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Utilization and Perceived Value of Genetic Counselors Within U.S. Hemophilia Treatment Centers

Caylynn Carls
Sarah Lawrence College

UTILIZATION AND PERCEIVED VALUE OF GENETIC COUNSELORS WITHIN U.S. HEMOPHILIA TREATMENT CENTERS

Caylynn Carls

Sarah Lawrence College, Bronxville, NY

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ABSTRACT

Rapid advancement of molecular genetics has transformed the diagnosis, treatment, and management of individuals with hereditary bleeding disorders. To provide effective, up-to-date genetic counseling, navigate the complexity of these conditions, and select appropriate molecular testing, genetics expertise is required. This study assessed the provision of genetic counseling services, the involvement of genetic counselors (GCs), and the perceived value of GCs within hemophilia treatment centers (HTCs) in the United States. A survey was emailed to 396 HTC providers. 115 responses were received, representing 68 of 149 U.S. HTCs (45.6% HTC participation rate). Responses were stratified by level of GC engagement. Although GCs have extensive training in genetics, genomics, and counseling skills, nearly one-third (34.9%) of respondents reported that a GC is not involved with the HTC nor are referrals made. 98% of GC-engaged HTC respondents and 95% of GC-referrals agreed that "GCs have a unique skill set that is highly valuable to an HTC clinic" compared to only 62% of non-GC-engaged respondents (p= .001). Additionally, many respondents noted positive implications of integrating a GC within their HTCs, stating that GCs are "ideal for optimal patient care." These results highlight the value of a GC within an HTC. This signifies the need to reassess the role of GCs among HTCs to reduce inconsistencies in the provision of genetic counseling and increase healthcare equity.

KEYWORDS: Hemophilia treatment centers, genetic counseling, genetic counselors, genetic testing, multidisciplinary clinic, service delivery, provider perspectives

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INTRODUCTION

Hemophilia treatment centers (HTCs) have been utilized for the comprehensive care of individuals with hereditary bleeding disorders in the United States since the early 1970s, with an emphasis on early diagnosis and intervention to prevent lifelong complications and address related comorbidities (Ruiz-Sáez, 2012). Several studies have demonstrated that optimal management of hereditary bleeding disorders can lower mortality rates, decrease hospitalizations, and improve overall quality of life in affected individuals (Ruiz-Sáez, 2012; Barlow, et al., 2007; Rhynders, et al., 2014).

HTCs are expected to adhere to specific standards and criteria as outlined in the National Hemophilia Foundation's 2002 Medical and Scientific Advisory Council guidelines document (NHF-MASAC), which defines expectations for the "Core Team", "Extended Team", and services provided. According to these guidelines, genetic counseling is a required component of the comprehensive HTC evaluation; however, this service can be delegated to but is not required to be performed by a genetic counselor (GC) (NHF-MASAC, 1994, Revised 2002). Consequently, the provision of genetic counseling services varies significantly among HTCs (Alabek, et al., 2015).

Since the publication of the 2002 NHF-MASAC guidelines, methods for molecular diagnosis have evolved significantly, directly impacting the management of patients with hereditary bleeding disorders. Since the 1990s, Sanger sequencing guided by clinical and laboratory phenotypes has been viewed as the "gold standard" for molecular diagnostics, despite being costly, time consuming, and having a lower sensitivity. With recent advances, Next Generation Sequencing approaches have become instrumental in molecular diagnosis, allowing for simultaneous sequencing of multiple genes, increased sensitivity, and quicker means to

providing diagnostic clarity to patients (Bastida et al., 2019). Recently updated management and diagnosis guidelines recommend molecular diagnosis for (1) individuals clinically suspected to have hemophilia, (2) at-risk carriers for future prenatal diagnosis, and (3) symptomatic females. This information also has implications in prediction of inhibitor development and response to immune tolerance induction therapy and determining the feasibility for potential gene therapies (Srivastava, et al., 2020). Similarly, recent updates to von Willebrand disease guidelines suggest genetic testing to confirm a diagnosis of type 2N, a genocopy of mild hemophilia A, and to distinguish between type 2A and 2B, which impacts treatment (Connell, et al., 2021).

Concurrent with the evolution of molecular diagnosis, the field of genetic counseling has witnessed rapid growth. The roles, recognition, and scope of practice of GCs have expanded dramatically within and across medical specialties particularly since the NHF-MASAC guidelines were first released in 1994. For example, BRCA1, the breast/ovarian hereditary cancer syndrome gene, had just been discovered in 1994, and only about 10% of GCs worked in the new specialty of cancer genetic counseling; by 2002, when the NHF-MASAC guidelines were last revised, over 40% of the growing number of GCs practiced in cancer genetics (Bennett, et al., 2003). Similarly, a year before the 1994 NHF-MASAC guidelines were released, the American Board of Genetic Counseling (ABGC) began certifying GCs and accrediting training programs, recognizing at that time fewer than 500 certified genetic counselors (CGCs); today, ABGC recognizes a workforce of more than 5,000 certified GCs, who "continue to integrate counseling services into an increasing number of medical specialties, such as oncology, cardiology and psychiatry" (ABGC, 2021; Ormond, et al., 2018). GCs now hold positions within various multidisciplinary clinics including but not limited to hematology, oncology, and cystic fibrosis (Hudson, et al., 2019; Langfelder-Schwind, et al., 2019).

Board-certified GCs must demonstrate twenty-two practice-based competencies categorized within four domains: genetics expertise and analysis, interpersonal, psychosocial and counseling skills, education, and professional development and practice (Accreditation Council for Genetic Counseling, 2019). Recent research has suggested both a lack of understanding of genetic counseling competencies as well as a lack of appropriate utilization of GCs by providers within multidisciplinary clinics. A study by Hudson, et al. (2019), revealed that many common roles that GCs are extensively trained for were either shared or performed by another provider. These include but are not limited to: eliciting pregnancy, developmental, or medical histories; psychosocial counseling; constructing a pedigree; developing a differential diagnosis; interpreting genetic testing results; and explaining genetic concepts and results of genetic testing. Although limited in scope, these results and the current paucity of data in this area demonstrate the need for further research to ascertain provider perceptions of GC scope, value and utilization of GCs in multidisciplinary settings.

A study examining the roles and perceived value of GCs in cystic fibrosis centers (CFC) demonstrated that in "GC-engaged" CFCs, i.e., those where GCs regularly staff CFCs, providers expressed a more positive perception of the expertise, value, level of understanding of cystic fibrosis (CF), accessibility, and cost of GCs as compared to provider perception in "non-engaged" CFCs who referred out to GCs or CFCs that did not utilize GCs. For instance, only 21% of non-engaged CFC respondents valued GCs as compared to 100% of GC-engaged CFCs. Similarly, just 39% of respondents in non-engaged CFCs agreed that GCs have a high-level understanding of cystic fibrosis (CF) compared to 100% of those involved in GC-engaged CFCs ($p<.001$). Additionally, the utilization of GCs within CFCs was found to improve the efficiency of the diagnostic resolution process when a newborn screened positive for CF. In contrast,

providers from CFCs that did not work closely with a GC had a more negative view of the same parameters (Langfelder-Schwind et al., 2019). These results suggest that integration of GCs within the CFC multidisciplinary setting positively influences overall perceptions about GCs as well as increases understanding of GC roles, which may lead to effective utilization of GC services and improvement in the management of patients with this complex genetic condition.

Provision of genetic counseling by non-genetics providers has resulted in various adverse outcomes due to inadequate genetic knowledge, limited time for patient encounters, and case complexity (Brierley, 2012). Documented negative outcomes include missed or delayed diagnoses, incorrect or unnecessary medical treatment, misuse of healthcare and patient dollars, elevated psychosocial stress and false reassurance, and increased morbidity and mortality (Bensend et al., 2014; Hudson et al., 2019; Farmer, 2019). In contrast, utilization of certified GCs allows for appropriate ordering of genetic testing, provision of accurate risk assessment, reduction of unnecessary testing and medical treatments, and a decrease in patient anxiety (Hudson et al., 2019).

As the uptake of genetic testing increases, providers must be increasingly proficient in genetics in order to properly navigate the ever-changing complexity of test selection and interpretation of molecular diagnostic testing, carrier testing, and prenatal diagnostic testing, while optimally integrating nuanced inheritance implications and genetic test results into clinical care and management and simultaneously relaying accurate information to patients and families. In the context of HTCs, it has been suggested that regularly involving a GC in the comprehensive care of patients lessens this burden on other providers while concurrently ensuring patients receive appropriate genetic testing, accurate results interpretation, provision of

recurrence risk information, and psychosocial support following a diagnosis of a hereditary bleeding disorder (Alabek et al., 2015).

In order to gain insight into the current utilization and perception of GCs within HTCs, this study assessed the provision of genetic counseling services, the involvement of GCs, and the perceived value of GCs among HTCs in the United States.

MATERIALS AND METHODS

An original web-based survey was developed using the Qualtrics platform and distributed via email to directors, nurse coordinators, and primary contacts of all 149 United States (U.S.) accredited HTCs located within the 10 regions of the HTC network, as listed on the Centers for Disease Control and Prevention (CDC) HTC database. The survey was sent in September 2020 to 396 email addresses, followed by two email reminders. The original web-based survey used for this study was inspired by instruments used by Langfelder-Schwind et al (2019).

Participation was voluntary and data was deidentified in order to provide anonymity. There was no compensation for participation however, upon survey completion respondents had the option to participate in a raffle for a one-hundred-dollar gift card. Prior to entering the survey, respondents were required to provide informed consent. Background information collected included their current role in the HTC and, for tracking purposes, the HTC number or city/state where the HTC is located. Survey questions were designed to elicit perceptions of various GC roles, to ascertain the current utilization of GCs in their HTC and to collect information regarding any barriers to employing a GC. Questions relating to GC roles were based on the twenty-two Accreditation Council for Genetic Counseling (ACGC) competencies. The survey used skip logic format, with Likert scale and multiple-choice question types, and all questions included an optional space for additional information on their choice selection.

This study received exemption from review by the Sarah Lawrence College Institutional Review Board.

Data Analysis

Responses were categorized by the level of GC-engagement within the HTC. A GC-engaged HTC was defined as one in which a GC is employed by the HTC or a parent institution and regularly staffs the HTC. A GC-referral HTC is one where a GC is not directly employed by the HTC or institution, and does not routinely staff the clinic, but referrals are provided as needed. Lastly, non-GC-engaged HTCs are ones in which a GC is not employed by the HTC or institution nor are referrals made.

Statistical analysis was performed in SPSS (Statistical Package for the Social Sciences), Version 27.0, using Fisher's exact test (threshold for significance: $p<0.05$) to determine association because of the small sample size. Qualitative responses were analyzed and manually coded for common themes. Coding was reviewed with two other team members. All responses, as well as partially completed surveys, were included in the analysis.

RESULTS

Of the 396 emails sent to recipients, 6 emails were returned as undeliverable, and 4 recipients noted that they either no longer worked within the HTC or were on leave. There were a total of 115 responses (29% response rate), of which 101 were fully completed. The majority of respondents were nurses (34.2%), medical directors (18.9%), and clinic administrative staff (18.9%) (Figure 1).

Out of the 149 U.S. accredited HTCs, 68 were represented in this study (45.6% HTC participation rate). 13 respondents provided a city or state location which has multiple HTCs, so investigators were unable to match the location with an HTC identification number. All 10

regions of the U.S. HTC network were represented in the dataset, with the majority coming from the Great Lakes region (n=18) where, coincidentally, the greatest number of HTCs (n=21) are located. The Mid-Atlantic (n=7), Great Lakes (n=9), and Northern States (n=9) regions had the most GC-engaged responses (Figure 2).

Close to half of the respondents (46.8%) reported that their HTC was GC-engaged (Figure 3). Nearly a third (34.9%) were categorized as non-GC-engaged and 18.4% were categorized as GC-referral. Those respondents who reported that GC services are provided by other HTC staff members (21.1%) were asked to specify which specific non-GC staff members were delivering these services to their patients. Physicians were most often noted to be the team member to provide genetic counseling, followed by nurses. Many mentioned that if needed, patients are referred to GCs after education from providers.

Competencies of GCs

Data analysis revealed significant variation when responses were stratified by level of GC-engagement. Due to skip-logic formatting, only responses categorized as GC-engaged (n=25) and non-GC-engaged (n=17) were included in this analysis. GC-referrals did not answer this question and were not included in this analysis (Table 1).

All GC-engaged respondents agreed that GCs provide a unique service to HTC families and possess a high level of understanding of hereditary bleeding disorders, compared to 65% of non-GC-engaged respondents (p= .002). Similarly, 100% of all GC-engaged respondents agreed that GCs exhibit understanding of healthcare systems and delivery models to promote responsible, equitable, and cost-effective integration of genetic services to improve patient care, compared to only 77% of non-GC-engaged respondents (p= .021). All GC-engaged HTC respondents believe that, ideally, a GC should be a part of an HTC team, with one respondent

noting “Our genetic counselors are invaluable to our HTC”, whereas only 59% of non-GC-engaged respondents agreed with this statement (p= .001).

Engagement stratification was marginally significant with respect to whether GCs show skill in research, self-education, and educating clients about a wide range of genetics and genomic information, with 100% of GC-engaged respondents agreeing with this statement as compared to 82% of non-GC-engaged HTC respondents (p= .059).

Although not statistically significant, it is important to note that over 95% of GC-engaged and non-GC-engaged respondents agreed that GCs tailor and communicate relevant information for various audiences, possess strong interpersonal skills to promote multidisciplinary relationships, and utilize advanced interviewing and counseling skills to empathetically respond to concerns. According to one respondent, “It was tremendously helpful to have a dedicated person in this role who was able to get to know our team and patients, and who could also do more education and outreach through our chapter events, programming, etc.” Respondents from both groups all agreed that GCs demonstrate a depth and breadth of genetics knowledge and constructs, and/or assess relevant, targeted, and comprehensive histories for bleeding disorder or carrier status risk assessment.

Issues Ideally Addressed by GCs

Due to skip-logic formatting, only responses categorized as GC-engaged (n=25) and non-GC-engaged (n=17) were included in this analysis. GC-referrals did not answer this question, therefore are not included in this analysis (Table 2).

While not statistically significant, over 95% of GC-engaged and non-GC-engaged HTCs agreed that ideally a GC should discuss genetic testing utilization/selection strategy, disclose genetic testing results, and explain the implication of genetic testing results. In particular, 96% of

GC- engaged respondents agreed that, ideally, a GC should obtain informed consent for clinical genetic testing, compared to 71% of non-GC-engaged respondents (p= .032).

Skills GCs Add to HTCs

Responses categorized as GC-engaged (n=22), GC-referrals (n=20), and non-GC-engaged (n=20) were included in this analysis (Table 3). Some questions were not answered by all 115 respondents due to skip logic formatting and inclusion of partially completed surveys; reported n values are variable for this reason.

While not statistically significant, responses reflected that 55% of non-engaged HTCs felt that their staff members were capable of providing the functions of a genetic counselor compared to only 27% of GC-engaged and 25% of GC-referral HTCs (p= .148).

Although not statistically significant, over 95% of all respondents agreed that a GC could explain genetic concepts of bleeding disorders, discuss testing strategy, disclose genetic testing results, and explain the various implications of genetic testing results to patients and families in an HTC, despite the level of engagement. These results were supplemented by additional open-ended responses in which respondents described the "valuable" skills that their GC adds to the HTC, including providing "educational information to our patients", assessing and coordinating "genetic testing for family members at risk", addressing "billing concerns", and "interpretation of genetic testing results." It was also noted that the knowledge of a genetic counselor could be advantageous in helping obtain insurance authorization for genetic testing.

Value of GCs within HTCs

Responses categorized as GC-engaged (n=47), GC-referrals (n=20), and non-GC-engaged (n=37) were included in this analysis (Figure 4).

When presented with the statement "I feel that a genetic counselor has a unique skill set that is highly valuable to an HTC clinic," 98% of GC-engaged and 95% of GC-referral respondents agreed, compared to only 62% of non-GC-engaged respondents ($p < .001$). Several respondents provided additional feedback. Multiple GC-engaged HTCs acknowledged that their GC is "very involved and highly valuable" and that their "program would be lost without their GC." One respondent remarked that the GC at their HTC "adds substantial benefit to the patient care that we provide for both established patients as well as their families and new patients. A GC is an essential part of the HTC team." Another respondent mirrored this by adding that having a GC available "is ideal for optimal patient care."

Barriers Employing a GC within HTCs

Due to skip-logic formatting, only responses categorized as non-GC-engaged (n=65) were included in this analysis. GC-engaged and GC-referrals did not answer this question, therefore are not included in this analysis (table 4).

26.2% of respondents identified the main barrier to employing a GC in their HTC as "a limited or uncertain perceived benefit"; 23.1% identified a lack of funding as the main barrier, and 20% noted other needs were a priority. Other reported barriers were staff concerns about knowledge and understanding of bleeding disorders on the part of the GC, space limitations, lack of success in recruiting a GC, and inability to replace a GC. Three respondents noted that they do not have enough patients to warrant hiring a full-time genetic counselor at their HTC.

Of those respondents who reported a lack of funding as a barrier to employing a GC in their HTC, 85.7% answered that they had not attempted to obtain funding for a GC (Figure 5). The other 14.3% chose "other", noting that the genetics department within their institution is short staffed and genetic counselors are "unable to dedicate time for this endeavor."

DISCUSSION

When GCs are utilized within HTCs, they are perceived as valuable and as having a unique skill set, consistent with findings from a survey of GC practice variation among cystic fibrosis centers (Langfelder-Schwind, et al., 2019). In particular, they are found to be most effective in providing genetic education to patients and families, coordinating genetic testing, facilitating informed consent, and disclosing, interpreting, and discussing implications of genetic testing results. In spite of this, our results suggest GCs are not being utilized by nearly a third of HTCs. Agreement about GC value and unique skill set was lower among HTCs without a GC as part of the care team. A lack of awareness among providers who have not worked closely with a GC in the past may contribute to the undervaluing of the potential contributions of GCs to multidisciplinary care. Defining how to effectively integrate GCs within HTCs could lead to an increase in GC utilization in this setting.

Our results demonstrate variation in the level of GC-engagement between the ten regions of the U.S. HTC network as well as within a single region (Figure 2). The regions reporting the highest GC-engagement align with results from the NSGC Professional Status Survey (PSS) identifying a higher number of GCs practicing within the states that make up the Mid-Atlantic (n=294), Great Lakes (n=253), and Northern States (n=276) regions (NSGC, 2020). Surprisingly, the Western States region reported the least amount of GC-engagement, in spite of having the highest number of practicing GCs (n=322) on the PSS (NSGC, 2020).

Disparities in the provision of genetic counseling creates unequal access to genetics expertise. HTCs are expected to provide or facilitate multidisciplinary comprehensive care to patients. Not having a GC on staff to address concerns places additional burden on HTC staff or unnecessary inconvenience if patients must seek and coordinate separate genetic counseling

appointments outside the HTC, potentially leading to lack of follow through. HTCs who have unsuccessfully attempted to recruit a local GC may consider the use of telegenetics to improve access to GCs. Patients often appreciate the convenience of telemedicine and report feeling more at ease in the comfort of their own homes (Vrecar et al., 2016).

It is crucial for patients with hereditary bleeding disorders to receive an accurate and prompt diagnosis which may be complicated by the genetic complexity of these conditions, unclear clinical testing results, and overlapping clinical features (Swystun, et al., 2016). Molecular diagnosis can provide diagnostic clarity to patients and their families in addition to facilitating proper management and treatment options. However, this requires accurate and thorough genetic counseling to ensure appropriate genetic testing is ordered and interpreted correctly. It is unrealistic to expect that non-genetics providers will have sufficient, ongoing and up-to-date genetics training as well as the time to provide a high level of care to patients (Farmer, et al., 2019). GCs have extensive training in assessing, ordering, and interpreting molecular genetic testing thereby providing them with the ideal expertise to significantly reduce ordering errors and unnecessary costs thereby optimizing patient care (Miller, et al., 2014; Foil, et al., 2018).

Our study reveals that those HTCs who do not utilize the services of a GC most commonly cite a limited or uncertain perceived benefit and lack of funding. The clear benefit and value noted by HTC providers who work closely with GCs suggests a need for increased awareness and further education across HTCs regarding the benefits of employing a GC within the HTC. It is noteworthy that the majority of respondents who reported a lack of funding had not attempted to obtain funding for a GC in their HTC. Although our survey did not assess

perspectives of GC-engaged HTCs relating to funding, Langfelder-Schwind et. al found that GC-engaged CFCs did not perceive having a GC to be cost-prohibitive.

Limitations

Our study has several limitations. A previously validated survey tool was not available for our study, so an original web-based survey was created. Several questions were designed to specifically reflect the ACGC practice-based competencies. The overall sample size of this study was relatively small which limited our ability to perform some statistical analyses.

The majority of responses were from GC-engaged HTCs which may demonstrate ascertainment bias as they may have been more likely to respond to this survey. Although there were multiple completed surveys from each region within the US HTC Network, our results are not representative of all 149 accredited HTCs. Of note, responses may be duplicated if there were multiple respondents from the same institution. This particular study focused on collecting information regarding the utilization of GCs, barriers to employing GCs, and providers' perceived value of GCs within HTCs. For this reason, patient perspectives were not assessed but could be informative for future studies.

Due to skip-logic formatting, only responses categorized as non-GC-engaged had the opportunity to answer questions relating to barriers of employing a GC within their HTC. In retrospect, it would have been informative to present this question to all respondents to assess the perspectives of GC-engaged and GC-referral HTCs to ascertain any barriers and issues with funding they experienced.

FUTURE DIRECTIONS

Despite the limitations of this study, our results highlight the need to reevaluate our clinical care models. Further investigation into the barriers of employing a GC in HTCs is

warranted to effectively address and improve access to GCs for individuals with hereditary bleeding disorders.

The contrast between HTC provider perspectives of a GC's value and unique skill set supports the need for increased education. For example, presentations to hereditary bleeding disorder providers can be utilized to promote the positive implications of incorporating a GC in our care models. In addition, future research focusing on the responsibilities of a GC within an HTC, patient perspectives of genetic counseling services received within HTCs and GC knowledge of hereditary bleeding disorders would be beneficial.

CONCLUSIONS

This study highlights significant variation in the provision of genetic counseling to patients and families between and within regions of the U.S. HTC Network. While there have been significant advances in genetic testing, our clinical models of care delivery have remained static; this has important implications for GC practice within HTCs. When GCs are integrated, HTCs endorse their high level of understanding of hereditary bleeding disorders, the unique and valuable services they provide, and support their inclusion as a part of the HTC core team. Reducing inconsistencies in the provision of genetic counseling may increase healthcare equity and optimize patient care.

In light of the rapid evolution of the genetic counseling field, advancements in the diagnosis and treatment of hereditary bleeding disorders, and the results of this study, consideration should be given to update recommendations for inclusion of a GC to the HTC core team.

CONFLICT OF INTEREST

All authors declare that they have no conflict of interest.

HUMAN STUDIES AND INFORMED CONSENT

All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinski Declaration of 1975, as revised in 2000 (5). Informed consent was obtained from all respondents for being included in this study.

Figure 1 Respondents Roles Within the HTC

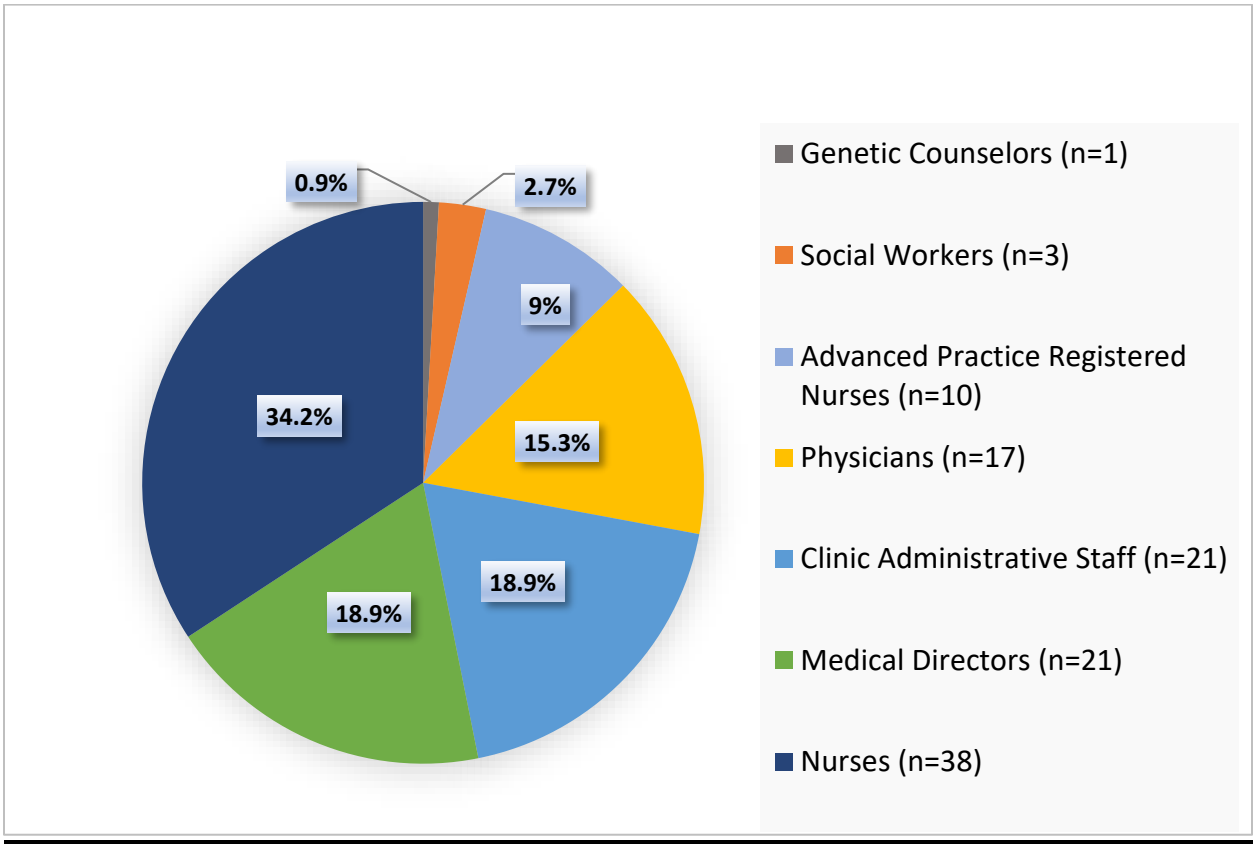
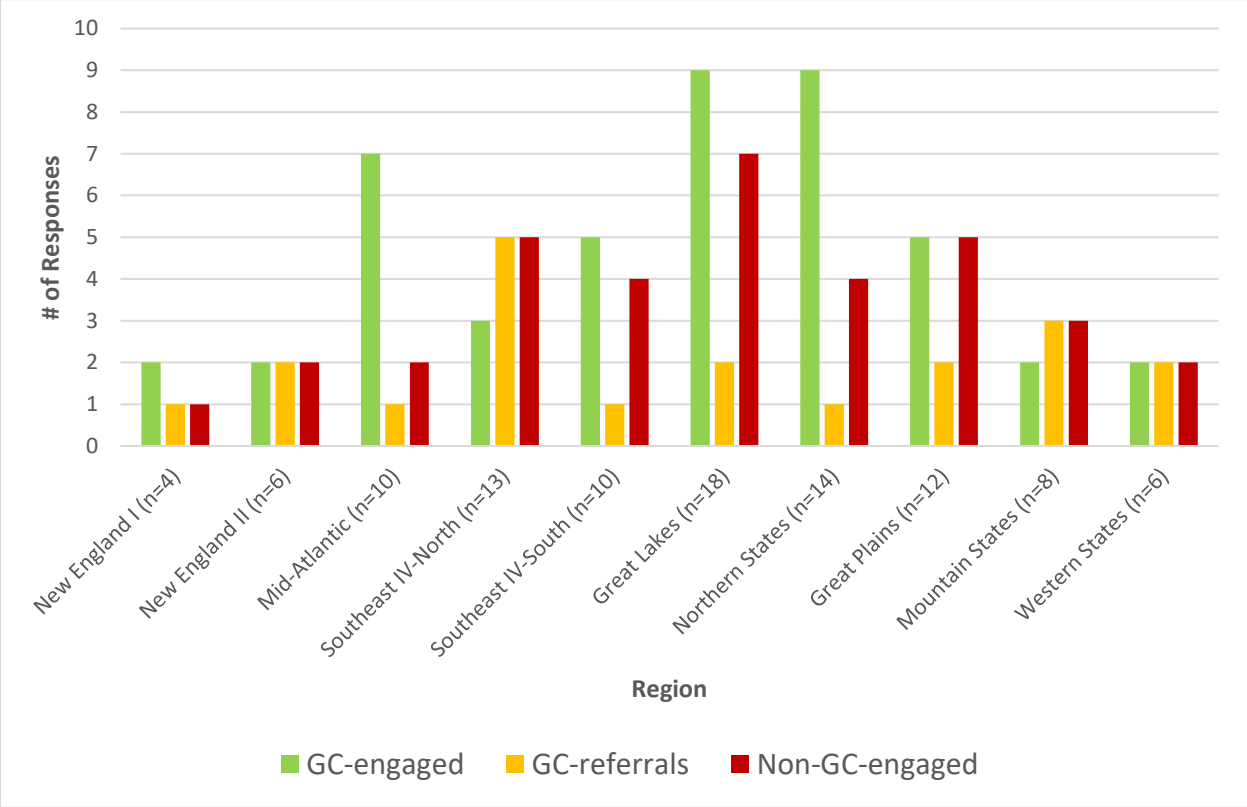
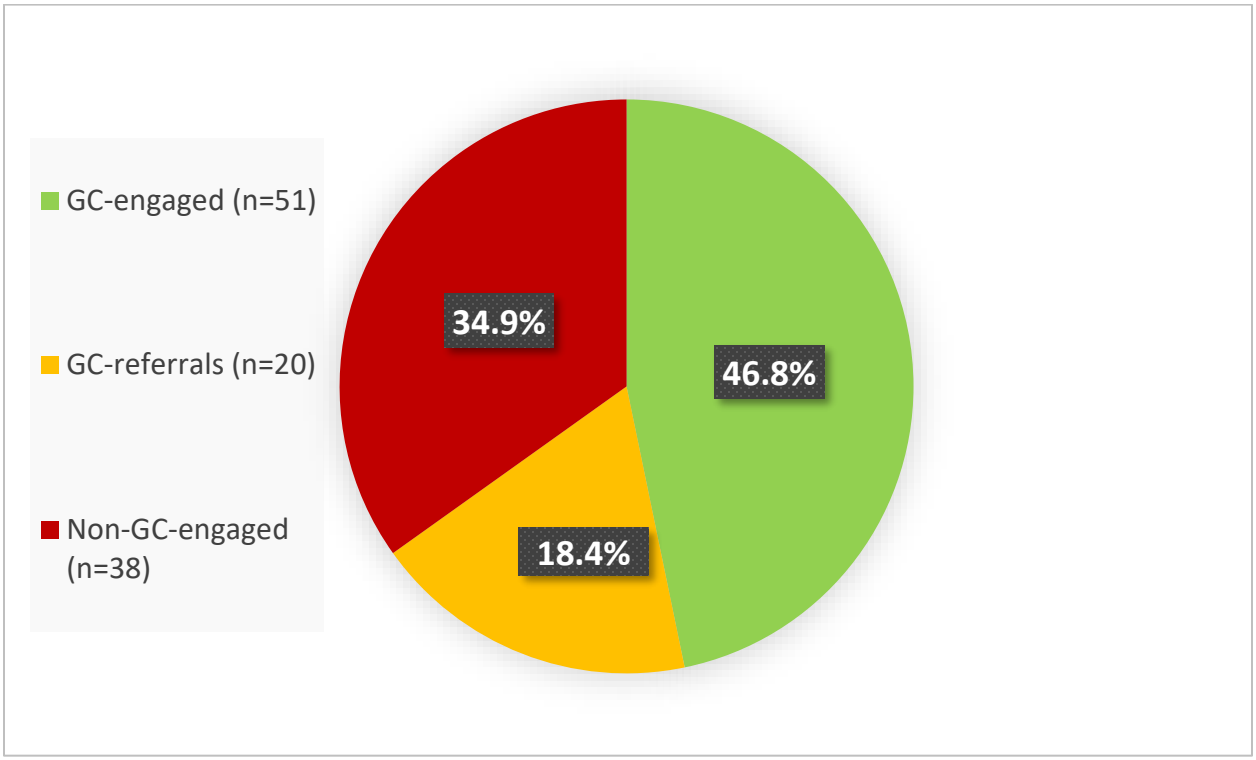


Figure 2 Variation of GC Utilization Among HTCs Across the Ten Regions of the U.S. HTC Network



Note: Total number of HTCs in each region are as follows: New England I (10), New England II (13), Mid-Atlantic (17), Southeast IV-North (14), Southeast IV-South (17), Great Lakes (21), Northern States (16), Great Plains (19), Mountain States (11), Western States (13).

Figure 3 Responses Stratified by Level of GC-engagement Within the HTC



Note: GC-engaged: a GC is employed by the HTC or a parent institution and regularly staffs the HTC; GC-referral: a GC is not directly employed by the HTC or institution, and does not routinely staff the clinic, but referrals are provided as needed; non-GC-engaged: a GC is not employed by the HTC or institution nor are referrals made

Table 1 HTC Provider Responses Regarding GC Competencies Stratified by Level of Engagement

Competency	Response	All (n=42) n (%)	GC-engaged (n=25) n (%)	Non-GC-engaged (n=17) n (%)	p-value
Demonstrates and utilizes a depth and breadth of understanding and knowledge of genetics core concepts and principles.	Agree Disagree Neutral	42 (100) 0 (0) 0 (0)	25 (100) 0 (0) 0 (0)	17 (100) 0 (0) 0 (0)	-
Tailors, translates, and communicates relevant information in a clear and unambiguous manner for a broad range of audiences based on their needs, characteristics, circumstances, and educational background.	Agree Disagree Neutral	41 (98) 0 (0) 1 (2)	25 (100) 0 (0) 0 (0)	16 (94) 0 (0) 1 (6)	.405
Shows skill in research and self-education, including identifying medical literature, experts, and relevant information to answer questions.	Agree Disagree Neutral	39 (93) 0 (0) 3 (7)	25 (100) 0 (0) 0 (0)	14 (82) 0 (0) 3 (18)	.059
Possesses strong interpersonal skills, resulting in successful patient relationships while maintaining appropriate boundaries, as well as positive relationships with professionals across health care disciplines to promote effective teamwork and care delivery.	Agree Disagree Neutral	41 (98) 0 (0) 1 (2)	25 (100) 0 (0) 0 (0)	16 (94) 0 (0) 1 (6)	.405
Exhibits understanding of healthcare systems and delivery models to promote responsible, equitable and cost-effective integration of genetic services to improve patient care.	Agree Disagree Neutral	38 (91) 0 (0) 4 (9)	25 (100) 0 (0) 0 (0)	13 (77) 0 (0) 4 (23)	.021
Constructs and/or assesses relevant, targeted and comprehensive personal and family histories and pedigrees for analysis of probability of bleeding disorders or carrier status based on pedigree, medical records review, test results, and other pertinent information.	Agree Disagree Neutral	42 (100) 0 (0) 0 (0)	25 (100) 0 (0) 0 (0)	17 (100) 0 (0) 0 (0)	-
Evaluates potential impact of psychosocial issues on decision making and adherence to medical management.	Agree Disagree Neutral	34 (81) 2 (5) 6 (14)	22 (88) 1 (4) 2 (8)	12 (71) 1 (6) 4 (23)	.384

Table 1 (Continued)

Competency	Response	All (n=42) n (%)	GC-engaged (n=25) n (%)	Non-GC-engaged (n=17) n (%)	p-value
Employs active listening, interviewing, and basic counseling skills to identify, assess and empathetically respond to concerns.	Agree Disagree Neutral	41 (98) 0 (0) 1 (2)	25 (100) 0 (0) 0 (0)	16 (94) 0 (0) 1 (6)	.405
Educates clients about a wide range of genetics and genomics information.	Agree Disagree Neutral	39 (93) 0 (0) 3 (7)	25 (100) 0 (0) 0 (0)	14 (82) 0 (0) 3 (18)	.059
Provides a unique service to our HTC families.	Agree Disagree Neutral	36 (86) 1 (2) 5 (12)	25 (100) 0 (0) 0 (0)	11 (65) 1 (6) 5 (29)	.002
As a staff member in the HTC, they possess a high level of understanding of hereditary bleeding disorders, including but not limited to etiology, clinical features, disease expression, and natural history.	Agree Disagree Neutral	36 (86) 1 (2) 5 (12)	25 (100) 0 (0) (0)	11 (65) 1 (6) 5 (29)	.002
Based on my experience, I believe that, ideally, a genetic counselor should be part of an HTC team.	Agree Disagree Neutral	35 (83) 1 (2) 6 (14)	25 (100) 0 (0) 0 (0)	10 (59) 1 (6) 6 (35)	.001

Note: Due to skip logic formatting respondents categorized as GC-referrals did not answer these questions. Only responses categorized as GC-Engaged and Non-GC-Engaged were included in this analysis. Some questions were not answered by all 115 respondents due to skip logic formatting and inclusion of partially completed surveys; reported n values are variable for this reason.

Table 2 HTC Provider Responses Regarding Issues to Ideally be Addressed by a GC Stratified by Level of Engagement

Issues Addressed by GC	Response	All (n=42) n (%)	GC-engaged (n=25) n (%)	Non-GC-engaged (n=17) n (%)	p-value
Explaining genetic inheritance and reproductive recurrence risks of the bleeding disorder.	Agree Disagree Neutral	39 (93) 0 (0) 3 (7)	25 (100) 0 (0) (0)	14 (82) 0 (0) 3 (18)	.059
Describing how the underlying genetic change results in the bleeding disorder and the potential impact on the individual and family.	Agree Disagree Neutral	38 (91) 0 (0) 4 (9)	25 (100) 0 (0) 0 (0)	14 (82) 0 (0) 3 (18)	.286
Discussing test utilization strategy/selection of genetic testing.	Agree Disagree Neutral	40 (95) 0 (0) 2 (5)	25 (100) 0 (0) 0 (0)	15 (88) 0 (0) 2 (12)	.158
Obtaining informed consent for clinical genetic testing.	Agree Disagree Neutral	36 (86) 0 (0) 6 (14)	24 (96) 0 (0) 1 (4)	12 (71) 0 (0) 5 (29)	.032
Obtaining informed consent for studies (research or industry-funded) that return genetic results.	Agree Disagree Neutral	32 (76) 1 (2) 9 (21)	21 (84) 0 (0) 4 (16)	11 (65) 1 (6) 5 (29)	.188
Addressing questions and concerns about gene therapy and/or informed consent for gene therapy.	Agree Disagree Neutral	30 (71) 2 (5) 10 (24)	19 (76) 0 (0) 6 (0)	11 (65) 2 (12) 4 (23)	.283
Disclosing genetic test results.	Agree Disagree Neutral	40 (95) 0 (0) 2 (5)	25 (100) 0 (0) 0 (0)	15 (88) 0 (0) 2 (12)	.158
Explaining the implication of genetic test results, including variant classification, variants of uncertain significance, and further testing recommendation for the patient and/or family members.	Agree Disagree Neutral	41 (98) 0 (0) 1 (2)	25 (100) 0 (0) 0 (0)	16 (94) 0 (0) 1 (6)	.405

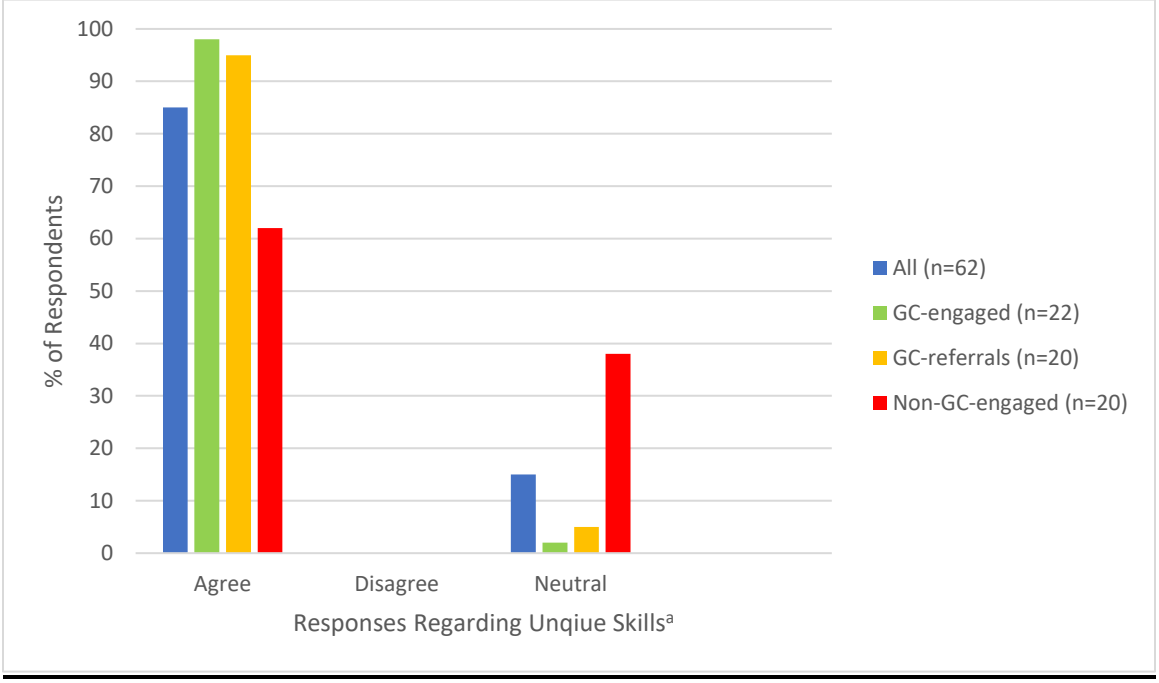
Note: Due to skip logic formatting respondents categorized as GC-referrals did not answer these questions. Only responses categorized as GC-Engaged and Non-GC-Engaged were included in this analysis. Some questions were not answered by all 115 respondents due to skip logic formatting and inclusion of partially completed surveys; reported n values are variable for this reason.

Table 3 HTC Provider Responses Regarding Skills a GC Can Add to an HTC Stratified by Level of Engagement

Skills a GC can add to an HTC	Response	All (n=62) n (%)	GC-engaged (n=22) n (%)	GC-referrals (n=20) n (%)	Non-GC-engaged (n=20) n (%)	*p*-value
Explaining genetic inheritance and reproductive recurrence risks of the bleeding disorder.	Agree Disagree Neutral	60 (97) 1 (1) 1 (1)	22 (100) 0 (0) 0 (0)	20 (100) 0 (0) 0 (0)	18 (90) 1 (5) 1 (5)	.201
Describing how the underlying genetic change results in the bleeding disorder and the potential impact on the individual and family.	Agree Disagree Neutral	56 (90) 3 (5) 3 (5)	20 (91) 0 (0) 2 (9)	19 (95) 1 (5) 0 (0)	17 (85) 2 (10) 1 (5)	.441
Discussing test utilization strategy/selection of genetic testing.	Agree Disagree Neutral	60 (98) 0 (0) 1 (2)	22 (100) 0 (0) 0 (0)	20 (100) 0 (0) 0 (0)	18 (95) 0 (0) 1 (5)	.311
Obtaining informed consent for studies (research or industry-funded) that return genetic results.	Agree Disagree Neutral	43 (72) 3 (5) 14 (23)	17 (81) 2 (10) 2 (10)	13 (68) 0 (0) 6 (32)	13 (65) 1 (5) 6 (30)	.255
Addressing questions and concerns about gene therapy and/or informed consent for gene therapy.	Agree Disagree Neutral	37 (66) 7 (13) 12 (21)	8 (42) 5 (26) 6 (32)	16 (89) 1 (6) 1 (6)	13 (68) 1 (5) 5 (26)	.029
Disclosing genetic test results.	Agree Disagree Neutral	60 (97) 0 (0) 2 (3)	22 (100) 0 (0) 0 (0)	20 (100) 0 (0) 0 (0)	18 (90) 0 (0) 2 (10)	.201
Explaining the implication of genetic test results, including variant classification, variants of uncertain significance, and further testing recommendation for the patient and/or family members.	Agree Disagree Neutral	61 (98) 0 (0) 1 (2)	22 (100) 0 (0) 0 (0)	20 (100) 0 (0) 0 (0)	19 (95) 0 (0) 1 (5)	.645
Our staff members at our HTC are capable of providing the functions of a genetic counselor for our patients.	Agree Disagree Neutral	22 (36) 26 (42) 14 (23)	6 (27) 11 (50) 5 (23)	5 (25) 11 (55) 4 (20)	11 (55) 4 (20) 5 (25)	.148

Note: All responses were included in this data analysis (GC-Engaged, GC-referrals, and Non-GC-Engaged). Some questions were not answered by all 115 respondents due to skip logic formatting and inclusion of partially completed surveys; reported n values are variable for this reason.

Figure 4 HTC Provider Responses Regarding Unique Skills of a GC



Note: All responses were included in this data analysis (GC-Engaged, GC-referrals, and Non-GC-Engaged). Some questions were not answered by all 115 respondents due to skip logic formatting and inclusion of partially completed surveys; reported n values are variable for this reason.

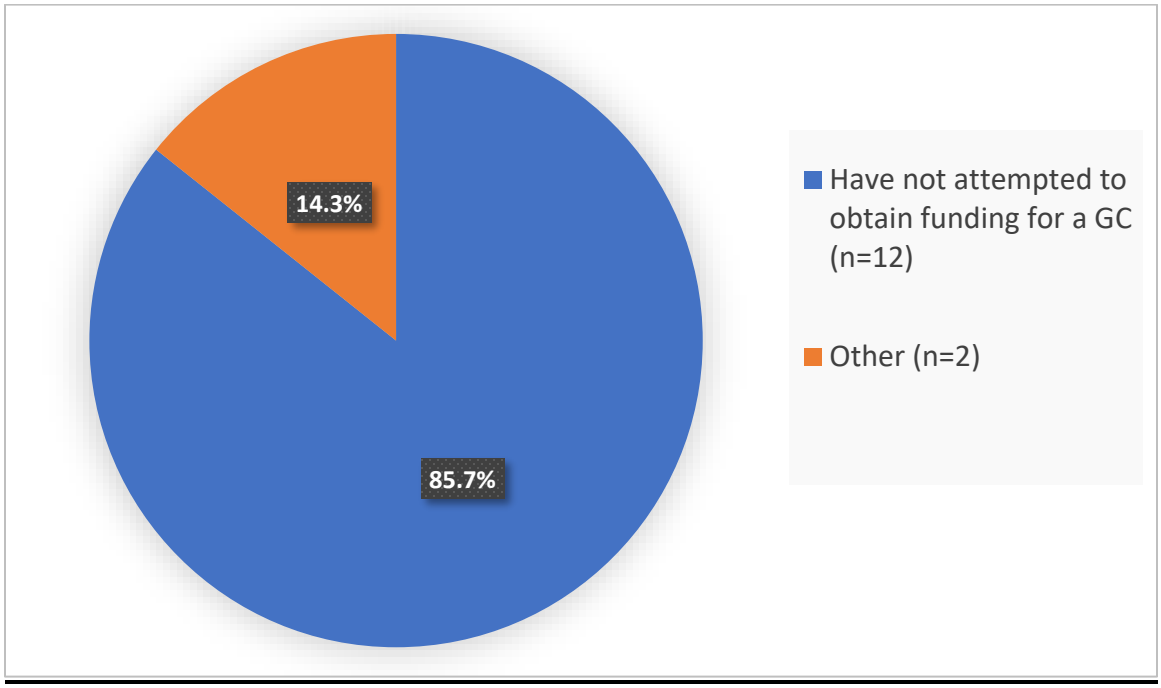
[a]The above reflects responses from the following statement: "I feel that a genetic counselor has a unique skill set that is highly valuable to an HTC clinic."

Table 4 Barrier to Employing a GC Within the HTC

Main Barriers	(n=65) n (%)
Lack of funding	15 (23.1)
Limited/uncertain perceived benefit	17 (26.2)
Staff concerns about knowledge and understanding of bleeding disorders on the part of the genetic counselor	3 (4.6)
Space limitations	5 (7.7)
Attempted unsuccessfully to recruit/hire a genetic counselor	4 (6.2)
Unable to retain/replace a genetic counselor	4 (6.2)
Currently other needs are a priority for our HTC	13 (20.0)
Other	4 (6.2)

Note: Some questions were not answered by all 115 respondents due to skip logic formatting and inclusion of partially completed surveys; reported n values are variable for this reason. This data includes responses categorized as non-GC-engaged only.

Figure 5 Description of the lack of funding for a GC within the HTC



Note: Some questions were not answered by all 115 respondents due to skip logic formatting and inclusion of partially completed surveys; reported n values are variable for this reason. This data includes responses categorized as non-GC-engaged only and those reporting a lack of funding as a major barrier to employing a GC within the HTC.

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