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Genetic Counselor Roles in the Undiagnosed Diseases Network Research Study: Clinical Care, Collaboration, and Curation

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Abstract

Genetic counselors (GCs) are increasingly filling important positions on research study teams, but there is limited literature describing the roles of GCs in these settings. GCs on the Undiagnosed Diseases Network (UDN) study team serve in a variety of roles across the research network and provide an opportunity to better understand genetic counselor roles in research. To quantitatively characterize the tasks regularly performed as well as professional fulfillment derived from these tasks, two surveys were administered to UDN GCs in a stepwise fashion. Responses from the first, free-response survey elicited the scope of tasks which informed development of a second structured, multiple-select survey. In survey 2, respondents were asked to select which roles they performed. Across 19 respondents, roles in survey 2 received a total of 947 selections averaging approximately 10 selections per role. When asked to indicate what roles they performed,

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Author Contributions

Authors JNK and EGK confirm that they had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis. All of the authors gave final approval of this version to be published and agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Conflict of Interest

Jennefer N. Kohler, Emily G. Kelley, Brenna M. Boyd, Catherine H. Sillari, and Shruti Marwaha declare that they have no conflict of interest.

Matthew T. Wheeler has equity interest in Personalis Inc (Menlo Park).

Human Studies and Informed Consent

This study was reviewed and granted an exemption by the Stanford University Institutional Review Board (protocol number: 57530). All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Implied informed consent was obtained for individuals who voluntarily completed the online survey and submitted their responses.

Animal Studies

No non-human animal studies were carried out by the authors for this article.

respondent selected a mean of 50 roles (range 22–70). Survey 2 data were analyzed via thematic coding of responses and hierarchical cluster analysis to identify patterns in responses. From the thematic analysis, 20 non-overlapping codes emerged in seven categories: clinical interaction and care, communication, curation, leadership, participant management, research, and team management. Three themes emerged from the categories that represented the roles of GCs in the UDN: clinical care, collaboration, and curation. Cluster analyses showed that responses were more similar among individuals at the same institution than between institutions. This study highlights the ways GCs apply their unique skill set in the context of a clinical translational research network. Additionally, findings from this study reinforce the wide applicability of core skills that are part of genetic counseling training. Clinical literacy, genomics expertise and analysis, interpersonal, psychosocial and counseling skills, education, professional practice skills, and an understanding of research processes make genetic counselors well suited for such roles and poised to positively impact research experiences and outcomes for participants.

Keywords

Genetic counseling; genetic counselors; professional development; expanded roles; research

Background

The field of genetic counseling has grown rapidly in recent years with the increasing integration of genomics into clinical research and medicine (Baty, 2018; Ormond, 2013; Ormond et al., 2018). The number of certified genetic counselors in the United States has more than doubled over the past 10 years, and the scope of practice has expanded since the profession's inception to include more roles in clinical, research, government, non-profit, and industry settings (National Society of Genetic Counselors (NSGC), 2020). As the era of genomic medicine and precision health continues to evolve, the roles of genetic counselors will continue to expand and diversify (Bamshad et al., 2018) while the core skill set will remain unique and in-demand across practice settings (Kohut et al., 2019).

The domains of practice-based competencies for genetic counselors (genetics expertise and analysis; interpersonal, psychosocial, and counseling skills; education; professional development and practice) (Accreditation Council for Genetic Counseling (ACGC), 2019) are clearly applicable across diverse settings. However, a comprehensive understanding of how genetic counseling competencies are applied in expanded roles is lacking. Genetic counselor roles in the public health and genomic testing industry sectors have been delineated in recent years, where studies have shown that genetic counselor core competencies are applied in new ways (Goodenberger et al., 2015; Goodenberger et al., 2017; McWalter et al., 2015; McWalter et al., 2018). For example, a laboratory genetic counselor applies genetics expertise and analysis to ensure appropriate test ordering; public health genetic counselors utilize education and interpersonal skills when performing educational outreach to providers and the public (Goodenberger et al., 2015; McWalter et al., 2015). A genetic counselor working in an industry setting versus a traditional clinical setting may be more likely to speak with patients independent of physician involvement; utilize telehealth to communicate information; contribute to new test

development; and implement innovative approaches such as chatbots, informational videos, brief communications approaches, or group pre-counseling to increase throughput (Hallquist et al., 2021; McWalter et al., 2018; Waltman et al., 2016).

Genetic counselors' variant interpretation skill set, a relatively recent addition to many genetic counseling programs' core curricula, is increasingly recognized as essential in multidisciplinary healthcare settings where specialists may not have expertise in genetics (Reuter et al, 2018). Beyond the clinical setting, these skills are fundamental to the increasing role of laboratory-based genetic counselors (Goodenberger et al., 2017; Swanson et al., 2014), whose services have been shown to both prevent inappropriate testing and result in substantial savings for hospitals and insurance companies (Wakefield et al., 2018).

Genetic counselors are well situated in research settings due to clinical expertise and research training, both requirements of genetic counseling training programs. In the 2020 National Society of Genetic Counselors' Professional Status Survey, 20% of genetic counselors indicated research as a primary professional role, and 38% of genetic counselors reported some level of involvement in research (NSGC, 2020). While the potential positive impact of incorporating genetic counselors into translational research teams was explored in 2011 (Zierhut & Austin), there remains little research describing how genetic counselors are fulfilling these roles (Biesecker, 2018; Kohut et al., 2019). Thus a clear opportunity and mandate exist to characterize the responsibilities of genetic counselors in the research setting.

Genetic counselors are integral in precision medicine where each patient is approached differently based on their unique presentation, as is done in the Undiagnosed Diseases Network (UDN) (Bamshad et al., 2018; Wicklund et al., 2018). The UDN is a research study that was established to provide a multidisciplinary diagnostic approach for the evaluation of diagnostically challenging cases and to identify the underlying mechanisms of newly described diseases (Splinter et al., 2018). Funded by the National Institutes of Health (NIH) along with participating academic institutions, the UDN was formed in 2014 and expanded in 2018 to consist of 12 clinical sites, one sequencing core, one metabolomics core, one central biorepository, two model organism screening centers, and one coordinating center (Undiagnosed Diseases Network, 2021). Detailed descriptions of the UDN research study have been published previously; briefly, adults and children with undiagnosed conditions may apply to the UDN, and 40% of applications are accepted as participants to enroll in the study (Splinter et al., 2018; Undiagnosed Diseases Network, 2021). Participants are typically evaluated via a multidisciplinary in-person visit with various specialists at one of the 12 clinical sites, spanning one to five days in length. The personalized evaluation typically also includes family-based clinical exome or genome sequencing and research exome and/or genome analysis. Follow-up research may include RNA sequencing of relevant tissues, metabolomics analysis, laboratory functional studies for candidate genes/variants, matchmaking via PhenomeCentral and public web pages, and creation of model organisms to provide evidence for variant pathogenicity in novel genes not yet associated with human disease. Participants in the UDN may receive a diagnosis during medical record review in the application phase, during the in-person evaluation, or days, weeks, months, or years after the evaluation. Thirty-five percent of participants in the study have received a diagnosis, and

dozens of new conditions have been identified (Splinter et al., 2018; Undiagnosed Diseases Network, 2021).

In the UDN, certified genetic counselors are members of the team at each clinical site, sequencing core, metabolomics core, and the coordinating center. The UDN Genetic Counseling and Testing Working Group (GCT WG) launched at the beginning of the study to advise development of consent forms, medically actionable findings policies, and return of results protocols. Over the past five years, the GCT WG has continued to provide input on study protocols and publish findings (Macnamara et al., 2019; Palmer et al., 2018), as well as serve as a monthly peer supervision group for genetic counselors and study coordinators.

The authors, together with the GCT WG, determined that characterizing the diverse roles of board-eligible and board-certified genetic counselors in the UDN would be useful in quality assurance/improvement. This study was therefore undertaken with the specific aim of characterizing the diverse roles of genetic counselors in the UDN.

Methods

Participants

Board-eligible or board-certified genetic counselors (hereafter referred to as genetic counselors) who were contributing to the UDN research study in any capacity at the times of the surveys were invited to voluntarily participate in this study.

Instrumentation

We sought to evaluate the roles and responsibilities of genetic counselors in the UDN using an iterative approach with two non-validated surveys that were developed by genetic counselors specifically for the purpose of this study. Survey 1 was exploratory in nature and meant to guide the development of survey 2, which aimed to comprehensively capture results in a structured manner. Questions focused on roles within the UDN study. Demographic questions were also asked, including years in practice and percent time dedicated to the UDN research study. “Genetic counseling” was intentionally not offered as a role option in the survey in an effort to allow for a nuanced assessment of which specific genetic counseling roles were being performed.

Procedures

This protocol was deemed exempt by the Stanford University Institutional Review Board (protocol number: 57530). In April 2018, survey 1 was distributed among members of the UDN GCT WG via email-based form after agreement from group members at a routine UDN GCT WG meeting. Members of the UDN GCT WG were asked to distribute survey 1 to any genetic counselors at their site who were not members. Survey 1 asked respondents to list up to five skills-based roles that they consistently performed in their roles in the UDN, categorized within 14 domains (see supplemental materials). Domains were based on the chronological stages of UDN research processes (e.g., application, evaluation, follow-up) as well as additional network and research components. Responses were collated and duplicate

or similar roles were collapsed by study author (JK) in an effort to create a list of discrete roles that were used to populate survey 2.

Survey 2 was developed in a multiple-select format by authors (JK, EK) based on the responses from survey 1 and was administered via Qualtrics (Qualtrics, Provo, UT) between January and March 2019. Demographic questions included clinical site, time with the UDN in months, and percentage of time on UDN as defined by salary. During data analysis, additional demographics were collected from respondents via email to include years in practice and the number of genetic counselors and site coordinators at each site at time of survey 2.

The survey 2 link was emailed to members of the Genetic Counseling and Testing Working Group and sent separately to any genetic counselors in the UDN who were not members. Up to four email reminders were sent to non-respondents over the course of six weeks. Organized by domain, respondents were asked to select (check boxes next to) all the roles they consistently perform in their current position as a genetic counselor in the UDN. Two free-text questions asked respondents to list the top five roles in which they feel most of their time is spent and the top roles that are most fulfilling to them. A free-text comments box was provided at the end of the survey for respondents to share any additional thoughts.

Data analysis

This exploratory research was conducted in a case study framework as a means to collect and present detailed information about a group of genetic counselors in a specific setting (Cresswell & Poth, 2018; Crowe et al., 2011).

Upon conclusion of data collection, roles were further collapsed or removed from the dataset for analysis if there was no selection, redundancy, limited possibility for interpretation (e.g., “perform duties as required”), or involved non-UDN professional development. The final dataset consisted of 96 roles falling into 20 codes across seven categories which were distilled into three main themes (see Table 2) in a process consisting of four phases of initialization, construction, rectification, and finalization (Vaismoradi et al., 2016).

To characterize the scope of the roles of genetic counselors in the UDN, roles were assigned to categories and themes in an iterative approach due to the exploratory and descriptive nature of this research. Three members of the study team (two genetic counselors and one non-genetic counselor study coordinator) developed an initial codebook by assigning a code to each role from survey 2. Codes were assigned equal levels of specificity and importance in a flat coding frame, rather than a hierarchical coding frame which relates codes to each other. Roles were then independently coded by three coders where roles were assigned a primary code, and a secondary code was assigned if the primary code was not felt to fully capture the role. To ensure coding reliability, codes were reviewed by three independent coders to determine consensus and a set of 20 non-overlapping codes were finalized. Codes were assigned descriptions through the consensus discussions and related codes were assigned to one of seven categories, which were distilled into three themes.

For an additional independent analysis of how role selections were related to each other, hierarchical clustering was performed on the above data that consisted of a matrix of 96 roles (listed under the 20 roles in the description column of Table 2) performed by the 19 respondents. The clustering algorithm grouped roles according to how often they are performed together. Similarly, respondents were clustered based on similarity of the roles they performed. Hierarchical clustering was run in R programming language (R version 3.6.0) (R Core Team, 2020). Euclidean distance was selected as a measure of distance and complete linkage was used to calculate the dissimilarity between clusters. To find the optimal number of clusters, consensus clustering (Monti et al., 2003) was applied in each case using the ConsensusClusterPlus package (version 1.48.0) (Wilkerson & Hayes, 2010) in R. The results of consensus clustering suggested using six to eight clusters for respondent analysis and 8 to 10 clusters for roles analysis; eight clusters were determined most fitting for both analyses (respondents and roles) based on manual analysis of the clusters identified. The manual analysis of the eight clusters assigned descriptive titles to each cluster for interpretation purposes. See Figure 1 for a summary of study procedures.

Results

Respondent characteristics

Fourteen genetic counselors representing all seven UDN clinical sites and the UDN Coordinating Center responded to survey 1 (response rate: 64%, 14/22). At the time of survey 1, the majority of respondents had been working on the UDN project for more than two years (n=11/14, 79%). Most respondents split their time between the UDN project and other professional responsibilities (n=8/14, 57%).

For survey 2, 19 out of 20 (95%) total eligible genetic counselors responded. Respondents again represented all seven clinical sites and the Coordinating Center. Twelve out of 14 survey 1 respondents responded to survey 2. Most survey 2 respondents had been working on the UDN project for more than two years (n=14/19, 74%), had been practicing as a genetic counselor for at least five years (n=13/19, 68%), and designated “genetic counselor” as their primary job title (n=12/19, 63%). Nine of 18 respondents split their time between the UDN project and other professional roles (50%; one respondent did not answer this question). Of the eight UDN sites represented by survey 2 respondents, six had more than one genetic counselor working on the study site’s team (range: 1–4). Respondent demographics of gender identity, race, and ethnicity were not collected.

Survey responses

A total of 378 roles were reported by survey 1 respondents. After removing duplicates and collapsing similar responses, 102 roles remained which were re-administered in survey 2.

Nearly all roles were selected by at least one respondent in survey 2; a single role was not selected by any survey 2 respondents (“Manage social media content relevant to UDN and local-site projects”) and was removed from subsequent analysis. Four roles were collapsed into two due to redundancy. Finally, another three roles were removed due to lack of

specificity (n=1) and lack of relevance to UDN work (n=2). A total of 96 roles were included in subsequent analyses.

Across the 19 respondents, roles in survey 2 received a total of 947 selections averaging approximately 10 selections per role. One role had the highest number of selections (n=18, "Participate in writing/reviewing manuscripts from UDN team") whereas two shared the lowest (n=1, "Act as participant advocate by accompanying them through most parts of their in-person visit" and "Manage UDN technology support operations"). On average, respondents selected 50 roles each with a range of 22–70.

Coding and Thematic Analysis

Twenty non-overlapping codes emerged from the data. A single code was assigned to each of 87 roles, while nine roles were assigned two codes each because it was felt that a single code did not sufficiently represent the role. The code receiving the highest number of selections (when normalized for the number of roles assigned per code) was "psychosocial counseling," which involves psychosocial support related to health conditions and risks, providing anticipatory guidance, and facilitating decision-making based on elicited values and goals of the participant.

"Clinical genetics" and "genomic testing management" codes also represented roles that were frequently selected by respondents. These involve activities such as eliciting family histories, overseeing genomic testing plan and ordering, results interpretation, and communication of results to clinical team and to participants.

Finally, roles coded "network liaising" were selected more on average compared to those falling under other codes. Network liaising reflects a variety of responsibilities and efforts that maintain connections among the multi-institutional UDN. These involve participating in network-wide meetings and working groups as well as managing individual communications with research cores, the Coordinating Center, and other clinical sites.

The 20 codes fell into seven categories: clinical interaction and care, communication, curation, leadership, participant management, research, and team management (Table 2). Three themes emerged from the categories that represented the roles of genetic counselors in the UDN: clinical care, collaboration, and curation.

Additional responses

When asked which roles provided most fulfillment, survey 2 respondents largely selected those involving clinical interaction and care, such as psychosocial counseling and discussing genetic findings. When asked where most of their time was spent, respondents listed a variety of roles with a slight overrepresentation of roles involved in participant management and curation of medical and research data.

Cluster analyses

Cluster analyses were performed to objectively explore associations between (1) respondents, and (2) roles in survey 2 data. The respondent dendrogram demonstrated closer relationships among genetic counselors at the same site rather than by degrees held or years

in practice (Figure 2). This indicates that genetic counselors at each UDN site tend to have more similar roles to each other than to genetic counselors at other sites. There did not appear to be association by highest degree achieved, years in the field, nor percent time allocated to the UDN, though it is possible that associations may be masked by the small sample size.

The role dendrogram grouped individual roles that were often performed by the same respondent (Figure S1). Unlike the coding and thematic analysis, these role groupings are influenced in part by their frequency of respondent selection. Upon manual review by the study team, six of the eight clusters reflected a clear theme: genomic testing coordination, genetic counseling, internal and external liaising, study coordination, leadership, and project management. Two groups, “genetic counseling” and “internal and external liaising,” included roles with the most selections, indicating these functions are most characteristic of UDN genetic counselors overall. In contrast to the seven role categories generated in the coding and thematic analysis, which primarily stem from shared underlying skill sets (e.g., genomics expertise, communication), these six clusters reflect groups of activities that tend to be performed by the same person and frequently involve multiple skill sets.

Discussion

This study provides a cross-sectional characterization of genetic counselors’ roles in a large, multi-site clinical research study. In this setting, genetic counselors’ roles differ across and within institutions, but were found to share similar core themes of collaboration, clinical care, and curation.

Out of 19 genetic counselors in the UDN that completed survey 2, half of the respondents split their time between the UDN study and other professional roles (50%). Institutions differed in their team structures, with genetic counselors at some sites performing study coordination activities and others specializing their involvement to primarily direct patient care, genomic analysis, and knowledge dissemination. While most (63%) had the primary job title of “genetic counselor”, the remainder had a range of titles--such as principal investigator, project manager, and director--reflecting varied ways in which genetic counselors’ skill sets are utilized in a multi-institutional research study.

Clusters

Exploration of associations between respondents and roles provided additional insight into professional contributions of UDN genetic counselors. A cluster analysis showed that genetic counselors’ roles clustered most closely by clinical site, indicating that genetic counselors at the same site had more similar responsibilities to each other than to genetic counselors at other sites. Each clinical site in the network (represented by 17 respondents) performs the same study activities as required by their NIH grants. Therefore, it is likely that the differences between genetic counselors’ roles primarily reflect site-specific organizational structures and application of genetic counselors’ skills within the context of their team environment. The second cluster analysis grouped roles that were often performed together and resulted in eight clusters, six of which represented key role categories: genomic testing coordination, genetic counseling, internal and external liaising, study coordination,

leadership, and project management. While research genetic counselors across the network are involved in a breadth of activities, these clusters suggest a degree of role specialization of UDN genetic counselors. Specializing in role types such as project management can allow for deep subject-area expertise in a research endeavor, but may not maximize genetic counselors' unique combination of skills.

Themes

The most performed roles share a core theme of communication, which demonstrates the primary responsibility of a genetic counselor in many settings and reflects the strong communication skill sets as outlined in the practice-based competencies. Genetic counselors are trained to communicate complex information across diverse audiences including patients, physicians, and researchers, and can utilize these skills in multiple content areas.

Collaboration, a core element of the UDN study, requires ongoing communication management. Genetic counselors actively communicate with local teams, the network, external collaborators, and with participants, sharing information most relevant to each. When communicating with international collaborators about planned functional studies or a potential matching patient, for example, the content differs significantly from when communicating with families. Roles coded as network liaising were selected more compared to other roles, reflecting the communication requirement of collaborative research. Large-scale research necessitates frequent cross-collaboration since multidisciplinary teams of experts come together to work on one project, and clear communication across collaborators is crucial for research progress.

UDN genetic counselors frequently performed roles involving synthesis of information, where large amounts of clinical, genomic, and/or research data must be understood and interpreted before disseminating to the appropriate recipients. This reflects an intersection of domain expertise and communication skill. Synthesizing and sharing relevant information continues to become more critical in genomic medicine as more conditions are identified and more data are generated (Riconda et al., 2018).

Clinical care and curation roles were also strongly represented in the data. "Clinical care" encompasses roles directly related to participants' UDN evaluations, including psychosocial counseling and other direct interaction with participants and families. Codes falling into this theme most closely resemble activities that are performed in a clinical setting--but applied in the context of a research study. Psychosocial counseling, for example, is a core component of genetic counseling and can be directly applied in the context of rare disease research. Given the level of uncertainty inherent to the undiagnosed and rare disease populations investigated as well as the longitudinal nature of the UDN study, genetic counselors in the UDN may find more opportunity for providing counseling and support, often with a longer duration of involvement with participants and families, than in a clinical setting. Similarly, discussions around risks and benefits of genetic testing in the UDN study resemble those in clinical settings; however, in the context of the UDN they are expanded to include risks and benefits of participation in the study as a whole. It is notable that the "psychosocial counseling" code received the highest number of selections after normalization for number of roles per code, indicating this is a valuable skill in a research setting such as the UDN.

It is not surprising that clinical care is a strong theme among roles of UDN genetic counselors. Genetic counselors are trained to be clinical providers involved in direct patient care and to understand research processes (Accreditation Council for Genetic Counseling, 2019). Furthermore, roles falling within the “clinical care” theme, including psychosocial counseling, bring high levels of professional fulfillment to UDN genetic counselors. When asked to list roles providing the most fulfillment, 14 of 17 (82%) respondents in survey 2 listed at least one activity involving direct participant care. We suspect this finding is not specific to UDN genetic counselors nor research genetic counselors generally but would be consistent among most genetic counselors with frequent patient interaction regardless of professional environment. Helping others has been identified as a common motivation for joining the genetic counseling field (Stoddard et al., 2021), and likely remains a source of professional fulfillment.

Finally, curation of information was a common theme among UDN genetic counselors, pertaining to clinical, genomic, and multi-omic data. Curation involves review, organization, and summarization of information most pertinent for the task at hand. Research with the UDN frequently requires creative and critical thinking, as working with participants with rare disease means pushing the boundaries of current gene-disease relationships and the use of novel diagnostic analyses. It requires a thorough understanding of the information to determine what information is most useful, as well as an ability to synthesize and distill it to diverse audiences. Genetic counselors are suited for such tasks given their medical literacy, training in statistics, genomics expertise, and communication skill set.

Interestingly, the individual role most performed by genetic counselors in the UDN was related to participation in writing and reviewing manuscripts. Genetic counselors in the UDN have led research studies, published on psychosocial issues in this research population, and authored various clinical reports (Cope et al., 2020; McConkie-Rosell et al., 2019; Schoch et al., 2020; Macnamara et al., 2019). Our findings that genetic counselors in the UDN are regularly involved in research project development, data analysis, and dissemination of results leads us to recommend additional training opportunities such as rotations in research settings to support development of these skills outside of thesis projects. One such research training experience is the genomics rotations developed at Stanford; in these rotations, for example, students develop plans for functional analysis of genomic candidates beyond what is available clinically, interact with external collaborators, and participate in manuscript preparation for cases they have worked on (Grove et al., 2019; Geng et al., 2019). These provide an opportunity for students to develop skills in genomic curation, translational science, and multidisciplinary collaboration. Research development, implementation, and dissemination capitalize on the interpersonal and collaborative skills that are key characteristics of successful genetic counselors.

Overall, the roles delineated in surveys 1 and 2 reflect the various ways genetic counseling skills are applied in the UDN research context. Key themes identified by this study align well with the domains of practice-based competencies for genetic counselors defined by ACGC (genetics expertise and analysis; interpersonal, psychosocial, and counseling skills; education; professional development and practice). Many roles performed by UDN genetic counselors overlap those performed in a traditional clinical setting, while others represent

genetic counseling competencies applied in nontraditional ways. For example, analysis of raw genomic and multi-omic data is typically not performed in a clinical setting, but relies upon a core skill in genetics expertise and analysis. Similarly, liaising with research collaborators is not common in clinical practice yet is grounded in strong interpersonal skill and professional practice to maintain interdisciplinary relationships and ensure research collaborations are conducted in an ethically sound manner. Roles performed by UDN genetic counselors demonstrate the relevance of genetic counseling competencies in this research setting, as well as ways these skills can be further developed to provide value in a translational research project.

Limitations

While this study represented 95% of genetic counselors in the UDN at the time of survey 2, the small sample size of 19 respondents limits its generalizability to other research and non-research genetic counselors. Only genetic counselors in the UDN were surveyed, so this study was not able to make comparisons between genetic counselors in other settings. Other factors may be associated with roles and responsibilities that were not captured by the data collected (e.g., years in practice or involvement with training programs) or that were not found to be associated by the cluster analysis due to limited power. Although the roles were found to be associated with clinical site location by the cluster analysis, this study was unable to draw conclusions about causality. Roles outside of UDN activities were not assessed and, since 50% of respondents split their time between the UDN and other roles, this study is limited in its ability to provide a holistic understanding of the professional activities of genetic counselors who spend some of their time working in research settings.

Implications for clinical practice / Future directions

This study highlights the ways genetic counselors apply their unique skill set in the context of a multi-site clinical research study. Findings emphasize the importance of variant interpretation skills particularly as exome and genome sequencing become more widespread and information about variant-gene-disease information grows, and variant curation and interpretation skills are increasingly being incorporated into genetic counseling training (Grove et al., 2019; Hooker et al., 2014). Additionally, findings from this study reinforce the wide applicability of core skills that are part of genetic counseling training. The landscape and goals of genetic counseling have evolved over time with the growing needs of the field, and professional roles have expanded outside the clinic. However, skills related to psychosocial counseling, genomics expertise, communication, and interpersonal skills remain central (Stoll et al., 2018). Exposure to expanded genetic counselor roles, such as those in research or industry settings, will be valuable in genetic counseling training to help trainees understand how to apply their skills in different settings. Genetic counselors in research and other expanded roles should be proactive in hosting trainee rotations as much as possible.

Future studies should survey genetic counselors across a wider variety of research studies and settings. Surveying genetic counselors across specialties (e.g., cancer, prenatal, cardiology) as well as setting (e.g., clinical, research, industry) would add to the overall

understanding of similarities and differences in professional roles of genetic counselors. Longitudinal studies assessing how individual genetic counselors' roles change over time as individual projects, study phase, and team structure evolve may further illuminate the differences found between sites in this study.

Conclusions

This study provides an in-depth evaluation of the roles of genetic counselors working in different positions on one large research study, finding that genetic counselors utilize their skills in collaborating, data curation, and clinical care. In this multi-institutional network study, roles were most similar among genetic counselors at the same site, which may be due to variation in site operational structuring and genetic counselors fulfilling specific needs of the study team. To our knowledge, this study is the first of its kind to characterize the varying and complex roles of genetic counselors in a clinical research setting. Genetic counselors in this study are frequently managing collaborations with diverse clinical and research groups. Clinical literacy, genomics expertise and analysis, interpersonal, psychosocial, and counseling skills, and an understanding of research processes make genetic counselors uniquely suited for such roles.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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Julian A. Martínez-Agosto

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Data Availability Statement

The data that support the findings of this study are available from the corresponding author upon request.

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What is known about this topic:

Genetic counselors are increasingly working in professional settings including research, but little is known about how genetic counselors fulfill these roles.

What this paper adds to the topic:

This paper provides insight into the professional roles and responsibilities of genetic counselors working in a multi-site research study, and further examines how core competencies of genetic counseling are applied in such roles. This evidence informs our understanding of genetic counselors' professional contributions and may inform future efforts in professional development.

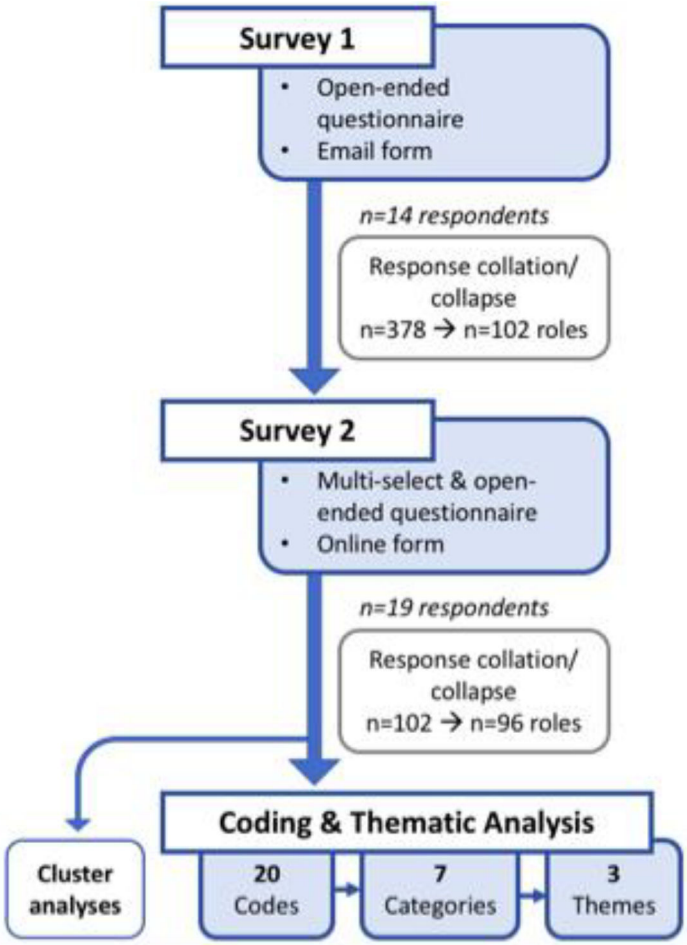

Figure 1: Study Procedures
Flowchart summarizing study procedures and analyses.

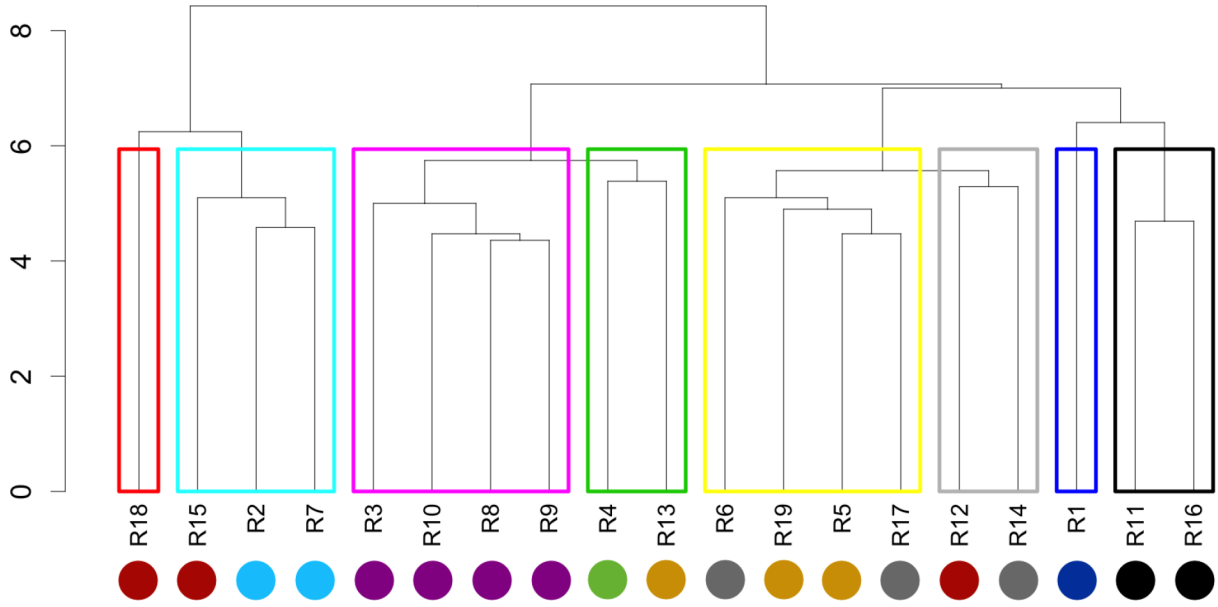

Figure 2: Clustering by Respondent

Hierarchical clustering of survey 2 respondents. Respondents are grouped by similarity of roles performed. Colored boxes delineate individual clusters (k=8). R1-R19 refer to individual respondent IDs (n=19). Colored circles beneath respondent IDs reflect the respondent's UDN site: Site 1 (red) =R18, R15, R12; Site 2 (light blue)=R2, R7; Site 3 (purple)=R3, R10, R8, R9; Site 4 (green)=R4; Site 5 (gold)=R13, R19, R5; Site 6 (grey)=R6, R17, R14; Site 7 (dark blue)=R1; Site 8 (black)=R11, R16.

Table 1:

Demographics (Survey 2)

Years practicing as a genetic counselor:	
<5 years	6
5–10 years	4
11–15 years	6
>15 years	3
Years working on UDN study (n=19):	
0–2 years	4
2–4 years	5
>4 years	10
Percent time allocated to UDN work (n=18):	
<50%	2
50%	4
51–99%	3
100%	9

Table 2:

Description of codes assigned to roles

Themes	Categories	Codes	Description
Clinical care	Clinical interaction & care	Consent	Obtain informed consent for UDN protocol and substudies
		Direct patient care	Accompany participants throughout the research visit, communicate recommendations and clinical test results to participants, including non-genetic findings
		Clinical genetics	Elicit family history, discuss genetic testing plan, return results, discuss management and treatment options
		Psychosocial counseling	Provide psychosocial counseling, elicit goals, provide anticipatory guidance, and facilitate decision-making
Clinical care	Participant management	Case management	Manage, coordinate, and/or communicate information related to specific participants or applicants, including participating in the development of the evaluation plan and overseeing participant progress in study
		Coordination	Facilitate, organize, schedule, and communicate study participation from the application to in-person evaluation
Collaboration	Communication	Network liaising	Activities and communications related to network-level work groups, committees, and meetings
		Outreach	Present to local and national groups to educate about UDN application, coordinating local grand rounds presentations, and participating in recruitment efforts
		Provider/ researcher communication	Communicate pertinent clinical or research information for expert consultation, to support transition of care, or to pursue a candidate diagnosis.
		Team communications	Communicate pertinent clinical or research information to a multidisciplinary team for application review, coordination of clinical evaluation, genomic or research results, and team projects
Collaboration	Leadership	Student professional development & mentorship	Formal and informal teaching and communications related to the professional education of trainees, particularly those pursuing advanced degrees (MS, MD, and PhD).
Collaboration	Participant Management	Data entry	Oversee and perform data entry into UDN database from application to follow-up
		Advocacy	Assist participants with accessing appropriate logistical, financial, and/or support resources and interact with participant advocates to optimize study experience
		Matchmaking and external data sharing	Manage external data sharing in public databases and matchmaking tools (e.g., UDN participant pages and GeneMatcher), involving synthesis of clinical and genomic information and evaluating potential matches
Collaboration	Team management	Personnel management	Hire and/or supervise site personnel (study coordinators, volunteers, research assistants/interns)
		Project management	Manage, coordinate, and/or communicate information related to site- or network-level functioning, including oversight of protocols, workflows, IRB compliance, and progress
Curation	Curation	Curation of research data	Assist in raw genomic data analysis, interpretation of -omics data, and participation in bioinformatic tool development
		Genetic testing management	Oversee genetic testing ordering, communication, analysis and reanalysis
		Medical record review	Interpret and summarize medical records from applicants and participants
Curation	Research	Research design, implementation, and dissemination	Develop and execute sub-studies, including grant writing, as well as disseminate research findings in manuscripts, presentations, and posters