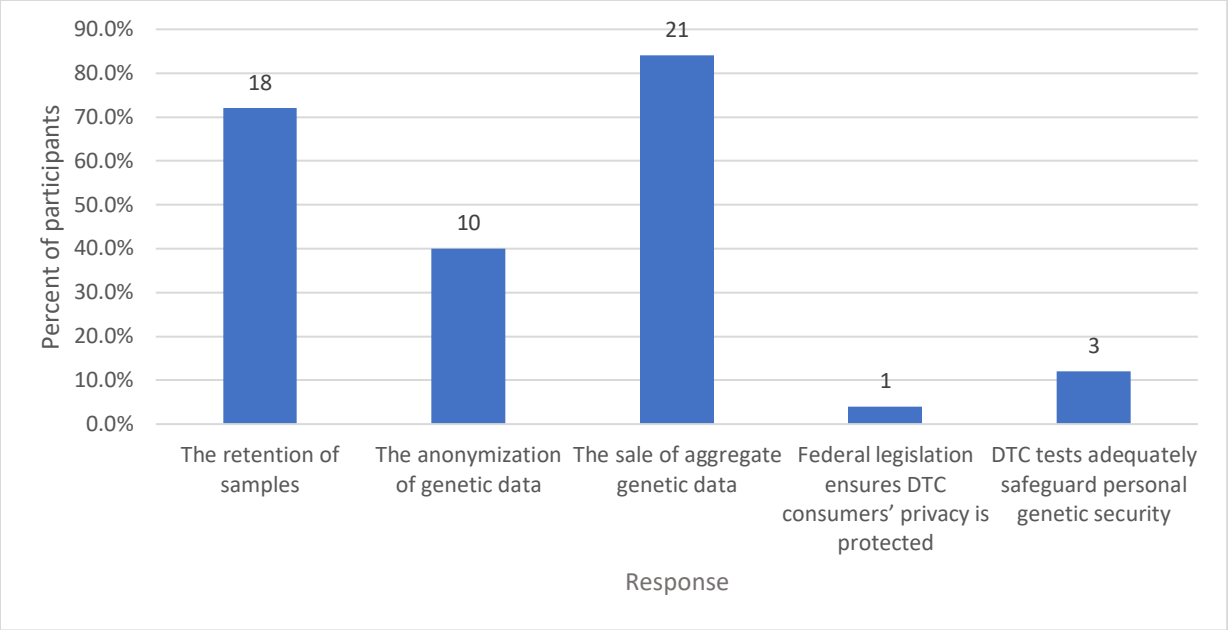

Figure 19: DTC test privacy concerns of GCs.

Most GCs were concerned about the sale of aggregate genetic data and the retention of samples. A minority of GCs believe the public is adequately protected from privacy concerns by federal legislation, and only one GC believes that DTC tests adequately safeguard personal genetic security. Note that participants were allowed to select multiple reasons, resulting in a higher count than number of participants for both categories. Participant counts also displayed.

GCs were also asked about what privacy protections they would like to see as a feature in DTC tests. 64% (n=16) GCs would like to see independent auditing of security features, 60% (n=15) selected company registration with a government agency as a change to implement, 56% (n=14) listed the encryption of data, and 44% (n=11) of GCs listed the aggregation of data as a privacy protection they wish to see in DTC tests. Only 12% (n=3) of GCs selected that privacy standards should be left to individual DTC company discretion (Fig. 20).

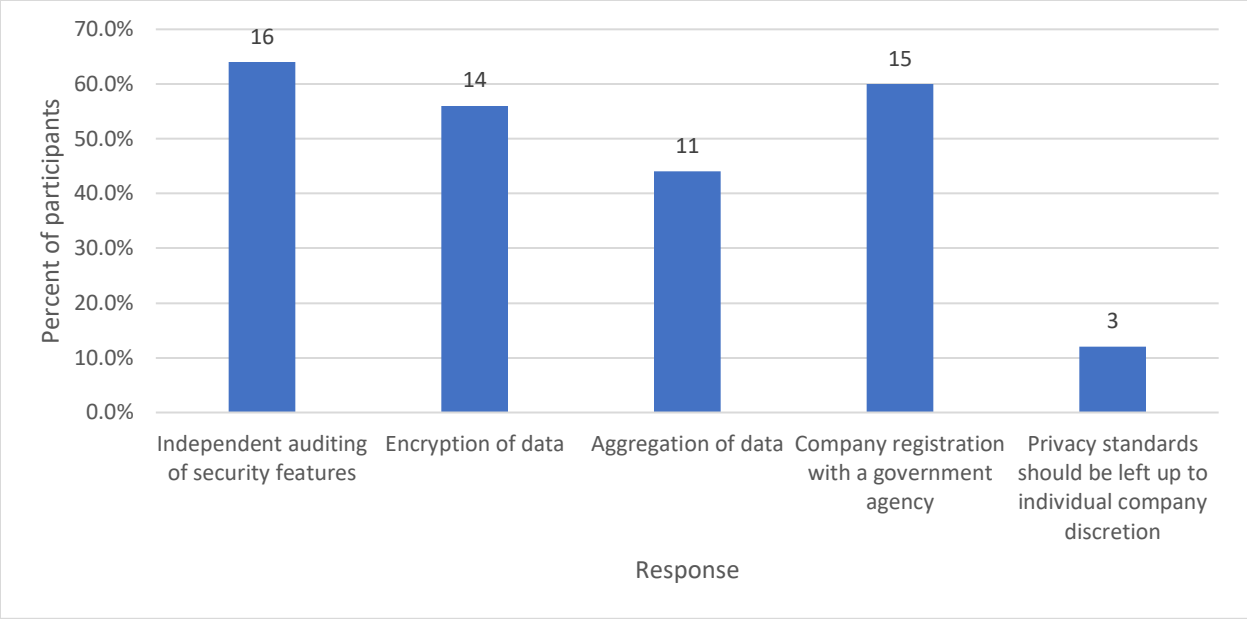

Figure 20: Privacy protections GCs would like to see in DTC tests.

Most GCs listed independent security audits, data encryption, and company registration as elements they would like to implement, and a minority of GCs believe that privacy standards should be left up to individual company discretion. Note that participants were allowed to select multiple reasons, resulting in a higher count than number of participants for both categories. Participant counts also displayed.

Asked whether they believe the profession of GCing needs to do more to protect the privacy of DTC consumers, 48% (n=12) answered in the negative, while 20% (n=5) answered yes, and 32% (n=8) were unsure (Fig. 11). Of those who responded no, 32% (n=8) of GCs said intervention was not feasible, 24% (n=6) listed that it was beyond the scope of GCing, and 24% (n=6) listed that intervention was a misuse of clinical time (Fig. 21). Of the GCs who responded that the profession needs to indeed do more to protect the privacy of DTC consumers, 20% (n=5) of GCs indicated that the lack of regulation of DTC company privacy policies is concerning, while 4%

(*n*=1) indicated that since informed consent is difficult to achieve without the mediation of a healthcare professional, privacy policies must be closely monitored (Fig. 21).

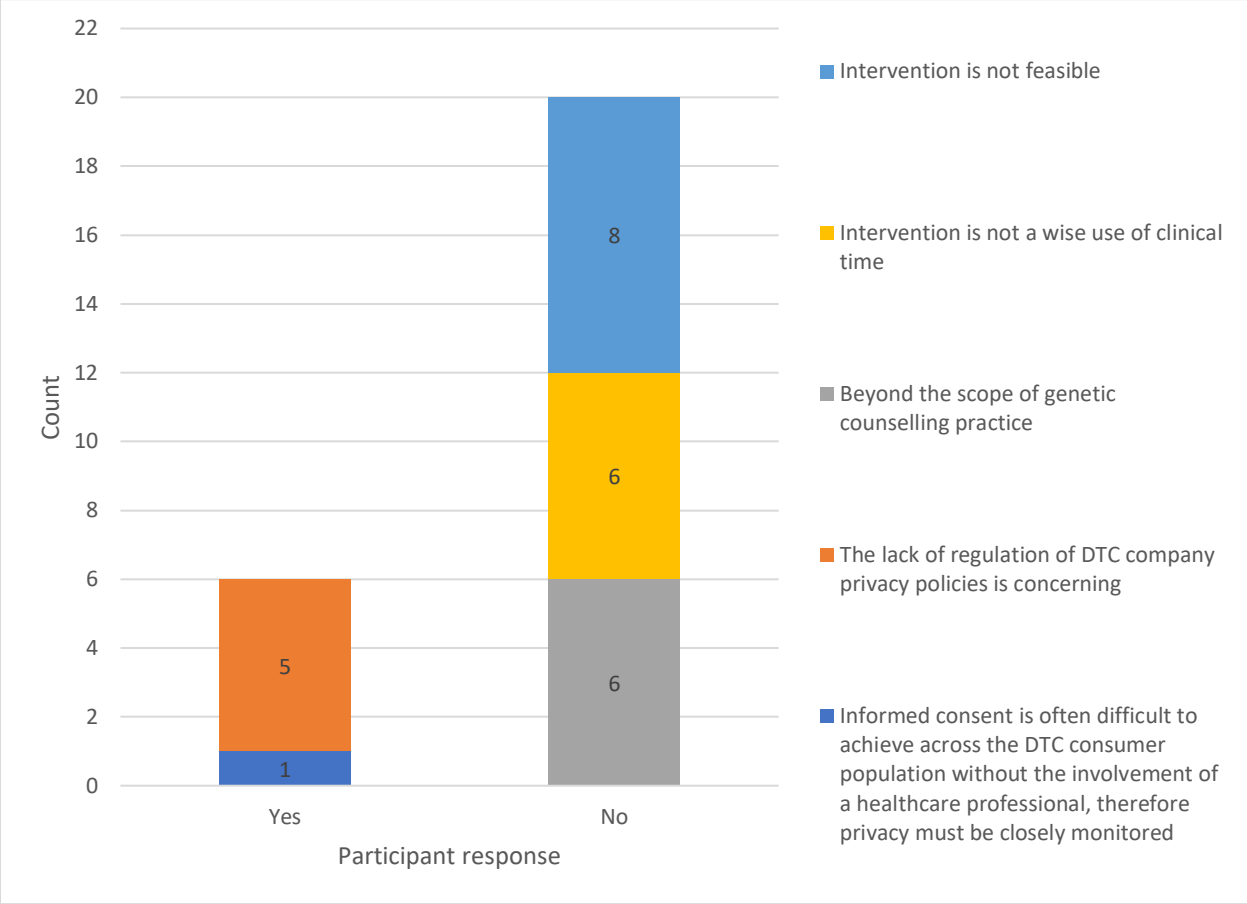

Figure 21: GCs do not believe the profession needs to protect the privacy of DTC consumers. *The majority of GCs do not believe GCs need to do more to protect the privacy of DTC consumers, while a minority of GCs believe GCs need to do more to protect the privacy of DTC consumers. Note that participants were allowed to select multiple reasons, resulting in a higher count than number of participants for both categories. Participant counts also displayed.*

When asked how the profession of GCing could respond to the privacy concerns of DTC tests, 76% (*n*=19) of GCs indicated that GCs could produce communication materials for the public, 68% (*n*=17) selected that GCs could collaborate with the Office of the Privacy

Commissioner of Canada to help educate the public, 36% (n=9) and indicated that GCs could advocate for reform through the CAGC. 20% (n=5) of GCs answered that responding to privacy concerns is outside the scope of the GCing profession (Fig. 22).

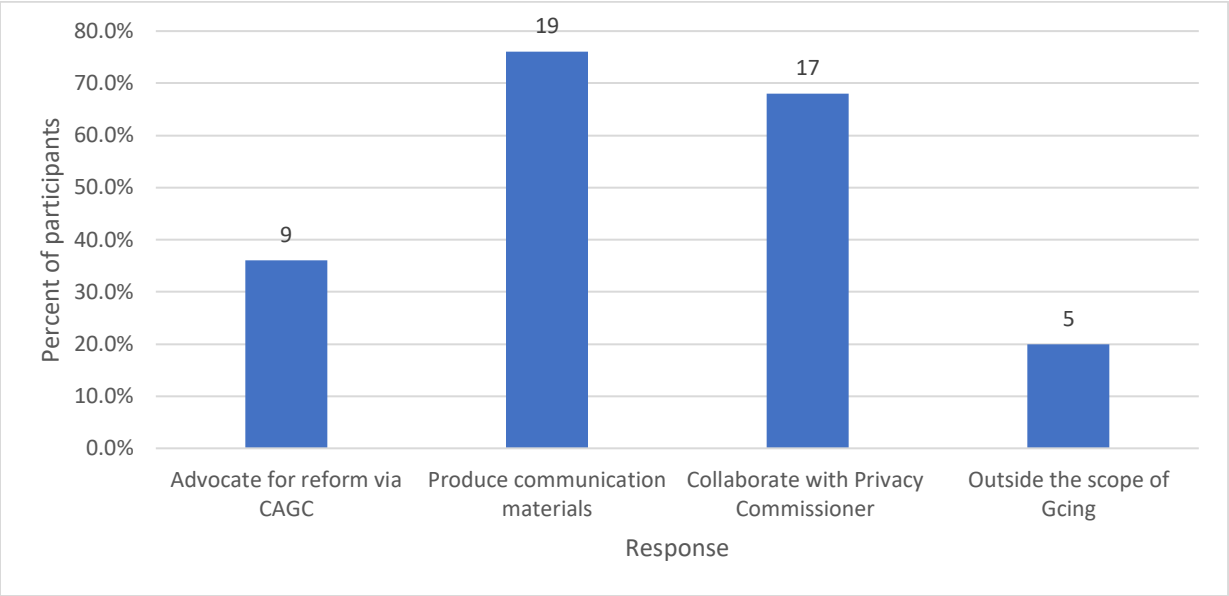

Figure 22: Possible actions GCs could take to respond to privacy concerns of DTC tests. *Most GCs responded they could produce communication materials or collaborate with the Office of the Privacy Commissioner to educate the public, as well as advocate for reform via the CAGC. A minority of GCs believe such actions are beyond the scope of the practice of GCing. Note that participants were allowed to select multiple reasons, resulting in a higher count than number of participants for both categories. Participant counts also displayed.*

The majority (68%, n=17) of GCs do not believe that the inclusion of DTC results in patients' medical records is likely to lead to genetic discrimination. 32% (n=8) of surveyed GCs did see this as a reasonable concern of DTC tests (Fig. 11).

4.5 Genetic counsellors' involvement in policy

GCs displayed a slight reluctance to involve the profession with patients who pursue DTC tests. When asked to rate how heavily the profession should involve itself on a scale of 1–5 (1—not involved, 5—heavily involved), the majority (36%, *n*=9) voted 3, 32% (*n*=8) selecting 4, 24% (*n*=6) voting 2, and only 8% of GCs (*n*=2) voting 5 (Fig. 23). The average answer was 3.24, with 0.93 standard deviation.

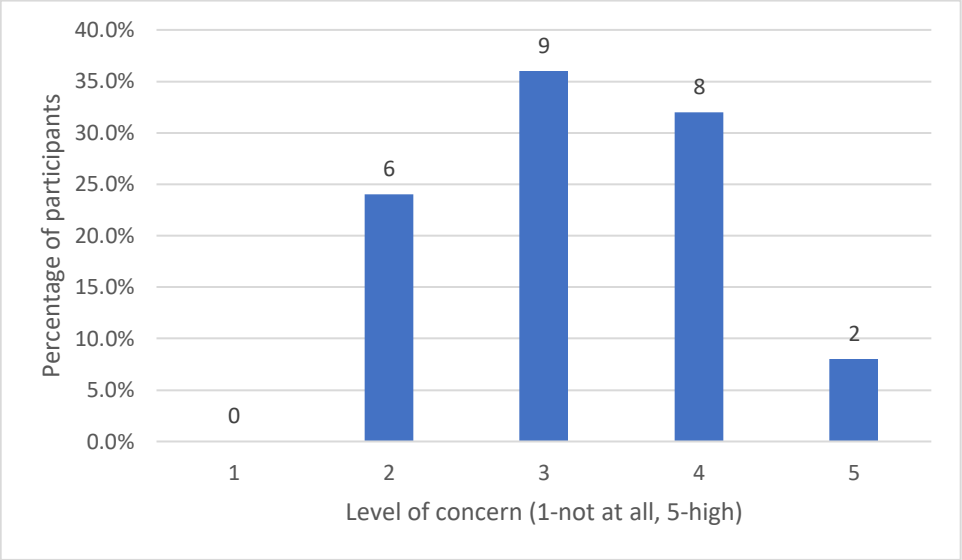

Figure 23: GCs' view on how heavily involved the profession should be with DTC tests and counselling consumers who pursue them.

On a scale of 1 (not involved at all) to 5 (heavily involved), the majority of GCs rated the potential involvement as a 3, average score was 3.24 with 0.93 standard deviation. Participant counts also displayed.

GCs were divided as to the impact that the lack of legal recognition as healthcare professionals has on their work counselling patients who pursue DTC tests. The majority of GCs (52%, *n*=13) do not view the lack of legal recognition as an impediment to their counselling work

with DTC consumers. 40% (*n*=10) GCs did see this lack of recognition as an impediment to counselling work, while 8% (*n*=2) refrained from answering (Fig. 11).

When asked to respond to the statement "DTC tests are sufficiently regulated", GCs were of a similar opinion; 76% (*n*=19) disagreed, while 24% (*n*=6) responded that they were neutral, and 0% agreed (Fig. 24).

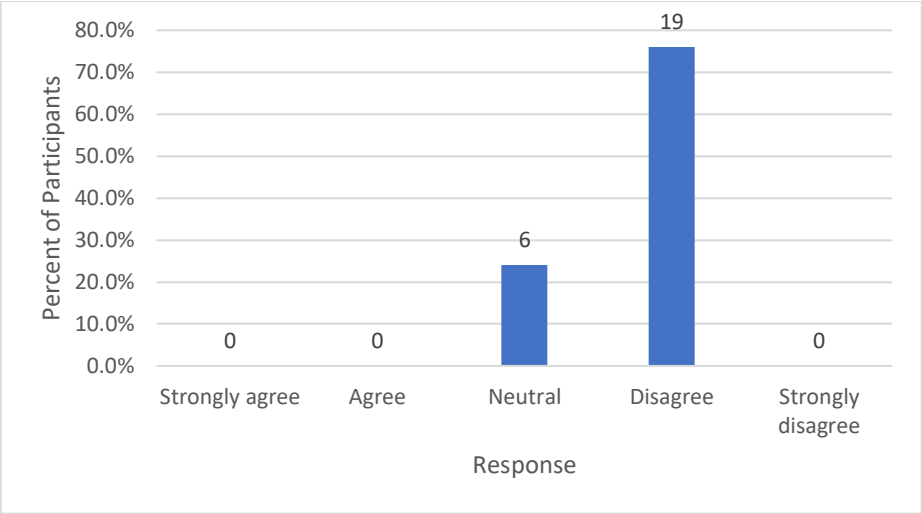

Figure 24: GCs do not think DTC tests are sufficiently regulated.

When responding to the statement "DTC tests are sufficiently regulated", the majority of GCs responded they disagree, while a minority were neutral. Participant counts also shown.

GCs held various opinions regarding how the CAGC could become involved with DTC evaluation. 20% (*n*=5) of GCs selected that the CAGC could grade tests via a list on their website, 12% (*n*=3) indicated that the CAGC could recommend tests through a list on its website, and 16% (*n*=4) indicated that the CAGC could accredit tests, similarly to how the Canadian Dermatology Association labels products. However, 56% of GCs (*n*=14) indicated that the CAGC should not attempt to evaluate DTC tests in any capacity, and 48% (*n*=12) selected that it is currently unfeasible for the CAGC to be involved in this process (Fig. 25).

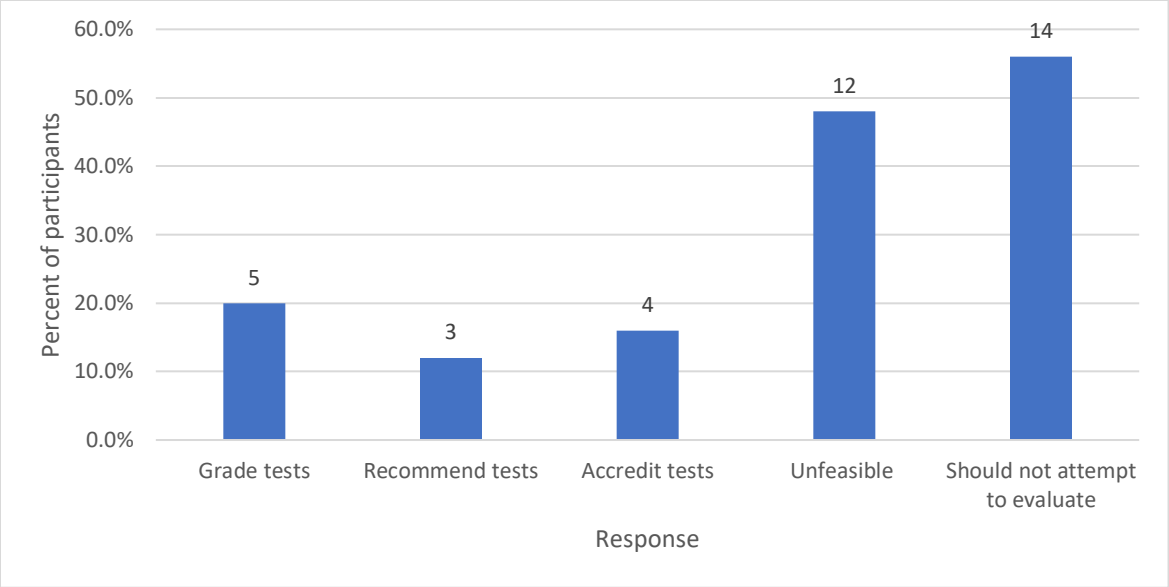

Figure 25: GCs are divided on whether the CAGC should attempt to evaluate DTC tests. *The majority of GCs selected that such actions were currently unfeasible, and that the CAGC should not attempt to evaluate DTC tests. Note that participants were allowed to select multiple reasons, resulting in a higher count than number of participants for both categories. Participant counts also displayed.*

The majority of GCs were in agreement (92%, *n*=23) that it is not feasible to offer pre- and post-DTC test GCing to patients who pursue DTC tests. Only 8% (*n*=2) of GCs believe this endeavour is feasible (Fig. 11). For those who voted no, 72% (*n*=18) of GCs responded that there are not enough GCs to offer this service, and the same number also responded that this should be the responsibility of DTC companies.

Asked whether offering counselling before or after taking DTC tests would potentially have the most impact on the consumer, 44% (*n*=11) GCs preferred to offer pre-counselling (helping to promote informed consent and educate consumers in advance of the test), 28% (*n*=7) of GCs indicated they would prefer to offer post-test counselling, to review results with consumers.

Seven additional GCs (28%) maintained that it is not feasible to offer any counselling to DTC consumers (Fig. 26).

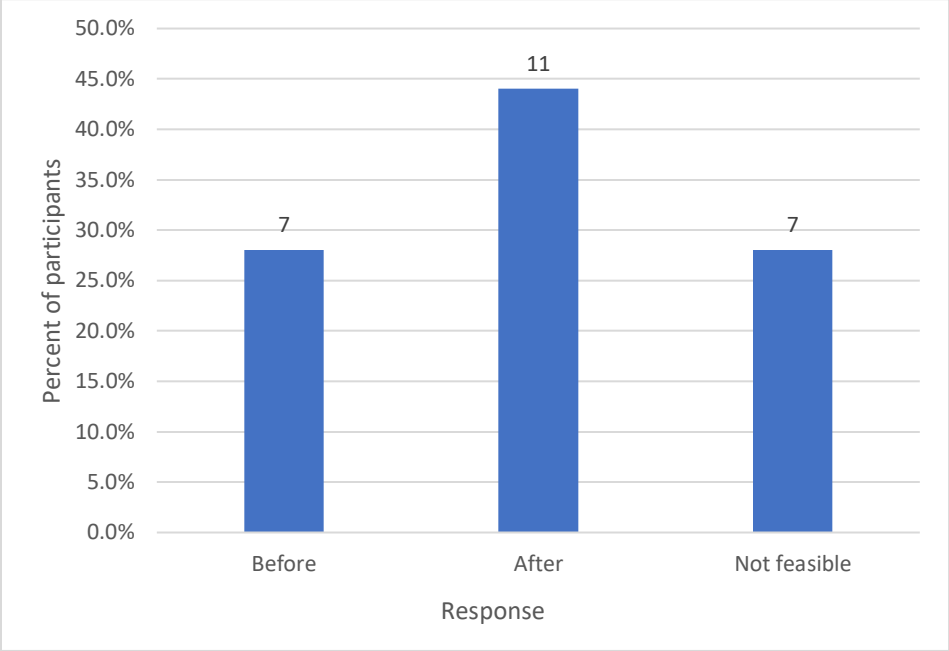

Figure 26: GC's views on pre- or post-DTC test counselling.

The majority of GCs view offering post-DTC test counselling to consumers as most beneficial to the consumers, while equal numbers of GCs preferred to offer pre-test counselling or that it was not feasible to offer any counselling to DTC consumers at all. Participant counts also shown.

GCs were also asked their opinions on alternative communication models of medicine, as the pandemic has caused healthcare professionals to adapt to fewer in-person interactions. Of these, 56% (n=14) GCs indicated they would like to use online resources managed by GCs to alleviate the burden of DTC test counselling, while 40% (n=10) GCs each responded they would like to utilize virtual conferences within their province and virtual conferences within the country. 40% (n=10) of GCs responded that it is not feasible to offer counselling to DTC consumers (Fig. 27).

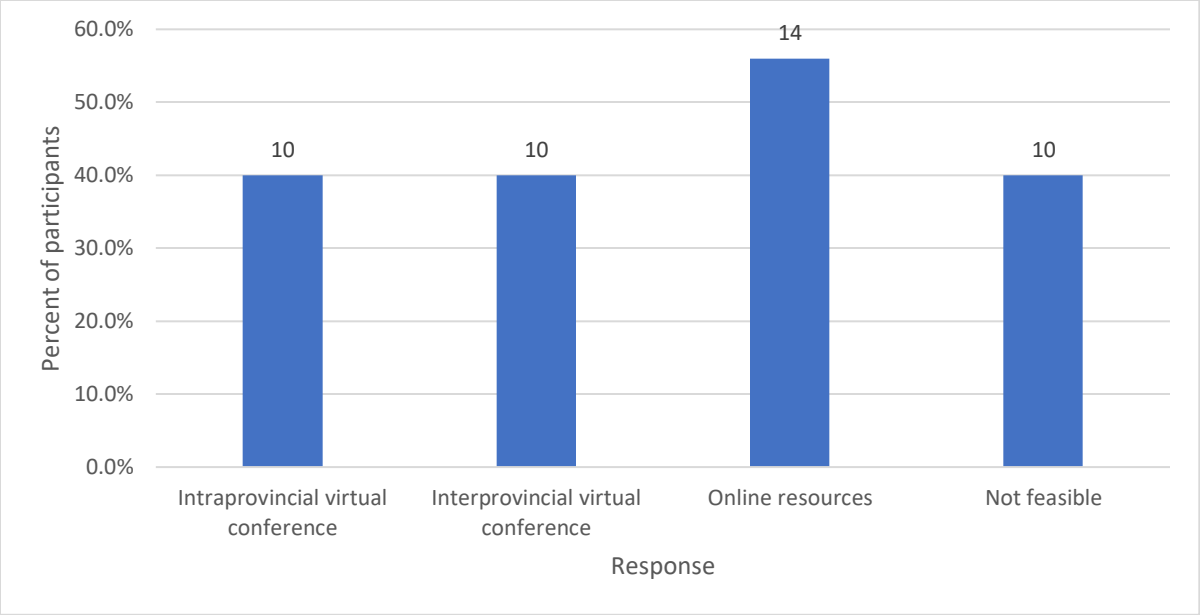

Figure 27: GCs' views on alternative communication models for counselling DTC consumers.

GCs see online resources as the best alternative communication models of medicine to help counsel DTC consumers, with many GCs also selecting intra-provincial and inter-provincial virtual conferences as viable, while some GCs also indicated that it is not feasible to offer counselling to DTC consumers.

In terms of what is currently feasible for the CAGC to offer, 68% (*n*=17) of GCs indicated that the CAGC could produce informative materials on DTC tests, 60% (*n*=15) of GCs selected that the CAGC could advocate for federal regulation of DTC tests (e.g., standardization of accuracy, consent forms, terms and service agreements, privacy policies), 48% of GCs (*n*=12) expressed interest in starting a Special Interest Group (SIG) at the annual CAGC meeting to develop a policy review of DTC tests, and 44% of GCs (*n*=11) indicated they would like to see seminars organized for primary care physicians or other healthcare professionals to prepare them to offer counselling to DTC consumers (Fig. 28).

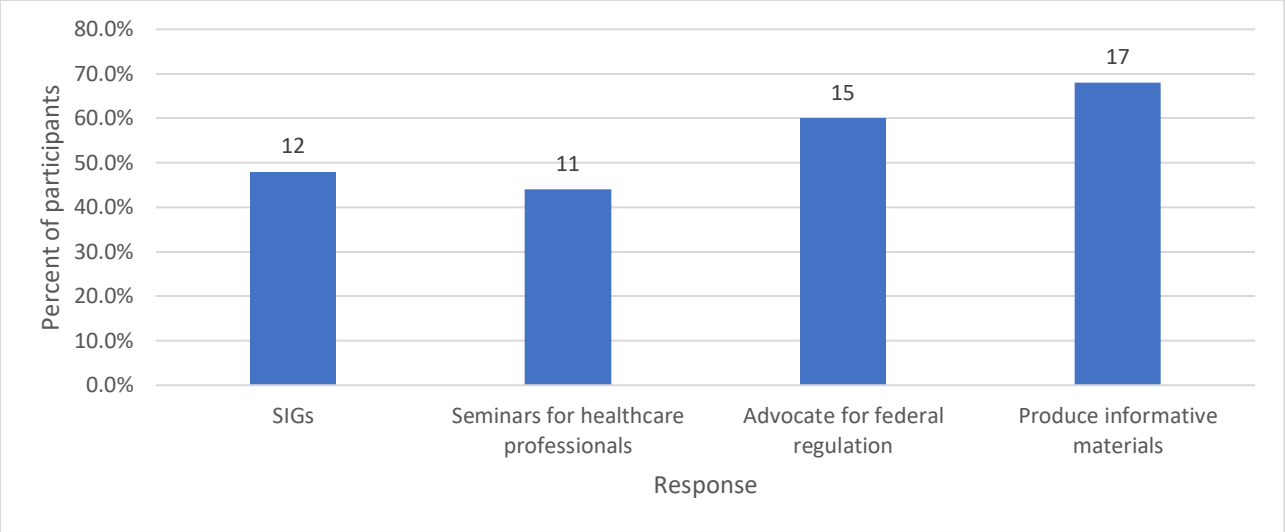

Figure 28: Feasible actions GCs would like to see the CAGC adopt to mitigate the high demand for counselling of DTC consumers.

Most GCs would like the CAGC to produce informative materials and advocate for regulation, while some GCs would also like to convene a SIG on DTC tests and organize seminars for healthcare professionals. Note that participants were allowed to select multiple reasons, resulting in a higher count than number of participants for both categories. Participant counts also displayed.

A recent study found that the majority of American GCs believe it is the responsibility of DTC companies to provide counselling to consumers, rather than clinical geneticists, and members of the CAGC responded unanimously (100%, n=25) that they agreed with this finding (Fig. 11). When asked why, 84% (n=21) of GCs responded that the DTC industry has sufficient resources to recruit GCs, and 60% (n=15) of GCs indicated that since the tests are not medically necessary, consumers do not merit access to the limited pool of GCs. 56% (n=14) of GCs responded that there are too few GCs in Canada to provide counselling, and the same number responded that the uneven distribution of GCs across Canada creates access issues. Finally, 40% (n=10) of GCs

responded that DTC consumers are not patients of the healthcare system, the same number responded that offsetting DTC counselling responsibility to clinical GCs helps the industry avoid accountability, and 12% (*n*=3) GCs responded that they do not view the counselling of DTC consumers as within the purview of clinical GCing (Fig. 29).

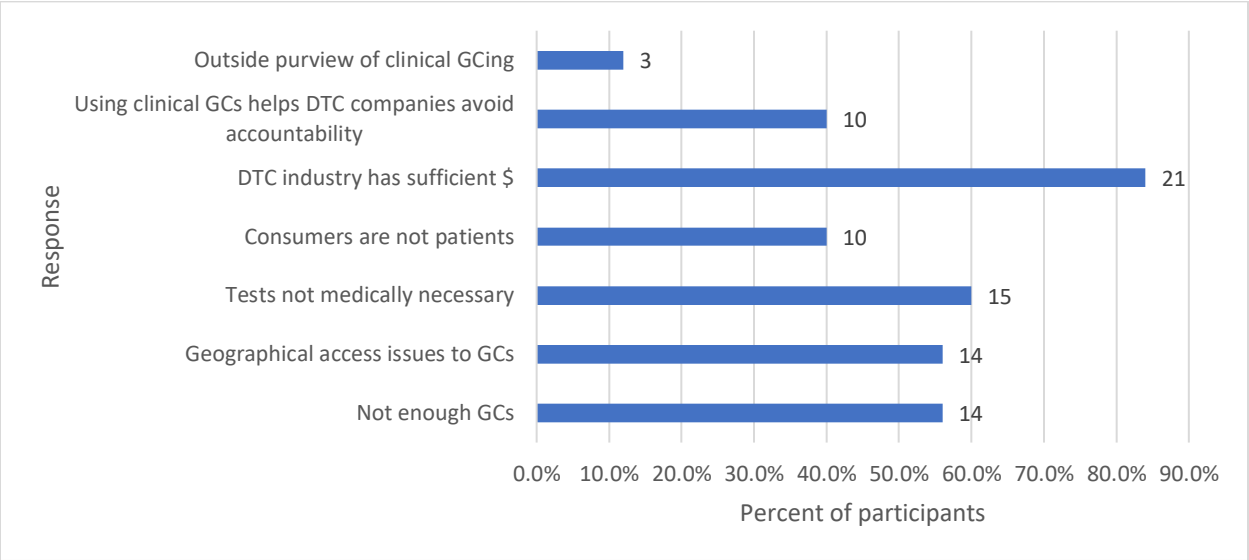

Figure 29: Reasons why GCs believe DTC companies should provide in-house GCing to consumers.

The majority of GCs indicated that the DTC industry has sufficient resources to offer. Note that participants were allowed to select multiple reasons, resulting in a higher count than number of participants for both categories. Participant counts also displayed.

Finally, GCs were asked how they see their role changing with regards to the DTC test industry. 60% (*n*=15) of GCs responded that they anticipate increased collaboration with primary care physicians, and 52% (*n*=13) responded that they anticipate increased collaboration with other healthcare professionals, while the same responded they anticipate increased work outside clinical roles, and the same number responded anticipating increased advocacy work. 24% (*n*=6) GCs

responded that GCs should focus on clinical patients, rather than patients who pursue DTC testing (Fig. 30).

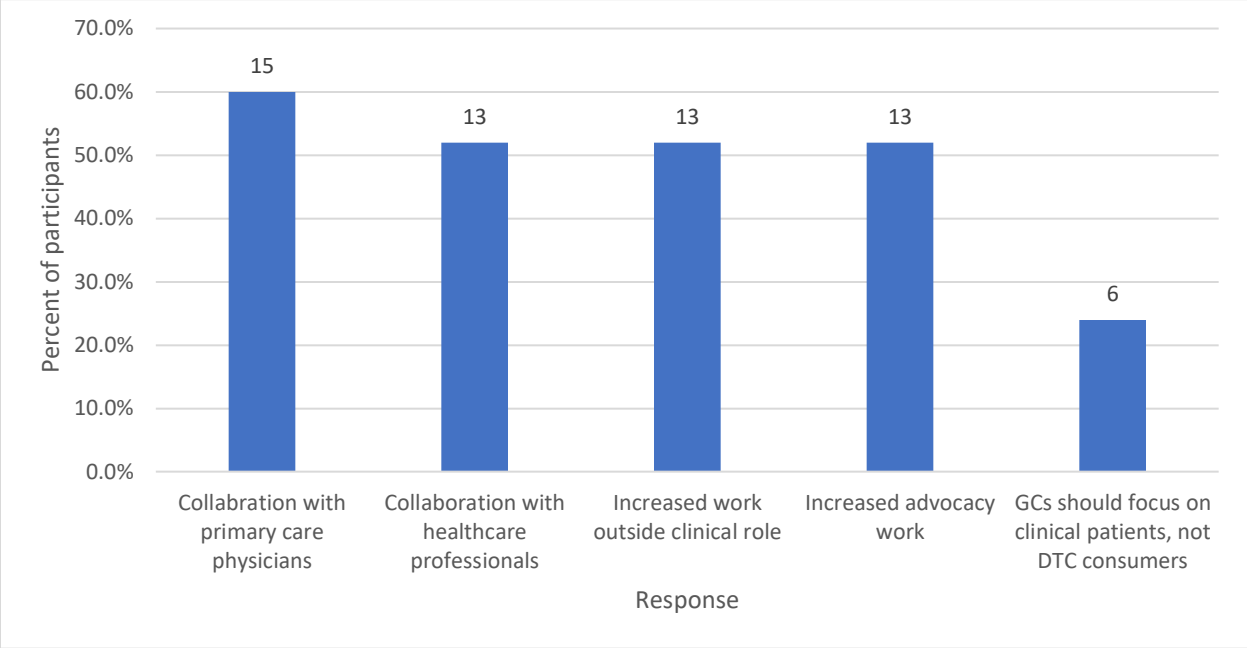

Figure 30: The changing role of GCs with respect to DTC consumers.

GCs anticipate increased collaboration with primary care physicians as the largest change to their role in the foreseeable future with the impact of DTC consumers. Participant counts also displayed.

Chapter 5: Discussion

5.1 Ethical concerns of DTC tests

DTC tests, at first glance, empower the autonomy of consumers who seek to understand some of the complex genetics underlying their health and ancestry. These tests grant one the ability to explore one's own health and identity in a way that was previously inaccessible to the public. As the latest form of personalized medicine, which seeks to tailor healthcare and interventions based on one's biology, DTC tests grant consumers the opportunity to implement diet or lifestyle

changes after receiving their genetic results, which is often conflated with increasing one's control over future health issues. True positive or negative results could allow individuals to better prepare, or act to reduce risks, before the onset of disease, or could offer emotional relief and lessen the number of checkups required for an individual from a family at high risk of developing a given disease, respectively[99].

For those who believe knowing one's genetic material promotes autonomy, the libertarian idea of moral right to self-ownership is a popular refrain. Using the libertarian concept that every person is the owner of their own powers and is able to use them as they please (provided they do not harm others), scholars elsewhere have argued that the genome is part of what a person is[100]. Therefore, every person is the moral owner of their genome, and is free to use it as they wish so long as they do not use their person and genome "aggressively against others"[100]. As such, every person should be free to take DTC tests so long as the tests are not performed by coercion of the test providers[100]. DTC test companies have themselves been arguing that people have the right to access their genetic data because such data is a fundamental element of one's body, identity, and individuality[73].

Underlying this argument is the idea that one's autonomy will be benefitted from having 'known' the genome through a DTC test, which is contested by the issues of scientific validity and clinical utility underscoring DTC tests, as previously discussed (Chapters 1.1 and 2.5)[4–6,17,79]. If all tests in Canada were guaranteed to provide accurate and useful results, one could plausibly acquire a more comprehensive understanding of one's health by knowing one's genetic health, and thus make more informed healthcare decisions. Unfortunately, this is not the case in Canada, where unregulated DTC tests do not necessarily provide accurate or evidence-based associations of

disease risk[4,6,8–13,19]. Given these concerns around the accuracy of DTC results, consumer autonomy and subsequent ability to make informed health choices may be challenged.

Furthermore, despite the rhetoric of self-empowerment that has emerged among DTC tests[7], the idea that DTC tests empower consumers to control one's future health is an "idyllic but entirely false vision", according to Schaper and Schicktanz, and is not evidence-based[101]. Studies have shown that genetic results are not consistently associated with diet and lifestyle changes[102], and only 27% of consumers report making a change in health behaviour post-DTC-testing, while no consumers ceased any previous high-risk health behaviours[103]. One meta-analysis of the literature reported that 12% of DTC consumers implemented improved dietary and exercise regimens in their lifestyles, while 19% quit smoking, totaling a 23% positive lifestyle change[1]. While these are promising initial results, the proportion of DTC consumers who implement positive health behaviour changes post-testing remains low. Therefore, DTC tests may not be so powerful in terms of informing one's decision-making abilities and autonomy, if the health behaviours of those who take DTC tests are largely unchanged. These values, however, may affect how consumers approach seeking healthcare for results interpretation and therefore may directly impact GCing.

While it is general knowledge that GCs are wary of integrating DTC results that are not validated, to our knowledge this is the first survey to explicitly interrogate Canadian GCs opinions on DTC tests and how their vocation and professional organizations could react to the emerging market. Of the surveyed GCs, the majority (84%, n=21) had experience working with patients who had pursued DTC testing, and thus the study results, while non-significant (see section 3.3), reflect the views of an experienced population. Indeed, such a number alone represents the growing normalcy of counselling DTC patients within the GCing profession. This level of experience

working with DTC consumers is a marked change from a 2011 study where only 14% of American GCs had encountered patients who requested interpretation of DTC results[104], and an increase from a 2016 survey in which 40% of American GCs had provided counselling post-DTC test[47]. This growth over time is consistent with the expanding DTC industry and raises interesting questions regarding the congruent expansion of public science literacy with DTC tests. However, such inquiries are beyond the scope of the current project.

Interestingly, of those who had worked with DTC consumers, GCs did not view their lack of professional legal recognition as an impediment; suggesting that GCs do not feel constrained by the lack of delegated acts in this area of their work. GCs working with patients who have taken DTC tests may enjoy working with a range of autonomy in their role, although in terms of ordering follow-up or validating results, only 13/25 GCs anticipated increased collaboration with healthcare professionals with the impact of DTC tests. This seems to imply that GCs do not prioritize follow-up genetic testing to DTC consumers, limiting their role to counselling and not clinical results validation.

Overall, this survey confirms the finding from Hsieh *et al.* (2021) that GCs hold negative views towards DTC tests[47]; only 56% of surveyed GCs believe patients who pursue DTC tests are within the professional scope of GCs. Indeed, all surveyed GCs were in consensus that DTC companies should provide in-house GCing. The most popular response was that the industry has sufficient funds to hire in-house counsellors, and that there were access issues and not enough GCs to provide counselling to DTC consumers. GCs also reported that using clinical GCs to counsel DTC consumers could help DTC companies avoid taking accountability for their services. This unanimous agreement that DTC companies should offer in-house counselling mirrors previous findings of a survey conducted through the NSGC[47].

In accordance with the above, the results of this preliminary study have demonstrated that most Canadian GCs are highly concerned about the privacy implications of DTC tests. GCs perceive privacy issues mainly around the sale of aggregate data and retention of samples, as well as the anonymization of samples. While many GCs expressed interest in implementing independent security auditing, company registration with a government agency, and data encryption, in contrast to their views on promoting informed consent, most GCs did not believe it is within their professional purview to do more to protect the privacy of DTC consumers. GCs reported that such actions would not be feasible, with a minority listing that it is beyond the scope of GCing and that such interventions would be a misuse of clinical time. Of note, the Office of the Privacy Commissioner of Canada maintains that DTC companies are subject to several Canadian privacy laws if they are located in Canada or have a "real and substantial link" to Canada[25]. Canadians are protected at the federal level by the *Personal Information Protection and Electronic Documents Act*[98], which applies to organizations that use, collect, or disclose personal information throughout commercial activities, and the provinces of Alberta, British Columbia, and Quebec all have provincial laws enacted to further safeguard the privacy of their residents[25]. Many organizations have also written about the necessity for privacy of genetic information, including the European Society of Human Genetics[83], American Society of Human Genetics[105], and the CMA[26]. These professional organizations have all stressed the importance of privacy regulations around genetic data, which could endanger consumers by threatening the integrity of sensitive information.

As for concerns regarding consent, GCs surveyed here do not believe the tests promote the informed consent of patients: terms and service documents poorly describe tests' benefits, limitations, and methodology to patients, and therefore fail to adequately disclose risk to

consumers. However, there is consensus in the literature that a healthcare professional should be involved with DTC consumers, despite GCs' opinions on their professional involvement. Among academics[5,6,9,11,45–47] and academic institutions[25–27,48–50] there have been many calls for healthcare professionals to be involved with DTC results. Surveyed public health professionals in Europe have similarly expressed that health professionals should be involved in the process of taking a DTC test[106].

Overall, GCs recognize the problems with DTC consent documents (in accordance with previous findings[104]) and the harm to informed consent that may arise from the lack of healthcare professionals mediating access to tests, but do not appear to agree on their professional duties in response and are wary of misusing clinical time and resources. However, there is a clear perception of need among the GC community for consumers to receive adequate information to give informed consent. The CAGC Code of Ethics urges that GCs provide, to the best of their abilities, autonomy to their patients in decision-making[51], but GCs themselves are divided as to whether DTC consumers should constitute patients under their care. GCs already provide post-DTC counselling to consumers on an *ad hoc* basis (i.e., when consumers are referred to their services or contact GCing clinics directly), but there exists no professional imperative to offer counselling to DTC consumers, nor to consumers of any other novel DTC genetic technologies.

These mixed sentiments were mirrored for discussions of consumer privacy; GCs do not think it is feasible nor a good use of clinical time to protect DTC consumer privacy at the individual level. Beyond being knowledgeable of privacy issues around the use, collection, disclosure and exchange of genetic information, the CAGC Practice Based Competencies[53] and Code of Ethics[51] do not compel GCs to take direct action on this matter. In the context of privacy protection, GCs already implement rigorous confidentiality measures in their clinical work, as mandated by the

CAGC Code of Ethics to "respect their patients' confidentiality in accordance with existing regulations in medical and research settings"[51], but according to this survey GCs do not believe any additional consumer privacy protection advocacy is warranted.

In terms of actionable change, however, GCs are content to take indirect action to improve consumer informed consent and privacy. They feel they are equipped with the necessary training and education to provide informed consent to people who pursue DTC testing, which is a marked change from previous surveys, which found that GCs do not feel comfortable providing counselling to DTC consumers due to a lack of knowledge (59.7% of n=482, self-reported)[47]. GCs are also amenable to producing more education materials for promoting public education on DTC tests, drawing again from the CAGC Code of Ethics and potentially in collaboration with the Office of the Privacy Commissioner. GCs are also interested in advocating for privacy reform through the CAGC. GCs are well versed in communication, and are highly trained in psychosocial aspects of counselling, making them well positioned to produce such materials for the broader public's use, and collaboration with governmental bodies or consultation with DTC companies directly could be an elegant solution to some of the major privacy concerns GCs perceive in DTC tests. With their deep understanding of the technical and psychosocial consequences of privacy breaches in handling sensitive genetic data, GCs are uniquely trained to be of service in this area. Indeed, a systematic review of the European general publics' view of DTC tests found that educational materials and programs would be beneficial towards increasing the capacity of individuals to make informed health decisions based on genetic information[107], and so enlisting GCs to produce such materials could similarly benefit Canadians.

An interesting theme that emerged out of the data was the implication that people who pursue DTC testing do not merit attention since the tests were not medically ordered; n=15/25

selected this response. This is an interesting finding, as it seems to conflict with the health-empowering, autonomous nature of DTC tests. Designed to allow the public unmitigated access to genetic health data, and possibly allow for faster response than the long waits often associated with public healthcare, there is an inherent tension here between the possible earlier diagnosis a DTC test could prompt, and the apparent value-judgement of GCs in preferring to treat patients of physician-ordered tests. GCs are heavily implying that patients outside the medical establishment do not deserve the time and attention of healthcare professionals. However, it is a well-known fact that access to GCs, and broader healthcare resources at all, is often difficult for consumers living in rural parts of Canada, or for those facing financial barriers[43,44], and DTC tests may be the easiest point of access to genetic data for these populations. While it may be acknowledged by GCs that DTC tests are often used recreationally and do not always warrant GCing services, this does not preclude the reality that DTC tests may serve as a practical tool for alerting consumers to possible health concerns that merit discussion with a healthcare practitioner. There is a need here, then, for operationalizable guidance for GCs to rely on to help them decide if a given DTC consumer warrants the use of scarce healthcare resources such as access to GCing or follow-up validation tests.

This attitude of GCs toward DTC consumers raises additional questions given the insensitivity of DTC test results towards POC (People of Colour); DTC algorithms may be insensitive to differences between racial and ethnic groups which can contribute to the vague or inaccurate return of results to POC consumers[12,13]. DTC companies rely on their own specific sample populations to develop algorithms[12] usually based off of homogenous nondiverse (White) datasets[37,108], creating a negative feedback loop and resulting in White populations being overrepresented in the training data, and biasing the algorithm towards White genetic biomarkers.

In the field of research ethics, generalizing results beyond the sampled population of a particular study to an unsampled population is known as construct or external validity[109], which, in the DTC context, is strengthened by diversity in sample recruitment. However, these proprietary algorithms are a symptom of a much larger problem concerning diversity in the field of genetics research: the systematic under-representation of POC in genetic research is a clear failure of the field to produce generalizable knowledge, and limits diverse populations' access to the benefits of precision medicine[37]. Inconsistencies in data collection, missing classifications and inconclusive results also stem from the issue of a lack of standards or guidelines for collecting and studying diversity data in genetics research[108]. This sampling bias within the entire field effectively means that although scientists have identified genetic associations with diseases, if there are genetic variants in susceptibility across different populations, researchers are unable to even stratify the data and perform subgroup analysis. Therefore, DTC tests which offer genetic health risks for such diseases may have a built-in blind spot for consumers of colour.

With emphasis on the principle of justice and the goal of equal access to medical services in the CAGC Code of Ethics[51], GCs may need to re-evaluate this apparent dismissal of all non-medically ordered genetic tests, since DTC tests may cause confusion and anxiety in population groups that are historically underserved by the medical establishment[110].

All this is not to say that all DTC consumers should be seen by GCs; such a mandate is not only impossible given the limited availability of GCs but is also an impractical use of their time and resources. While GCs have many valuable skills to leverage in the DTC dilemma, mandating that they consult with all DTC consumers is beyond the carrying capacity of the vocation and would offset responsibility from DTC companies, thus enabling poor DTC company practices

while overburdening an already strained healthcare profession. For further discussion of the appropriate updated roles of GCs, see section 5.2.

As well, GCs have good reason to employ the threshold of medically ordered tests when deciding whether to accept patients. There is consensus in the literature that DTC tests lack scientific validity[4,10–14] and the tests could easily return false positive results, leading anxious consumers to waste valuable resources in seeking confirmatory medical tests. See section 5.2 for additional discussion of this topic. It is also important to consider that this was a close-ended survey, and this response did not originate from GCs, but rather was selected from a predesigned list of responses.

As an interesting final note from the survey, GCs surveyed here do not anticipate that the inclusion of DTC results in medical records will lead to genetic discrimination. This finding is in apparent opposition with the literature, where scholars have noted that the discrimination inherent in DTC tests through insensitive algorithms leads to inaccurate or misleading information which should not be consulted by healthcare professionals for diagnostic purposes[111]. While not directly leading to genetic discrimination, since DTC tests carry questions of scientific validity[10,12,13], consulting such data in healthcare decisions or even treatment could lead to substandard care in racialized populations and thus, indirectly, genetic discrimination. This has grave potential for harming individuals and racialized populations when health decisions are made from misleading or inaccurate genetic data[111]. GCs offering counselling to POC DTC consumers would be a powerful impediment against such alarming consequences.

Despite the professional hesitation to engage with patients who have pursued DTC testing, GCs hold the most potential for aiding this novel patient group. The literature review indicated that DTC consumers have the potential to overwhelm healthcare providers, and physicians have

reported a lack of confidence counselling DTC patients[112] with their limited training in genetics[43]. Indeed, recent studies have reported a 26% error rate in physician interpretation of DTC genetic results[80]. An error rate of over 25% remains concerning and would hold serious consequences for patients if diagnoses are delayed and necessary confirmatory tests are withheld. In contrast, GCs are uniquely trained in psychosocial counselling as well as genetics to hear and address the emotional values and technical questions that may emerge during the DTC odyssey, and to refer patients to physicians for clinical validation if necessary. In light of these factors, GCs are the leading healthcare provider with the knowledge and psychosocial skills to counsel DTC consumers.

Overall, results from this survey suggest that GCs remain fundamentally divided over several key issues regarding consumer informed consent and privacy, and that GCs additionally remain divided over to the appropriate role individual GCs and the GCing profession ought to play in this age of unprecedented public access to novel biotechnologies. Revisiting the Traditional Duties of GCing may contribute some much-needed clarity on this matter to distill exactly what the role of GCs ought to be in the DTC era.

5.2 Points to consider: Genetic counselling traditional duties and the DTC counselling session

In light of these survey findings, ideas around GCing and DTC consumers must be revisited. Notably, the CAGC recommends that DTC consumers seek pre- and post-DTC test counselling[27]. However, there is dissonance between the CAGC recommendation and the results of this survey, wherein GCs reported they have adequate training to counsel DTC consumers but lack the appropriate resources to be able to offer individual counselling to these additional patients. To reconcile this tension, and to shape the discussion of counselling in the context of DTC consumers, let us revisit the Traditional Duties of GCs, as introduced in section 2.3. Note that for

the purposes of this discussion, DTC consumers are referred to as 'patients' once they seek GCing services.

The overarching theme to this discussion is, of course, the low number of GCs currently practicing in Canada. GCs are few in number and are overworked and burned out after demand has risen exponentially for their services over the last few years with the rise of genomics in healthcare. It is this fundamental fact that must guide any discussion of GCing services in Canada, and the survey findings presented here are discussed in light of this reality.

To preface, DTC consumers present novel challenges to Canadian GCs who chose to offer them counselling. DTC consumers risk receiving distressing results that could identify unexpected family relations or ancestry information, or even anxiety-inducing health information with unclear clinical utility or unconfirmed gene-to-disease-phenotype correlation[21]. Indeed, non-profit organizations have sprung up to support consumers who have received distressing ancestry test results[113], and third party apps and websites are emerging to help consumers 'demystify' DNA results[114]. Ergo, GCs amenable to counselling DTC consumers will have to adapt their services to meet the needs of this distinctive patient group as they strive to meet their professional obligations.

As a foundational principle of GCing, The Duty to Inform is defined as providing non-directive information and risks on the diagnosed condition, as well as treatment options, consequences, risks, and alternatives, and other patient support groups/organizations. In the DTC context, the GC Duty to Inform should not be updated to include any additional duties for patients who have pursued DTC testing; most GCs do not wish to further engage with DTC consumer informed consent or pre- and post-test counselling of DTC consumers and to force all GCs to accept the additional patient load would be too great a burden to impose on the already overworked population.

While the Duty to Inform as a theoretical Duty should not be changed in light of DTC consumers, in practice GCs must still uphold the fundamental values of this Duty if they choose to offer counselling to DTC consumers, as they would for any other patients. However, DTC patients may present unique challenges to this Duty of GCing.

In terms of the actual counselling session, GCs may feel they are unable to satisfy their Duty to Inform when pre-counselling DTC patients. GCs offering DTC patients pre-DTC test counselling would be forced to contend with the lack of available information on each tests' clinical validity and utility, as DTC companies rarely provide detailed information on their methodology. The same logic holds true for the Duty to Inform in regard to the privacy issues of DTC tests; GCs would be unable to control the privacy policies of DTC companies, stalling counselling efforts. The privacy policies may be unclear, vague, or entirely absent, which challenges what GCs can counsel patients on in terms of how the patient's genetic data may be stored, used, or shared to third parties, thus undermining the GC Duty to Inform. Therefore, the GC Duty to Inform, and to provide informed consent and privacy information to patients is challenged by DTC tests.

Additionally, GCs would encounter tension with the Duty to Inform when offering post-DTC test counselling on the return of results. DTC companies have varying policies on the return of incidental findings; policies can be vague or even entirely absent, and some companies may return incidental findings to anxious consumers ill-prepared to deal with the information. Given the varying methodologies and coverage[19] of some of the tests, and even the availability of this information, GCs may struggle to counsel anxious patients on results that may be false positives, or provide warning of false negatives in this context. Without further medical verification of suspicious test results, GCs may feel their Duty to Inform has not been satisfied. GCs will thus

have to use their professional judgement to decide if follow-up clinical validation is required, which will be impacted by availability of resources in the given health jurisdiction and will potentially incur additional costs for the health system[7,10,17,18]. Since false positives and negatives are common[14] and can severely impact an individual's medical choices and health behaviours[9,10], GCs who work with patients who have pursued DTC testing will require additional support in their provision of care.

DTC tests additionally challenge the GC Duty to Inform through obfuscating results interpretation for POC. GCs who provide counselling to patients who pursue DTC testing will need additional support in how to address the sensitive topics such as the racial bias in tests when counselling POC patients with confusing or vague results, stemming from a lack of construct validity and sample diversity in genetics research[37]. GCs may not be adequately trained to cover discussions of ethnic limitations or racial bias of DTC tests. In *A Gide to Genetic Counselling*, a foundational textbook of the field, the authors discuss "multicultural counselling", and make clear that diversity is cultural, not biophysical[61]. The chapter discussion is centered around the diverse values, assumptions, and beliefs of patients, and how these may impact the counselling session[61]. While this broad discussion provides useful context for many instances in medicine, as the field of genomics faces epistemic and construct validity challenges in terms of a lack of diversity in studied populations[37], GCs need to be aware of the consequences this may hold for genetic testing and DTC tests. Diversity, inclusion, and non-discrimination are vital for the ethical distribution of healthcare services and are receiving much-needed attention from the biomedical research community right now, and this will affect how GCs talk about these issues with patients.

In sum, the definition of the Duty to Inform as a theoretical Duty of GCing should not be updated to mandate GCs to counsel DTC patients to avoid overburdening the profession. However,

for those GCs who voluntarily chose to counsel DTC patients, this Duty will present novel, but not insurmountable, challenges in operationalization.

Conversely, the GC Duty of Competence, as a defined theoretical Duty of GCing, has potential for modification in light of DTC patients. This Duty describes the essential skills of the GC profession, as defined in the knowledge-based[55] and practice-based[53] competencies[52]. Since GCs have here reported confidence in providing informed consent to patients who pursue DTC testing, the Duty of Competence could be expanded to include staying abreast of innovations in DTC genetic tests, or perhaps even DTC biotechnologies at large. Integrating DTC genetic tests in this Duty would expand the knowledge competencies of GCs and would ensure all GCs are prepared to counsel DTC patients at any stage in the DTC odyssey, without mandating all GCs to offer such services. An expanded Duty of Competence could fall within the GC Knowledge-Based Competencies under Unit 3: Molecular Genetics, where GCs are expected to:

> Understand clinical molecular testing techniques including indications for, limitations of and the types of mutation they identify. Examples include, but are not limited to sequencing, panels, whole exome sequencing, MLPA, and linkage analysis[55].

Amending this Duty would ensure that GCs in Canada are adequately prepared to offer counselling to DTC consumers if they chose to expand their practice, but would not force GCs to offer counselling, and would therefore represent an elegant solution to the availability of counselling services for DTC patients.

The Duty of Confidentiality, while extremely important for preserving the privacy of GC patients, is not particularly relevant to discussions of DTC patients as GCs would need to respect this obligation regardless of the patient.

Returning to the practical counselling of DTC patients, GCs broadly must be prepared to adapt to DTC patients' specific values. Unlike patients of the medical system seeking counselling to inform future genetic tests or to discuss results of medically accredited tests, DTC patients may approach the counselling session with unfounded beliefs about what information the tests can provide. GCs must be prepared to discuss the limits of DTC tests to potentially frustrated and confused patients, as the debate over whether DTC tests empower patients' autonomy to make informed health choices or change health behaviours may affect how patients who have pursued DTC testing approach the counselling session.

Additionally, the GCing profession may need to reassess the provision of in-person care given the rising number of DTC patients. To address the accessibility concerns of GCing, this survey interrogated GC's views on virtual conferences. Here, a minority of GCs reported interest in providing intra- and inter-provincial virtual conferences (which would have no legal consequences given that GCing is not a licensed profession), while an equal number listed any such action as not feasible. Within the literature, there is debate around implementing tele- or electronic-based counselling, which could open access to genetics services to rural populations, help triage available GCing resources, and decrease costs while in theory reducing wait times for medical decisionmaking[115]. In contrast, it has also been argued that offering remote GCing services carries with it the potential to increase stress and anxiety over results, increase potential distractions and misunderstanding, and lead to inefficient counselling as GCs miss patient nonverbal cues and face additional challenges in providing emotional support[45,115]. However, to meet the needs of a rising DTC patient population, virtual counselling options may be critical for ensuring access to GCing for the appropriate patients to receive care.

When directly asked at what stage in the DTC odyssey they would prefer to offer counselling, GCs reported a preference to provide counselling after receiving DTC results, presumably to help with results interpretation (ranked as the leading concern of patients who had sought DTC testing). However, I contend that preemptive action to promote informed consent could help reduce the number of DTC consumers seeking results interpretation by promoting consumers' knowledge of testing risks and benefits before they take a DTC test. This work, stemming from the theory of harm reduction, could intercept downstream consumer anxieties before they manifest through anticipatory education, and would thus represent an efficient use of GCs' limited time and resources.

To satisfy this goal, in lieu of individual-oriented consent processes, perhaps alternative media forms, such as short videos or more graphic representations of the ethical and scientific concepts embedded in DTC tests would attract consumers' attention and promote informed consent. After reading/viewing these materials, consumers could be asked to take a quick quiz to demonstrate their understanding as evidence of informed consent before finalizing the purchase of a DTC test. These educational materials could also be adapted for the type of DTC test being purchased; the amount of material needing to be viewed and length of the quiz could be stratified by seriousness of the medical risks interrogated by the test, similarly to how the FDA ranks DTC tests for review based on the medical purpose risk, and the likelihood of results impacting medical care[29].

This could be an area in which the CAGC or individual GCs could consult, to help design educational materials for both the public and other healthcare practitioners that adequately convey technical concepts and prepare consumers for the implications of test results. Results of this survey suggested that GCs would be open to producing educational materials on DTC tests and

collaboration projects with the DTC industry to promote informed consent, and the funds resulting from such collaborations could be further used for GC research or undertaking initiatives relating to GC professional recognition. While there is no established industry standard for informed consent, Canadian GCs have the knowledge, training, and expertise in communication, counselling, and genetics to guide the development of informed consent materials for DTC companies as a primary mechanism to improve the pre-DTC test consent process. When asked to rank what they think their overall engagement with the DTC industry ought to be, 76% of surveyed GCs indicated interest in being involved, and so these measures could very well be a feasible first step.

While GCs surveyed here do not support a direct DTC consumer counselling role for their profession, in the literature, Wade and Wilfond (2006) suggested that GCs do have a duty to interpret DTC test results. These authors suggest that the duty of care GCs owe to their patients should extend to DTC consumers, which includes referral to other physicians or specialists[116]. In their view, any DTC consumer who requests counselling after taking a test should be seen by a GC, or a physician in the case where the GC does not feel confident providing results interpretation[116]. In the United States, using the NSGC Code of Ethics, others have similarly argued that GCs have a professional duty to offer pre- and post-genetic test counselling under the premise that the NSGC strives to be the healthcare resource that members of the public access for questions about genetic testing[117]. Ideally, this would be the case in the Canadian healthcare context, and the CAGC does call for pre- and post-DTC test GCing[27]. However, after over two years of battling COVID-19, combined with the small number of practicing GCs, Canada's healthcare infrastructure is extremely fragile and not equipped to deal with a high influx of DTC

patients if such a counselling duty were to be codified. GCs themselves do not see this as their responsibility nor their obligation, as demonstrated by the survey results.

Regardless of the changes DTC tests may or may not bring to the traditional duties of GCs, these and other emerging biotechnologies will continue to enact changes in the role of GCs and other healthcare practitioners. Here, GCs report increased collaboration with primary care physicians and other healthcare practitioners, increased work beyond their traditional clinical boundaries, and increased advocacy work as a direct consequence of DTC tests. With their unique combination of psychosocial counselling knowledge and genetic expertise, this population of Canada's healthcare workers are ready and able to meet the challenges of DTC tests, and lessons learned will be of use for other factions of the healthcare sector as biotechnologies continue to emerge.

5.3 Novel considerations

The GCing literature, while rich with analysis of DTC tests, has left one very important element largely untouched: the impact of DTC tests on Indigenous peoples. These tests present new challenges to Indigenous identity and sovereignty and raise important questions around Indigenous access to GCing services. Here too, the GC Duty to Inform could be adapted to ameliorate the situation and potentially provide much-needed support to communities.

The systematic oppression of Indigenous peoples by the Canadian state through the forces of colonization, assimilation, relocation, and globalization has resulted in the degradation of the cultures, knowledge, and traditional knowledge systems of Indigenous peoples[118], and created a deep distrust for Western science and the medical establishment. Historically, Indigenous bodies were treated abysmally by knowledge-producing cultures and practices which still shape the way

research is conducted today[119], and these atrocities have left a lasting impact on the way many Indigenous communities interact with modern scientists and the medical community.

One of the main functions of DTC tests is to provide consumers with a quantifiable, evidence-based identity. However, Indigenous researchers take issue with the premise of using DNA as a basis to assign identity and citizenship as this Western construct only complicates the problems stemming from biology, White supremacy, slavery, and dispossession that caused the issues in the first place[119]. Indigenous communities favour a self-defined identity as an expression of sovereignty and self-government[33], and so DTC companies attempting to define Indigeneity through genomic tests represents a radical alteration to the definition of Indigeneity[33]. Indeed, researchers have described how conflating genetic identity with Indigenous identity creates issues for Indigenous identity claims and thus challenges the existing historical and legal foundations of Indigenous governance authority[119]. Scholars elsewhere have coined the term 'biocolonialism' to describe this contemporary form of colonialism over Indigenous sovereignty and identity[120]. With the commercialization of these tests, Indigenousness is constructed and reinforced by the lens of capitalism, and the Indigenous genetic identity is controlled by DTC companies.

Indeed, the creation of an Indigenous haplotype is in itself a paradox; scholars have noted how 'Native American DNA' emerged out of the colonial history of Western settlers labelling the entire continent of pan-racial peoples as 'different' from the settlers and is based on the notion of the 'purity' of the Indigenous genome, pre-colonization[119]. However, assuming there was "a moment, a human body, a marker, a population back there in space and time that was a biogeographical pinpoint of originality" inherently contradicts the theory of evolution which underlies the entire field of genetics[119], and recent studies have found that various waves of traveling populations in fact settled on the North American continent over different time periods[121].

Thus, Native American DNA is an object that is defined by, and yet functions as, scientific data to support the idea of this 'pure' and 'original' founding population – an inherent contradiction. The ongoing effort by scientists to craft new identities for Indigenous peoples out of this flawed scientific narrative also perpetuates Indigenous distrust of the knowledge-producing methods of Western scientists.

A concerning trend that has arisen through the popularization of DTC tests has been non-Indigenous people increasingly seeking out such tests to claim Indigenous ancestry[122]. Indigenous legitimacy, in the contemporary era, carries political implications in terms of qualification for state benefits, sovereignty, and registration[122], and therefore should be defined by Indigenous communities rather than genetic ancestry. It has been noted how even the commercialization of these tests have included acts of appropriation: the notions of race and tribe, which are historically and culturally charged and belong to Indigenous peoples, are used in the marketing and interpretation of these tests[32]. Thus, the tests are both a product of, and reinforce, ideas of race and tribe, which takes the power of self-identification away from Indigenous communities and instead leaves them to be re-defined by DTC companies[32]. DTC tests enable the obsession with proving long-lost Indigenous ancestry and perpetuate the concept of gene fetishism: the idea that DNA is the sole source of identity[32] and thus directly undermines community-based forms of Indigenous identity.

DTC tests are presenting novel challenges to Indigenous identity and thus sovereignty and governance. With the growing demand for DTC tests, Indigenous communities and activists would be greatly benefitted from support from within Western medical establishments to lend additional support. As mentioned in the Introduction, GCs are mainly concentrated in metropolitan areas, working at large urban academic medical centers and requiring referral for access to their

services[43]. This geographical access issue is compounded by financial and psychosocial burdens, as patients must cover the cost of travel and accommodation, while taking time off work, and leaving their family and support systems to access GCing services[43]. It is well-established that barriers in access to health care is associated with worse health outcomes in rural populations[43,123], but DTC tests challenge these barriers by providing personal genetic data to consumers regardless of their location.

Here, I contend that the GCing profession should shift toward a more proactive, supportive role for Indigenous communities facing DTC test-associated challenges through the Duty to Inform. Under the CAGC Code of Ethics, GCs are obligated to inform patients such that they "benefit from advances in medical genetics", with "the goal of equal access to medical services" , and the Code encourages GCs to "act in the best interest of their patients and… offer appropriate clinical and psychosocial support, advocate…as necessary"[51,52]. Indigenous people deserve to share in the benefits of DTC tests, which include the ability to make informed lifestyle choices or change health behaviours, as well as receive early warnings of potentially disease-associated genetic variants to discuss with healthcare providers. For rural communities, often with insufficient access to medical care, offering remote GCing services to counsel on DTC test results could represent a significant step towards integrating genomics-based healthcare into underserved Indigenous communities.

Additionally, the CAGC Code of Ethics highlights how GCs have a "particular responsibility to ensure vulnerable patients are treated with due care"[51]. Due to their authority in the medical community, GCs who offer counselling to patients who pursue DTC testing could make special efforts to discuss with consumers the ways in which DTC tests erode community-based Indigenous identities, to intercept foreign genetics-based claims of Indigeneity identity

before communities are affected. This proactive work could make an important contribution to preserving Indigenous traditions regarding identity and community and thus support Indigenous sovereignty and self-governance. Such clarification in DTC counselling sessions could circumvent genetic essentialism in Indigenous discourse and identity, and so utilizing GCs to stress the importance of culture and tradition over DNA in constituting Indigenous identity could help defend Indigenous sovereignty and self-identity.

The CAGC Code of Ethics further emphasizes that GCs should "seek opportunities for continuing education"[51]. To proactively support Indigenous communities, GCs may need additional training to discuss Indigenous affairs and specifically the governance and identity concerns stemming from DTC tests. This cultural and historical training could be implemented through the courses and internships offered in GCing programs or through the CAGC. In learning to work alongside Indigenous communities, the work of Linda Alcoff may serve as a useful framework. Alcoff describes how one must be cautious of the position/location of the speaker and the context of the discussion in order avoid speaking over the voices one is aiming to support[124]; in the context of the ongoing impacts of colonialism and DTC tests, GCs offering counselling must be cognizant of the power dynamics at play in their relationship with Indigenous community groups and leaders. Alcoff suggests using self-questioning practices such as analyzing one's incentive, one's metaphorical location, practicing self-accountability and responsibility, and analyzing the possible future consequences of the speech[124]. GCs would benefit from additional training in this cultural sensitivity before working with Indigenous groups to ensure productive and beneficial counselling sessions. However, with the empathy and communication skills already ingrained in the practice of GCing, GCs are uniquely positioned to meet the needs of Indigenous communities in a way many others in the scientific community would be ill-equipped to attempt.

The challenges DTC tests present to Indigenous populations remains an understudied area and has great potential for the expansion of the role of GCs in the era of DTC biotechnologies. However, further training may be required before GCs could adopt a more proactive role in working with Indigenous communities. GCs, though, with their scientific and psychosocial expertise, have the potential to become powerful supporters in the broader quest to counter some of the challenges DTC tests pose to Indigenous identity and sovereignty.

5.4 Future directions

This preliminary survey has described the sentiments of Canadian GCs towards counselling patients who pursue DTC tests and the many implications this additional role holds for the vocation. While this survey gathered rich data, the participant response rate was not high enough to reach statistical significance, and thus the results may not be generalizable across the entire GC profession. To continue to clarify GC opinions on DTC tests and their professional obligations, interviews with GCs in clinical, academic, and industry roles could shed valuable insight on the feasibility of the proposed changes to the traditional duties and novel activist role for GCs proposed here. The role of the GC is facing new challenges with the expansion of biotechnologies into the hands of consumers and additional research in this area will help to identify the most efficient use of the limited time and resources of Canadian GCs.

Chapter 6: Conclusion

DTC genetic tests present a valuable knowledge translation tool to increase public engagement with genetics and may one day precipitate the democratization of personalized medicine. However, in the present era these tests are still entangled in questions of scientific validity and clinical utility, and the removal of healthcare providers from discussions of genetic and genomic health pose great concern for consumer informed consent and interpretation of

results. GCs may form part of the solution, as is the consensus in the literature[5,6,9,11,45–47] and in the Canadian and American Medical Associations[26,49] and Genetic Counselling Associations[27,50]. However, there is a paucity of research regarding what roles Canadian GCs could adopt outside of their traditional clinical roles and how this latest biotechnology challenges existing paradigms of the vocation. By means of a qualitative, closed-ended survey, this M.Sc. thesis has explored the challenges DTC tests pose to the traditional duties of Canadian GCs. GCs from across Canada, working in academia, industry, and clinical settings responded to the survey, representing a preliminary yet rich response to the themes and ideas interrogated in this survey.

The primary objective was to evaluate the current practices within Canadian DTC companies regarding the communication and delivery of results, which was established as preliminary research that informed the survey questions. The terms and conditions, privacy policies, and consent forms of DTC tests available for purchase in Canada were consulted to guide question development.

The second objective, to examine the ethical and policy issues DTC tests present to the traditional duties of GCs, was explored through the literature review, where academic discussion of DTC tests was explored, alongside the literature on the traditional duties of GCs.

The third objective, exploring how practicing Canadian GCs view DTC tests in Canada, was addressed through the survey of CAGC members. The survey results were analyzed through descriptive statistics to make recommendations for updating the traditional duties of Canadian GCs in light of DTC tests. The data gathered here suggest that the Duty to Inform should not be updated to include a mandate for GCs to provide counselling to patients who pursue DTC tests, as this additional burden is a misuse of clinical resources and time. However, since GCs report feeling confident in providing informed consent to DTC consumers, the Duty of Competence could be

updated to include staying informed of innovations in DTC tests, which would fit thematically with existing duties in the CBGC Knowledge-Based Competencies document[55] and would prepare GCs to counsel DTC patients if they chose to do so. Tasking GCs with having a working knowledge of DTC tests is not exceeding what is already common in the profession, according to the results of this survey, and therefore this update to the Duty of Competence is justifiable. From the findings of this survey, GCs are prepared to produce more education materials to inform the public about genetics and the science behind DTC tests, but GCs have very limited resources and clinical time, and many are wary of providing substandard care to patients of the healthcare system through allocating resources to provide counselling to DTC consumers. Therefore, while GCs may reasonably be tasked with staying up to date on innovations of DTC tests, in order to facilitate counselling if they choose to do so, it is ill-advised to task GCs with directly providing counselling to DTC consumers. The CAGC may adopt a greater advocacy and educational role with respect to DTC tests, and with the integration of genomics into Canadian healthcare the expertise and utility of GCs is critical and must be conserved.

This thesis also discussed the ways in which GCs who offer counselling to patients who have pursued DTC tests will have to adapt their services to meet the needs of this novel patient group. Drawing from the theory of harm reduction, I here contended that pre-counselling on DTC tests would help reduce consumer anxiety and perhaps minimize the number of DTC patients seeking follow-up counselling and test validation, and that virtual conferencing sessions with GCs could help to improve access to GCing services for patients in remote parts of the country. Additionally, DTC patients may bring a unique set of values to the counselling session, driven by unfounded beliefs about what information DTC tests provide, and the ongoing debate regarding whether DTC tests truly empower patient autonomy to make informed health choices or

behaviours may affect how DTC patients approach GCing sessions. DTC tests also inherently challenge one of the core obligations of the GCing profession: the Duty to Inform. GCs must contend with potentially being unable to counsel patients on false positives or negatives, due to a lack of access to test methodologies. Thus, GCs will have to rely on their professional judgement whether to recommend clinical validation, which is additionally impacted by resources in their health jurisdiction and may represent an additional, unnecessary toll on their healthcare system. As well, GCs would be unable to truly satisfy the Duty to Inform on the privacy concerns of DTC tests, as companies may not have publicly available privacy terms and GCs would be unable to provide patients information on sample retention, storage, use, anonymization, or sharing to third parties. DTC tests also challenge the Duty to Inform through a built-in bias; due to underlying systemic problems of construct validity in the field of genetics, DTC tests may provide vague or even inaccurate results to POC. GCs may feel unprepared to address such sensitive topics when counselling POC DTC patients and may require additional support to discuss diversity, inclusion, and non-discrimination in the DTC counselling context.

DTC genetic tests, and future direct-to-consumer biotechnologies, will inevitably bring changes to the role of GCs and healthcare practitioners. GCs have unique and critical training in psychosocial counselling and genetic expertise, and as novel biotechnologies continue to emerge, the role must continue to adapt. The final section of the thesis highlighted how the GCing literature has not yet discussed in detail how DTC tests are impacting Indigenous communities. DTC tests challenge Indigenous self-identity through privileging 'scientific' definitions of Indigeneity over community traditions, which holds serious implications for Indigenous sovereignty and self-governance[33,120]. Drawing from the CAGC Code of Ethics, GCs could begin to proactively support Indigenous communities while expanding their access to genomics-based healthcare through

offering counselling for DTC tests, while taking a more assertive stance in their counselling of other DTC consumers against DTC-based Indigenous identity claims. With additional training in Indigenous affairs and cultural sensitivity, GCs hold great potential for supporting Indigenous communities in Canada through some of the challenges DTC tests pose.

This research has helped to clarify the traditional duties and additional roles GCs may adopt given the challenge of DTC tests and will be useful to GCs and physicians to understand how GCing curricula and expectations of knowledge could be adapted to meet this disruptive biotechnology. Additionally, insights from the survey are beneficial for informing future guidelines of DTC testing communication materials and informed consent processes in Canada, and aid in characterizing the most efficient use of GCs' limited time and resources to support Canadian DTC consumers. While Canadian GCs are not yet prepared to accept professional responsibility for counselling DTC patients, the expansion of the GC role to include knowledge of DTC tests, increased production of GC-supported public education materials, and expanding professional GC knowledge competencies for DTC tests strengthens the argument for the legal recognition of the GCing profession by augmenting their role in the Canadian healthcare and public health systems.

DTC tests may one day help facilitate a preventative model of healthcare, empowering patients with their genetic health data to make informed health and lifestyle choices and saving the healthcare system untold time and resources when predispositions are caught early, and appropriate changes are implemented. However, until DTC tests become scientifically valid and clinically useful, the knowledge transfer abilities of such tests remain an untapped potential.

Chapter 7: References

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Appendix A: Online survey (EN)

Online survey for the study: "Direct-to-consumer genetic tests: Is there a role for Canadian genetic counsellors?"

Student Investigator: Cassandra Haley

Supervisor: Dr. Ma'n Zawati

Inclusion criteria

Thank you for your interest in this research study. The goal of this study is to understand the traditional duties, future roles, and involvement of Canadian genetic counsellors in regards to direct-to-consumer genetic tests.

1. Are you a Canadian- or American-certified genetic counsellor?
    a) Yes
    b) No
2. Are you a genetic counsellor currently working in Canada?
    a) Yes
    b) No
3. In your role as a genetic counsellor, have you worked with patients?
    a) Yes
    b) No

IF THE RESPONSE TO ANY OF THE ABOVE ITEMS IS 'NO', PROMPT AS INELIGIBLE FOR THE STUDY AND END SURVEY.

1. Informed Consent

1.1. Do you consider it part of your professional obligations to work with direct-to-consumer genetic test [DTC tests] consumers?

a) Yes—DTC tests fall within the scope of professional practise

b) No—DTC tests are outside of the scope of professional practise

1.2. Currently, terms of service documents vary widely in the DTC industry. If you are familiar with DTC terms of service documents, do you believe they sufficiently describe the limitations of DTC tests?

Terms and service documents:

a) Are too varied across the industry

b) Clearly describe the tests' benefits and limitations

c) Poorly describe the tests' benefits and limitations

d) Do not describe the tests' benefits and limitations at all

e) Unfamiliar with DTC terms of service documents

1.3. If you responded e) for question 1.2, which of the following elements would you prioritize for inclusion? Select all that apply.

- Layman's description of services provided
- Disclaimers: not medical diagnoses/devices, no guarantee accuracy/security, not liable for damages, will disclose to law enforcement
- Limitations of DTC tests (comprehensibility, gene-environment interaction)
- Retention, use, storage, disposal of sample plan
- Recommendation to speak with genetic counsellor

1.4. Most DTC terms of service documents describe the sequencing technology and/or assays used in the test. Do you believe the language used sufficiently promotes consumer informed consent?

a) Yes

b) No

c) Unfamiliar with DTC terms of service documents

1.5. How would you rate following statement: DTC tests remove genetic counsellors from discussions of genetic health data, which negatively impacts consumer informed consent.

a) Strongly disagree

b) Disagree

c) Neutral

d) Agree

e) Strongly agree

1.6. Would standardization of consent forms across the industry better inform DTC consumers?

a) Yes—standardization would improve informed consent

b) No—standardization would not benefit consumers' informed consent

c) Not feasible

d) Unsure

1.7. Do you interpret the Canadian Association of Genetic Counsellors [CAGC] Code of Ethics tenant to "promote awareness of the roles of medical genetics professionals" to include collaboration with the DTC test industry?

a) Yes (select why):

- Would allow for evaluation of DTC tests
- Would allow for standardized informed consent
- DTC company funding could allow for research & development in the genetic counselling field

b) No (select why):

- Inappropriate—beyond our scope of practice
- Conflict of interest

1.8. The CAGC Code of Ethics states that genetic counsellors should:

> "… promote awareness of the roles of medical genetics professionals through activities such as participation in multi-disciplinary teams, providing public education, contributing to policy-making and provincial/national consultation".

Do you think that "providing public education" should include information on DTC tests?

a) Yes

b) No

1.9. Do you believe the profession of genetic counselling needs to do more to protect the informed consent of DTC customers compared to traditional patients seeking counselling?

a) Yes—DTC customers merit additional safeguards to ensure informed consent because:

- The tests contain significant limitations which may be unclear to consumers
- The absence of healthcare professionals creates additional challenges to informed consent that merit attention
- There is limited information about the accuracy of DTC test results

b) No—DTC customers currently receive adequate support to give informed consent because:

- The tests are not medically necessary and therefore do not merit intervention from healthcare professionals
- Ensuring informed consent for each DTC customer would be an inefficient use of clinical time and resources
- Genetic counsellors are not adequately trained to counsel DTC customers on informed consent

1.10. DTC tests continue to improve quality and accuracy of results every year. How does the continual improvement of DTC test results impact the genetic counselling profession? Select all that apply.

- Genetic counsellors will be expected to work with larger medical teams to interpret more specific results
- It will prolong the relationship with DTC customers to offer counselling as results are updated
- It will expand the knowledge required of genetic counsellors beyond a reasonable understanding of DTC tests, diverting time and energy from clinical patients
- It does not impact the practice of genetic counselling—existing medical genetic tests also continue to improve

2. Privacy

2.1. As an expert in handling sensitive genetic data, how concerned are you with the privacy policies of DTC companies operating in Canada? 1-unconcerned, 5-strongly concerned.

Sliding scale 1–5

2.2. How do you believe DTC tests challenge customer privacy, if at all? Where do you perceive privacy issues in DTC tests? Select all that apply.

- ☐ The retention of samples
- ☐ The anonymization of genetic data
- ☐ The sale of aggregate genetic data
- ☐ The federal *Genetic Non-discrimination Act* and *Personal Information Protection and Electronic Documents Act* ensure DTC consumers' privacy is protected
- ☐ DTC tests adequately safeguard personal genetic security

2.3. Would you like to see any of the following privacy protections in DTC tests? Select all that apply.

- ☐ Independent auditing of security features
- ☐ Encryption of data
- ☐ Aggregation of data
- ☐ Company registration with a government agency
- ☐ Privacy standards should be left up to individual company discretion

2.4. Do you believe the profession of genetic counsellors needs to do more to protect the privacy of DTC customers compared to traditional patients seeking counselling?

a) Yes (select why):

- ☐ Informed consent is often difficult to achieve across the DTC customer population without the involvement of a healthcare professional, therefore privacy must be closely monitored
- ☐ The lack of regulation of DTC company privacy policies is concerning

b) No (select why):

- ☐ Beyond the scope of genetic counselling practice
- ☐ Intervention is not a wise use of clinical time
- ☐ Intervention is not feasible

c) Unsure

2.5. How can the genetic counselling profession respond to privacy concerns of DTC tests? Select all that apply.

- ☐ Advocate for reform through CAGC
- ☐ Produce communication materials to educate the public
- ☐ Collaborate with the Office of the Privacy Commissioner of Canada to educate the public
- ☐ Responding to privacy concerns is outside the scope of the genetic counselling profession

2.6. The inclusion of DTC results in patients' medical records could potentially open the door to genetic discrimination practices. Do you see this as a reasonable concern of DTC tests?

a) Yes

b) No

3. Genetic Counsellors' Involvement in Policy

3.1. Generally speaking, how heavily do you think the genetic counselling profession should be involved with DTC tests? 1-not at all involved, 5-heavily involved.

Sliding scale 1–5

3.2. Do you see the lack of recognition as healthcare professionals as an impediment to your work with DTC test customers?

a) Yes

b) No

3.3. Please rate your response to the following statement: DTC tests are sufficiently regulated.

a) Strongly agree

b) Agree

c) Neutral

d) Disagree

e) Strongly disagree

3.4. What level of involvement should CAGC hold with respect to DTC test evaluation? Select all that apply.

- Grade tests (maintain a list on CAGC website)
- Recommend tests (through the CAGC website)
- Accredit tests (label tests similarly to how the Canadian Dermatology Association labels products)
- It is currently unfeasible for CAGC to be involved in this process
- CAGC should not attempt to evaluate DTC tests in any capacity

3.5. Given the limited number of genetic counsellors in Canada, and the high volume of Canadians taking DTC tests every year, do you believe it is feasible to offer pre- and post-clinical genetic counselling to DTC customers?

a) Yes

b) No (select why):

- There are not enough genetic counsellors to offer this service
- This should be the responsibility of DTC companies

3.6. If you do not believe it feasible to offer both pre- and post-counselling, which intervention has the potential for the most impact on the consumer?

a) Before—promote informed consent/educate consumers in advance

b) After—review results

c) It is not feasible to offer counselling to DTC customers

3.7. Alternative communication models of medicine have emerged throughout the pandemic as healthcare professionals adapt to fewer in-person interactions. Do you believe that any of the following methods could alleviate the burden of DTC test counselling? Select all that apply.

- Provision of virtual conference within province
- Provision of virtual conference within the country
- Online resources—managed by genetic counsellors
- It is not feasible to offer counselling to DTC customers

3.8. Given the high demand for counselling of DTC customers, what is currently feasible for CAGC to offer? Select all that apply.

- Establish a special interest group (SIG) at the annual CAGC meeting to develop policy review of DTC tests
- Organize seminars for primary care physicians or other healthcare professionals to prepare them to offer counselling of DTC results
- Advocate for federal regulation of DTC tests: scientific validity, consent forms, terms and conditions agreements, privacy standards
- Produce informative materials on DTC tests

3.9. A recent study by Hsieh *et al.* (*J. Genet. Couns.* **30,** 191-197, 2021) found that the majority of surveyed genetic counsellors believe it is the responsibility of DTC companies to provide counselling for consumers rather than clinical genetic counsellors. Why do you agree or disagree with this finding? (Select all that apply)

a) I agree with this finding because:

- ☐ There are too few accredited genetic counsellors across Canada to provide this service
- ☐ The uneven distribution of clinical genetic counsellors across Canada creates access issues
- ☐ The tests are not medically necessary; therefore, customers do not merit access to a limited pool of professional genetic counsellors
- ☐ DTC customers are not patients of the healthcare system
- ☐ The DTC industry has sufficient resources to recruit genetic counsellors
- ☐ Offsetting DTC counselling responsibility to clinical geneticists helps the industry avoid accountability
- ☐ I do not believe counselling DTC customers is within the purview of clinical genetic counselling

b) I disagree with this finding, clinical genetic counsellors should provide counselling services to DTC customers

3.10. How do you see the current role of genetic counsellors changing with regards to the DTC test industry? Select all that apply.

- ☐ Increased collaboration with primary care physicians
- ☐ Increased collaboration with other healthcare professionals

- Increased work outside clinical roles
- Increased advocacy work
- Genetic counsellors should focus on clinical patients rather than DTC customers

4. Demographic Information

4.1. In your career, how often have you worked directly with DTC customers?

- 0–5 individual customers
- 6–10 individual customers
- 11–20 individual customers
- 21–50 individual customers
- 50+ individual customers

4.2. In your work with DTC customers, what are the concerns most often raised by clients? Rank from highest to lowest concern.

a) I have worked with customers:
   1. Interpretation of results
   2. Implication for family members
   3. Privacy concerns
   4. Lack of informed consent

b) I have not worked with DTC customers

4.3. In providing counselling to DTC customers, have you collaborated with other healthcare professionals?

a) Yes

b) No

c) I have not worked with DTC customers

4.4. Out of which province/territory is your position currently based? (Select from drop-down menu)

- Alberta
- British Columbia
- Manitoba
- New Brunswick
- Newfoundland-Labrador
- Northwest Territories/Nunavut
- Nova Scotia
- Ontario
- Prince Edward Island
- Quebec
- Saskatchewan
- Yukon

4.5. In what year did you graduate from your genetic counselling training program?

Drop down: Years 1970-2021

4.6. Which of the following roles best describe your <u>current</u> genetic counselling position(s)? (Select all that apply)

- Clinical
- Laboratory
- Research
- Academic/education

- Industry
- Other

4.7. Which of the following best describes the work setting of your current genetic counselling position(s)?

- A genetic clinic
- A non-genetic department/division within a healthcare institution (public sector)
- Industry
- Private-pay clinic
- University
- Government
- Other

4.8. Which of the following roles best describe your previous genetic counselling position(s)? (Select all that apply)

- Clinical
- Laboratory
- Research
- Academic/education
- Industry
- Other

4.9. Which of the following best describes the work setting of your previous genetic counselling position(s)?

- A genetic clinic
- A non-genetic department/division within a healthcare institution (public sector)

- ☐ Industry
- ☐ Private-pay clinic
- ☐ University
- ☐ Government
- ☐ Other

END OF STUDY

Appendix B: Survey consent form

Informed consent form for the study: "Direct-to-consumer genetic tests: Is there a role for Canadian genetic counsellors?"

MCGILL UNVERSITY

INFORMED CONSENT FORM FOR PARTICIPATION IN A RESEARCH STUDY

Study Title: *Direct-to-consumer genetic tests: Is there a role for Canadian genetic counsellors?*

Student Investigator: Cassandra Haley

Supervisor: Dr. Ma'n Zawati

Institution: McGill University

INTRODUCTION

You are being invited to take part in a research study. Your participation in the study is voluntary, and you are free to refuse to participate. Should you decide not to be in the study, it will not affect your relationship with the researcher or McGill University.

This research study looks at the roles of Canadian genetic counsellors in relation to the emergence of direct-to-consumer (DTC) genetic tests. We are asking you to participate in this study because you are a Canadian or American certified genetic counsellor, who is working in Canada, and works with patients.

DTC tests are at-home genetic tests that Canadians may purchase to learn more about their genetic information without referral by a healthcare practitioner. The tests offer insights into one's ancestry but can also provide information on genetic diseases and predispositions to customers. The DTC test market is rapidly growing in Canada but is not currently regulated by Health Canada, leading experts to question the scientific validity and clinical utility of the tests. The Canadian Medical Association and the Canadian Association of Genetic Counsellors both recommend pre-

and post-counselling by genetic counsellors to promote informed consent and explain results, in that order. Although similar recommendations have been made in the literature, no framework currently exists for how this service ought to be provided.

There are no conflicts of interest to declare related to this study.

WHY IS THIS STUDY BEING DONE?

This study is being undertaken to investigate the ways in which the expertise and traditional duties of Canadian genetic counsellors are challenged by the DTC test industry. Indeed, the purpose of this exploratory study is to describe the current roles of genetic counsellors in regard to DTC tests, to analyze alternative strategies of involvement with DTC tests, and to consider the implications of genetic counsellors engaging with DTC tests in the ethico-legal context, as genetic counsellors are not legally recognized as healthcare professionals across Canada.

WHAT IS INVOLVED IN THIS STUDY?

If you agree to participate in the study, you will be asked to complete an online survey. The survey questions will be close-ended and will address information about your experiences working with DTC test consumers, your opinions of how genetic counsellors should work with DTC test consumers and companies, and how you think the traditional roles of genetic counselling should be changed because of the tests. The survey will also collect demographic information including your province of work and previous work experience. The survey should take 20-30 minutes to complete.

No contact information will be solicited in the survey.

If you have any questions, you may contact the principal investigator using the contact information provided below.

WHAT ABOUT CONFIDENTIALITY AND PRIVACY?

If you decide to participate in this study, your participation will be kept confidential. Your survey response will not be directly identifying. Your survey response will be assigned an alpha-numeric code, and all data will be aggregated for analysis. At the end of the study, data will be anonymized; the separate document linking individual surveys with the alpha-numeric code will be destroyed.

It is expected there will be 180-200 participants across Canada. Therefore, there is minimal risk of being re-identified through indirectly identifying information collected in this study. Information that will be collected in this study includes province of practice, work setting, and field of practice. This demographic information in combination may make re-identification possible. Re-identification poses a personal risk as a major goal of the study is to evaluate the potential legal and ethical duties of genetic counsellors in regard to DTC tests.

The following measures have been implemented to protect your privacy:

4) All surveys will be assigned an alpha-numeric code. Participants' identifying information will not be collected.
5) It is expected that the data collected in this study will be used in analyses and it may be published or presented to the larger scientific community at meetings or in the literature. Therefore, any publications related to this study, including supplemental materials, will minimize information regarding each individual participant to reduce the likelihood of re-identification. Demographic data will be either grouped, representing multiple participants, or will be limited as it relates to individual participants. For example, when referring to your specific work setting and field of practice, your province of practice will not be included.

These protections are in place to reduce the risk of re-identification, but a small risk remains. Despite the protections being in place, there is also a risk of unintentional release of information.

Authorized representatives of the McGill Research Ethics Board may look at your original consent (possibly identifiable), to check that the information collected is correct and follows proper laws and guidelines.

As required by the McGill Regulation on the Conduct of Research, your survey responses will be retained for 7 years, or until you decide to withdraw your participation from the study.

MAY I LEAVE THE STUDY?

You can choose to end your participation in this research study at any time, without having to provide a reason and with no consequence. If you do not complete the entire survey, you will automatically be withdrawn from the study. Otherwise, you may provide written notice to the student investigator by email.

WHAT ARE THE RISKS OR HARMS OF PARTICIPATING IN THIS STUDY?

There is a small risk of being re-identified based on combined non-identifying information (demographic information), which could be unique to you as a participant. Though we have implemented strict measures to ensure privacy, there is always a small risk that your demographic information could lead to you being re-identified.

WHAT ARE THE BENEFITS OF PARTICIPATING IN THIS STUDY?

There are no personal benefits to participation in this research study. However, your participation in the study will allow those in the field of genetic counselling to better understand the appropriate professional involvement of Canadian genetic counsellors in the unregulated and volatile market of DTC genetic tests. Results of this study may also help to uncover additional roles for genetic counsellors within the healthcare system or DTC industry to protect anxious

customers, which will be of use in the context of legal recognition and regulation of the genetic counselling profession.

WHAT ABOUT COST AND COMPENSATION?

Participation in this study will not involve any costs to you, nor will you be compensated for taking part in this study.

WHAT ARE MY RIGHTS AS A PARTICIPANT IN A RESEARCH STUDY?

You have the right to ask questions about the study at any time.

Your participation in this research study is voluntary. You may choose not to participate, or withdraw from the study at any time, for any reason, without penalty.

Your rights to privacy are legally protected by federal and provincial laws that require strict safeguards to ensure your privacy is respected.

By accepting these terms, you do not give up any of your legal rights against the researcher or involved institutions for compensation. This form does not relieve the researcher or their agents of their legal and professional responsibilities.

WHOM DO PARTICIPANTS CONTACT FOR QUESTIONS?

If you have questions about participating in this study, you can talk to the research team, or the person who oversees the study at McGill University.

Student Investigator: Cassandra Haley

Contact: cassandra.haley@mail.mcgill.ca

Supervisor: Dr. Ma'n Zawati

Contact: man.zawati@mcgill.ca

If you have questions about your rights as a participant or about the ethical issues related to this study, you may contact the McGill Faculty of Medicine and Health Sciences Institutional Review Board (McGill IRB), which is not involved in this study at all.

Contact: (514) 398-3124

ATTESTATION

☐ I attest to the following:

- The study has been explained to me and all of my questions have been answered to my satisfaction.
- I do not give up any of my legal rights by signing this consent form.
- I agree to participate in this study.