I, Courtney Lewis, hereby submit this original work as part of the requirements for the degree of Master of Science in Genetic Counseling.

It is entitled:
Genetics Laboratory Directors’ Perspectives on the Role of Genetic Counselors in Acquired Mutation Testing: Current and Expanded Opportunities

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Genetics Laboratory Directors’ Perspectives on the Role of Genetic Counselors in Acquired Mutation Testing: Current and Expanded Opportunities

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By

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ABSTRACT

**Background:** Advances in genetic testing have enabled the characterization of recurrent acquired mutations, which are now recognized as part of the classification criteria for certain hematopoietic malignancies. The genetic counselor’s (GCs) role in genetic testing for constitutional mutations has been well established; however, the opportunity for GCs to apply their skills to acquired mutation testing has not been previously studied. The purpose of this study was to describe the roles for laboratory GCs in acquired mutation testing as well as discuss training to help prepare GCs for these roles.

**Methods:** An online survey to assess the current and expanded roles for GCs regarding genetic testing for acquired mutations related to oncogenesis was sent to 387 genetics laboratory directors. To be eligible for the study, participants had to identify as directors working in laboratories offering genetic testing services for acquired mutations.

**Results:** Thirty-nine participants met eligibility criteria and completed the survey; of which, 20 (Group A) currently employed a GC and 19 (Group B) did not employ a GC. Time spent on acquired mutation testing accounted for 1-25% of the GCs total effort in the laboratory. Acting as a provider resource was the most common role currently performed by laboratory GCs related to acquired mutation testing and GC roles were reported to be similar to those related to constitutional mutation testing. The two groups significantly differed on their level of agreement regarding the role of GCs in acquired mutation testing, with Group A agreeing more strongly compared to Group B (p=0.05). A GC master’s training program and on the job training were cited as the best routes to prepare GCs for a role in acquired mutation testing.

**Conclusion:** Laboratory GCs are currently performing roles related to acquired mutation testing. There is support for an expanded GC role in this area, particularly as perceived by directors who currently employ GCs. Additionally, this work supports GC training programs incorporating didactic coursework on laboratory-based genetic counseling that may include topics to prepare GCs for a role in testing for acquired mutations.

**Key Words:** genetic counseling, genetics laboratory, acquired mutations, oncogenesis, genetic counseling training
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INTRODUCTION

Cancer is one of the leading causes of mortality in the world.\textsuperscript{1,2} The proportion of deaths attributed to cancer is a major healthcare concern and exploring methods (e.g., early detection, genetic profiling, targeted therapeutics) to reduce cancer mortality rate is a key clinical and research focus.\textsuperscript{1} Cancer is caused by an accumulation of genetic mutations in a group of cells. Acquired mutations, or genetic alterations that are not present in the germline, can accumulate in a single cell and lead to unregulated mitotic cell division resulting in cancer development, or oncogenesis. If the accumulation of these mutations does not trigger the cell to enter apoptosis, the mutations can confer a selective advantage for unregulated cell division and promote the development of blood vessels to supply nutrients to the tumor.\textsuperscript{3}

Certain types of acquired mutations play a key role in the initiation and progression of a cancer and can be analyzed through molecular and cytogenetic methods.\textsuperscript{1,4} Acquired mutations that occur in genes that are important for regulation of the cell cycle (i.e., proto-oncogenes, tumor suppressor genes, and DNA repair genes) can precipitate oncogenesis.\textsuperscript{1} For example, consistent mutations have been well described in hematopoietic cancers and The World Health Organization now includes cytogenetic profiling in the assessment of diagnosis and prognosis for patients with hematopoietic malignancies, such as acute myeloid leukemia.\textsuperscript{5, 6, 7, 20}

Since the identification of the Philadelphia chromosome in 1960, the development of genetic testing and interpretation methods have enabled the identification of acquired mutations associated with oncogenesis in malignant cells.\textsuperscript{8} Cancer genomic profiling can aid in medical management of a patient by 1) guiding treatment, 2) providing alternative therapies if drug resistance occurs, and 3) improving the implementation of individualized medicine
practices. The development and availability of complex genetic tests is expected to increase with advancing technology. In an article about the future of personalized oncology, Ross (2011) reported up to a 50% growth rate for gene-based and molecular diagnostic testing. Additionally, in the next decade, Ross (2011) proposed that the clinical application of molecular diagnostics will transform the application of personalized oncology practice. It is imperative to explore how specialized genetics healthcare professionals, such as genetic counselors, can assume an intermediary role between genetic testing laboratories and ordering providers.

Genetic counselors (GCs) are trained to educate patients and healthcare professionals about the genetic contributions to disease. Among many responsibilities, GCs assist in risk assessment, discuss genetic testing options, and synthesize information to help guide informed medical management decisions. Training in hereditary cancer predisposition syndromes is a foundational aspect of the graduate-level curriculum for GCs; however, training in acquired mutations has not been historically incorporated into the training curriculum for GCs. Genetic counselors play an integral role in assessing risk and coordinating genetic testing for inherited cancer predisposition in the traditional clinic setting. Several studies have underscored the importance of GCs in non-traditional settings, specifically, genetic testing laboratories.

Over the past decade, the proportion of laboratory-based GCs has nearly doubled, from 6% to 11.2%. This increase in laboratory-based employment opportunities for GCs seems to indicate that genetics laboratory directors, the primary decision makers for hiring and staffing of laboratory personnel, have identified specific needs that are fulfilled by the expertise of trained GCs. Furthermore, this trend emphasizes the importance of understanding the current
responsibilities for laboratory GCs as well as identifying opportunities for expanded growth from the laboratory director’s perspective.

Recognizing the complexities of the genetic testing process, several studies have supported the integration of GCs into the laboratory setting.\textsuperscript{12, 13, 14, 17, 18} A range of roles for laboratory GCs has been described; however, the existing literature had yet to describe the role of laboratory GCs in acquired mutation testing or to elicit the perspective of genetics laboratory directors on the role of laboratory GCs. Among the most common roles for GCs in laboratory, acting as a liaison for the ordering provider, calling out results, discussing testing logistics are the most commonly cited. A study to assess the interactions between genetic counselors and molecular genetics laboratories found that nearly 60\% of clinical genetic counselors contact the laboratory prior to ordering a test to inquire about specimen requirements, turnaround time, and cost of testing.\textsuperscript{19} Many genetic counselors in this study also reported contacting the laboratory after receiving test results to obtain clarification about the interpretation information included in a test report (83\%), information about testing methods (82\%), interpretation of results and implications of negative results (39\%).\textsuperscript{19} Over half of respondents (57\%) indicated they speak with a laboratory genetic counselor before sending a specimen to a testing facility as opposed to interacting with another laboratory employee (i.e., client services employee (19\%), laboratory director (16\%), clinical consultant for the laboratory (12\%), or laboratory supervisor (7\%)).\textsuperscript{19} Overall, these studies have concluded that laboratory GCs not only act as a physician resource in guiding testing decisions, but assume a variety of roles at all stages of the testing process.
While it has been well established that GCs have a prominent role in the laboratory setting, the GC’s role specifically in acquired mutation testing has not previously been studied. This study is the first to describe the role of genetic counselors in acquired mutation testing. The purpose of this study is to describe the current and expanded roles for laboratory GCs in acquired mutation testing and identify potential training mechanisms to prepare GCs for a role in acquired mutation testing.
MATERIALS AND METHODS

Participants and Procedures

Genetics laboratory directors (director, assistant director, or associate director) working in a laboratory offering genetic testing for acquired mutations related to oncogenesis met inclusion criteria for this study. Individuals who did not identify as laboratory directors or genetics laboratory directors who did not work in laboratories offering genetic testing for acquired mutations related to oncogenesis were excluded from the analysis. Genetics laboratory directors were identified by: 1) querying the Genetic Testing Registry (GTR) hosted by the National Center for Biotechnology Information (NCBI) and 2) accessing a list of American Board of Medical Genetics (ABMG) Accredited Laboratory Training Programs. Study results contains self-reported data from the perspective of genetics laboratory directors.

The terms “cytogenetics”, “molecular” and “genetic” were queried under the “Lab” search tab in the GTR during May 2013. The results were filtered by laboratories in the United States only. Available contact information for laboratory directors listed under the resulting laboratories and the ABMG list was abstracted. Attempts were made to obtain missing contact information by searching the director’s name on academic or commercial websites. Duplicated laboratory directors resulting from the three GTR queries combined with the list of ABMG directors were removed prior to finalizing the list of prospective participants. SurveyMonkey automatically removed individuals who previously opted out of completing online surveys hosted through their site. Using the GTR infrastructure, it was not feasible to determine which laboratory directors worked in laboratories offering genetic testing for acquired mutations prior to survey dissemination. An eligibility assessment question was included at the start of the
online survey to exclude those who did not work in laboratories offering genetic testing for acquired mutations at the time survey responses were collected. This study was approved by Cincinnati Children’s Hospital Medical Center (CCHMC) and the University of Cincinnati Institutional Review Boards (CCHMC IRB#: 2013-3007)

Survey Development

The survey tool was developed to assess the perspective of genetics laboratory directors on current and expanded roles for genetic counselors regarding genetic testing for acquired mutations related to oncogenesis. The survey tool was developed and reviewed by the authors and pre-tested for validity and reliability by seven colleagues (i.e., genetic counselors, training program faculty, and peers) in the Division of Human Genetics (DHG) at CCHMC. Individuals who met eligibility criteria for the study were asked to complete one of two versions of the survey depending on whether they employed a genetic counselor in their laboratory. Laboratory directors who currently employed a genetic counselor in their laboratory completed APPENDIX B and laboratory directors who did not employ a genetic counselor at the time of the survey completed APPENDIX C. The components of both surveys fell into the following domains: 1) evaluation of a GC’s role in genetic testing for acquired mutations 2) description of knowledge and training content that may be relevant to preparing GCs for a role in genetic testing for acquired mutations, 3) laboratory characteristics regarding testing for acquired mutations and 4) participant demographics. The roles of a GC described in the survey were developed through consultation with clinical and laboratory personnel in the DHG at CCHMC, literature review, and review of the NSGC Scope of Practice statement.11, 14, 17
Survey Measures and Dissemination

The first domain of the survey assessed laboratory director’s perspectives on either the current or future scope of a GC’s role in genetic testing for acquired mutations. For laboratory directors who reported working with a GC in their laboratory (Group A), the questions in this domain addressed: 1) the proportion of time GCs dedicate to acquired mutation testing, 2) strength of agreement regarding a role for GCs in acquired mutation testing (scaled from strongly agree to strongly disagree), 3) the roles genetic counselors currently perform related to acquired mutation testing and the range of time spent on each role and 4) the participant’s perspective on whether GCs should be performing a set of roles related to acquired mutation testing (yes, no, I don’t know). For laboratory directors who did not report working with a GC in their laboratory (Group B), the questions in this first domain addressed similar themes but in a hypothetical manner as well as assessed reasons why the laboratory did not currently employ a genetic counselor.

The remaining three survey domains were the same between Groups A and B. The second survey domain assessed the knowledge base and training necessary for GCs to have a role in genetic testing for acquired mutations. An open-ended question was included in this section to allow researchers to gather additional input. The third domain assessed the following characteristics of the laboratory: genetic testing volume (acquired vs. constitutional), incidence of incidental findings and follow-up measures, number of laboratory personnel, sample population (adult, pediatric, both), and laboratory certification status (CLIA, CAP, etc). The last domain of the survey gathered information about respondent demographics, which included: gender, race, ethnicity, professional title, specialty, and certification.
Genetics laboratory directors from our compiled list were invited by email to complete the online survey hosted through SurveyMonkey. Recipients were able to opt out of receiving future invitations for study participation. Non-responders received the invitation to participate one and two weeks following the initial dissemination of the survey. Respondents who did not meet the eligibility criteria for the study were sent to a disqualification page with a thank you message. Respondents who met the eligibility criteria for the study and completed the online survey were sent a follow-up email with an electronic gift card.

**Data Analysis**

Survey responses were collected in January 2014. Survey responses in SurveyMonkey were exported and organized into SPSS (v. 22) for data analysis. Data were summarized using mean and standard deviation or median and range (depending on data distribution) for continuous variables and percentage and frequency for categorical variables. Open-ended questions were post-coded to identify common themes and the frequency for these themes was tabulated.

The level of agreement (assessed using a 10-point scale ranging from strongly disagree to strongly agree) regarding the role of laboratory GCs in acquired mutation testing was compared between respondents who currently employ a GC (Group A) and those who do not employ a lab GC (Group B). During study design, authors hypothesized that Group A would rank the role of GCs in acquired mutation testing higher (agree more strongly) than Group B. After assessing the normality of the data distribution, the non-parametric Kruskall-Wallis Test (1-way analysis) was performed to assess a statistical difference between the two groups.
RESULTS

Participant Demographics and Laboratory Characteristics

In total, 39 laboratory directors met the eligibility criteria for the study and completed the online survey. An additional 19 invited individuals expressed interest in study participation, but did currently not work in laboratories offering genetic testing for acquired mutations and were excluded (13 were disqualified during the online survey and 6 contacted the study contact person directly indicating they did not offer genetic testing for acquired mutations). The overall response rate for the study was 15% (58/387). Upon excluding directors who self-identified as ineligible for study participation, our inclusion rate was 10% (39/387). Since it was not feasible to determine which laboratory directors worked in laboratories offering genetic testing for acquired mutations prior to survey dissemination, an accurate calculation of the response rate from our eligible population is difficult to determine.

Twenty (51.2%) respondents reported working with at least one GC in their laboratory and 19 (48.7%) respondents did not currently work with a GC. Eighty-five percent of respondents were laboratory directors and 43.6% of the total participants considered themselves the primary decision maker regarding staffing and hiring in their laboratory. Molecular genetics was the most common specialty (71.8%) reported and the majority (41%) of respondents indicated they had been Board-certified between 6-10 years. The mean age of participants was 47.5 years (SD 10.2). In our sample population, 61.5% of respondents self-identified as Caucasian and 61.5% reported their gender as female (Table 1).

All laboratories were CLIA certified and 84.6% were CAP accredited. Laboratories that employed a GC had a median workforce that was over three-times larger when compared with
laboratories without a GC, and the mean number of GCs employed by the laboratories was 2.8 (SD 2.1). One-third (30.8%) of respondents indicated that testing for acquired mutations accounted for >51% of the total services in the laboratory. The majority of laboratories represented in the study reported receiving fewer than 50 (43.6%) or between 50-100 samples (18.4%) for acquired mutation testing per month. Most laboratories reported receiving fewer than 50 orders for acquired mutation testing per month and samples were generally received from both pediatric and adult patients (Table 2).

**Current Roles for GCs in Acquired Mutation Testing**

The majority (65%) of respondents who reported working with a GC indicated that GCs dedicate between 1-25% of their total time in the laboratory to acquired mutation testing. Furthermore, one respondent indicated that ≥26% of the GC’s time in their laboratory was dedicated to acquired mutation testing (Table 3). Laboratory directors who work with a GC (n=20) indicated the most common roles GCs currently perform related to genetic testing for acquired mutations included: acting as a provider resource (80%), discussing incidental constitutional abnormalities with healthcare providers (70%), clarifying test orders (70%), calling out test results to the ordering provider (60%), collecting clinical data related to the case (60%), developing patient materials (60%), coordinating research studies (55%), performing administrative duties (50%) and marketing (50%) (Figure 1). Thirty (79%) laboratory directors indicated they encounter at least one incidental reportable constitutional abnormality each year during acquired mutation testing, while 7.7% (n=3) reported ≥21 incidental constitutional abnormalities per year (Table 4).
For the most common roles currently performed related to acquired mutation testing, the percent effort dedicated to each role was most often reported as 1-25%. In addition, some respondents indicated that GCs in their laboratory dedicate >50% of their time related to acquired mutation testing on the following five roles: discussing incidental constitutional abnormalities with healthcare providers (5%), calling out test results to the ordering provider (5%), collecting clinical data related to the case (10%), developing patient materials (5%), and marketing (5%) (Table 5).

**Expanded Roles for GCs in Acquired Mutation Testing**

Laboratory directors who work with a GC were given the option to indicate whether a given role was not currently performed by GCs in their lab, but could be performed related to acquired mutation testing. Additionally, directors who did not work with a GC were asked to indicate which roles they felt GCs could perform related to acquired mutation testing. Common roles cited as increased opportunities for laboratory GCs in acquired mutation testing included: calling out test results to the ordering provider, collecting clinical data related to the case, discussing incidental constitutional abnormalities with healthcare providers, acting as a provider resource, coordinating research studies and clarifying of test orders (Figure 2).

Of the 14 lab GC roles related to acquired mutation testing delineated in the survey, three roles were commonly cited as not relevant for GCs (as reported by directors who currently employ a lab GC) or not cited as possible opportunities for lab GCs (as reported by directors who do not employ a lab GC). Ninety-seven percent of respondents (n=38) indicated signing laboratory reports was either not a relevant role or not a possible role for lab GCs in acquired mutation testing (Figure 3). Additionally, the majority of respondents indicated that
creating laboratory reports (51%) and interpreting test results (49%) related to acquired genetic abnormalities were not relevant GC roles (Figure 3).

Training for GCs to Prepare for an Expanded Role in Acquired mutation Testing

Respondents were asked to rank the importance (1= most important, 10= least important) of educational topics for GCs to learn to expand their role in acquired mutation testing. The four most important topics for GCs to learn about as reported by laboratory directors included interpretation of pathology reports related to acquired mutation testing, natural history of cancer, psychosocial skills, and understanding of sample types amenable to testing (Table 6). Conversely, the four least important topics included patents for acquired mutation testing, flow cytometry, psychosocial skills and bench protocols for carrying out acquired mutation analysis (Table 6).

To learn about the most important topics related to acquired mutation testing, laboratory directors most often cited training during a genetic counseling master’s program (41%) or on the job training (38%) as the best modes of training. A small proportion of respondents indicated obtaining an advanced research degree (3%) or certificate training (8%) as the best training for GCs to learn about acquired mutation testing topics (Figure 4). Respondents were given the opportunity to provide qualitative responses regarding additional training and/or educational opportunities for GCs to expand their role in acquired mutation testing. Upon post-coding the data for themes, four primary themes were identified among the 10 respondents who provided qualitative responses (Table 7). The most common themes included: more education in genetics and genomics (n=4), teaching/interacting with other
healthcare providers (n=4), attending conferences (n=3), and a combination of on the job training and additional master’s course work (n=2) (Table 7).

Differences in the Level of Agreement Regarding the Role of GCs in Acquired Mutation Testing between Genetics Laboratory Directors

If the responding director indicated that they did not currently employ a GC (n=19), they were asked to select reasons why a GC is not directly employed by the lab. Among the reasons listed in the survey, lack of funding for a position and a limited role for a GC in the laboratory were cited as the most common reasons for not employing a GC (47.4% and 36.8%, respectively). Additionally, 42.1% of respondents reported that they collaborate with GCs in other departments in their “other” response option (Table 8).

The level of agreement regarding a role for GCs in acquired mutation testing was compared between respondents who employ a GC and those who do not. The level of agreement was ranked on a 10-point scale (1= strongly disagree GCs have a role in acquired mutation testing, 5=neutral, 10= strongly agree GCs have a role in acquired mutation testing). The median level of agreement for directors who employ a GC was 8, as compared to 5 for directors who do not employ a GC (range 1-10 for both groups). The median level of agreement for directors who currently employ a GC was significantly higher than the median for directors who do not currently employ a GC (p=0.050) (Figure 5).
DISCUSSION

This is the first study that aimed to characterize the role of laboratory GCs in genetic testing for acquired mutations related to oncogenesis. Furthermore, the perspective of genetics laboratory directors on the role of GCs in laboratory had not been described previously. Although responsibilities related to germline testing predominated GCs’ total effort in the laboratory, the study found that most GCs who work in laboratories that offer acquired mutation testing dedicate between 1-25% of their total time on acquired mutation testing.

The results of this study provide evidence that the core skills of GCs already have been adapted and are perceived as efficacious to the facilitation of acquired mutation testing. Among these roles, acting as a resource for ordering providers was the most common role currently performed by GCs related to acquired mutation testing. Furthermore, if the GC was not currently performing this role or if the respondent did not currently employ a GC, acting as a provider resource remained an appropriate role for GCs when considering increasing responsibilities in acquired mutation testing. This finding is in parallel to a study by Christian (2012) which aimed to characterize the current roles for laboratory genetic counselors. In this study, 95% of respondents indicated that they act as a customer liaison in the laboratory. Additional roles described by Christian (2012) that mirrored common roles in acquired mutation testing identified in our study included: calling out test results to the ordering provider, coordinating research studies, administrative duties, and marketing. This finding suggests that while the content and implications of acquired mutation testing and germline mutation testing may differ, the roles of the lab GC are generally the same irrespective of
testing type. Kotzer (2014) specifically described the importance of collecting relevant clinical information related to a test order, which was a common role that was currently performed by GCs (60%) and also was commonly cited as an appropriate role for GCs to expand into related to testing for acquired mutations (50%).

Incidental constitutional abnormalities present a unique challenge within the realm of testing for acquired mutations. Prior to this study, however, the laboratory GCs role in reporting incidental constitutional findings had not been described previously in the literature. Discussing incidental findings with healthcare providers was the second most common (70%) role currently performed by GCs working in laboratories that offer acquired mutation testing and was also reported as an expanded opportunity for laboratory GCs in our study. Of note, 79% of laboratory directors indicated they encounter at least one incidental reportable constitutional abnormality each year during acquired mutation testing. Together, these results indicate that incidental constitutional findings are not uncommon and GCs possess the skillset to discuss these findings and their implications with providers.

Christian (2012) found that 44% of participating laboratory GCs reported being involved in signing laboratory reports. This finding is in contrast to the laboratory director’s perceptions in our study in which 97% of respondents indicated signing lab reports was not a relevant role for GCs, with regard to reports for acquired mutation testing. As GCs are typically not certified to sign off on laboratory reports in the United States regardless of testing type, the response frequency in Christian et al. could represent GCs that are involved in co-signing reports with a laboratory director or reflect a misunderstanding of the survey question. The creation of
laboratory reports and the interpretation of results were also cited as some of the least relevant roles for laboratory GCs related to acquired mutation testing. From anecdotal evidence, laboratory GCs are an important part of the creation of laboratory reports and interpretation of constitutional results; however, the results from our study may indicate that these roles may be less relevant in the field of acquired mutation testing. It is possible that training and education regarding the development and interpretation of acquired mutation reports results these roles would be perceived as more relevant and in the scope of their practice.

This study showed that training during a genetic counseling master’s program in addition to on the job training were the best mechanisms for equipping GCs with the knowledge to assume a role in acquired mutation testing. As such, it will be important for training programs to adapt their curricula to include courses about laboratory genetic counseling as well as offer clinical rotations in the laboratory to enable students to reach their full potential in the genetics laboratory setting. The idea of integrating laboratory-focused training opportunities into the existing master’s curriculum was most recently echoed by Swanson (2013) and exists as part of the required curriculum for genetic counseling training programs seeking accreditation through the American Board of Genetic Counseling. Integrating information about the most salient topics related to acquired mutation testing into master’s level courses on laboratory genetic counseling may minimize the amount of on the job training needed to prepare GCs to assume roles in acquired mutation testing.
In terms of curricula development, it may be important to highlight the subject areas that were identified as the most important for GCs to expand their role in acquired mutation testing (i.e., interpretation of pathology reports, natural history of cancer, psychosocial skills, and understanding of sample types amenable to testing). Many of these topics are currently covered in genetic counseling training programs and are equally important to learn about to fulfill a variety of capacities in the laboratory. Comparing these topics to the most common current and expanded roles reported for GCs in acquired mutations, it is possible to infer why these topics were ranked as the most important.

To adequately clarify a test order and collect relevant clinical data, a working understanding of the pathology, the natural history of cancer, and sample types amendable to testing would be imperative. Furthermore, interacting with external healthcare providers during the genetic testing process (i.e., by acting as a provider resource, discussing incidental findings, and calling out results) may require, in a less traditional sense, psychosocial counseling skills. It is important to note that “psychosocial counseling” was frequently ranked as one of the most important topics as well as one of least important topics for GCs to learn about to expand their role in acquired mutation testing. Laboratory directors who employed a GC ranked psychosocial counseling as one of the most important topics more frequently than directors who did not employ a GC (6 versus 3). This finding could suggest that directors who currently employ a GC place a high value on a GCs psychosocial counseling skills in the laboratory. This finding also could suggest that respondents were unclear on the way the question was worded.
Interestingly, there were differences in how often directors ranked certain topics as most important based on whether they currently employed a GC or not. Laboratory directors who currently employed a GC ranked “clarification of sample type” as an important topic more frequently when compared to laboratory directors who did not employ a GC (5 versus 1). This finding may suggest that laboratory directors who currently employ a GC have recognized the importance of this role among the GCs in their laboratory. Furthermore, laboratory directors who did not employ a GC ranked “natural history cancer” as an important topic more frequently when compared to laboratory directors who currently employed a GC (7 versus 1).

For the least important roles for a GC to expand their role in acquired mutation testing, laboratory directors tended to be in agreement regardless of whether they currently employed a GC.

The statistical power necessary to draw meaningful comparisons between directors who employed a GC (Group A) vs. directors who did not employ a GC (Group B) on the role of laboratory GCs in acquired mutation testing was impeded by our small sample size. However, a statistically significant difference (p=0.050) was noted between the two groups as related to the degree in which they agreed that laboratory GCs have a role in acquired mutation testing. Group A more strongly agreed that GCs have a role in acquired mutation testing, as compared to Group B who were generally more neutral as to the role of GCs in acquired mutation testing. If the laboratory director was unfamiliar with the role of a GC, the director may not have seen the benefit in the integration of a GC in testing for acquired mutations. Furthermore, although not directly employed by the genetic testing laboratory, 42.1% of those in Group B reported collaborating with GCs in other departments. This finding may suggest that a more conventional
perception of clinical-based GCs may exist among directors who do not directly interact with laboratory GCs as compared to those in Group A. It is also reasonable to apply this conclusion to the role of laboratory GCs in general. Laboratory directors who currently employ a GC may likely agree there is a role for GCs in the laboratory setting as they are familiar with their contributions; whereas laboratory directors who do not employ a GC directly may not be as familiar with their responsibilities outside of a clinical setting and perceive their role in the laboratory more neutrally.

Limitations

Due to the low response rate and small sample size in this study, the generalizability of our findings may be limited. Additionally, the list of prospective participants was restricted to laboratories/corresponding directors identified through a manual query of the GTR and/or through a list of training sites. This method of identifying potential participants may have inhibited the authors’ ability to elicit participation from all directors in the U.S., and was not selective for laboratories offering genetic testing for acquired mutations. However, many of our findings related to director’s perceptions of the GC’s role in acquired mutation were mirrored by prior studies describing the overall role of GCs in the laboratory. Furthermore, the self-reported nature of the data may not provide a complete description of the GC’s role in acquired mutation testing.

Future Studies

Future studies will address the perspectives of clinical oncology health professionals on the role of genetic counselors in genetic testing for acquired mutations related to oncogenesis.
Furthermore, it will be important to assess whether GCs (laboratory-based GCs and clinical cancer GCs) would be interested in increasing their role in acquired mutation testing. Future studies also are needed to validate our findings on the roles of GCs related to acquired mutation testing.

Conclusions

This study provides additional support for the applicability of traditional clinic GC roles to non-traditional work settings and demonstrates that a GC’s impact is not limited to facilitating genetic testing for constitutional mutations. Additionally, our results support the need for genetic counseling training programs to incorporate didactic coursework about laboratory-based genetic counseling, which may include topics regarding the genetic counselor’s role in testing for acquired mutations related to oncogenesis. As the field of personalized oncology expands in proportion to the clinical application of complex genetic tests, the role of the laboratory genetic counselor will become increasingly important to bridge the gap between the ordering provider and the testing laboratory.
REFERENCES


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### Table 1. Participant Demographics (n=39)

<table>
<thead>
<tr>
<th>Professional Title</th>
<th>n(%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Laboratory Director</td>
<td>33 (84.6)</td>
</tr>
<tr>
<td>Other Director Type (Associate, Assistant, Other)</td>
<td>6 (15.4)</td>
</tr>
<tr>
<td><strong>Primary Decision Maker for Staffing/Hiring</strong></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>17 (43.6)</td>
</tr>
<tr>
<td>No</td>
<td>22 (56.4)</td>
</tr>
<tr>
<td><strong>Specialty</strong>*</td>
<td></td>
</tr>
<tr>
<td>Cytogenetics</td>
<td>17 (43.6)</td>
</tr>
<tr>
<td>Molecular Genetics</td>
<td>28 (71.8)</td>
</tr>
<tr>
<td>Other</td>
<td>1 (2.6)</td>
</tr>
<tr>
<td><strong>Length of Time Board-Certified (years)</strong></td>
<td></td>
</tr>
<tr>
<td>≤5</td>
<td>4 (10.3)</td>
</tr>
<tr>
<td>6-10</td>
<td>16 (41.0)</td>
</tr>
<tr>
<td>11-15</td>
<td>5 (12.8)</td>
</tr>
<tr>
<td>&gt;15</td>
<td>11 (28.2)</td>
</tr>
<tr>
<td><strong>Gender</strong></td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>14 (35.9)</td>
</tr>
<tr>
<td>Female</td>
<td>24 (61.5)</td>
</tr>
<tr>
<td><strong>Race</strong></td>
<td></td>
</tr>
<tr>
<td>White</td>
<td>24 (61.5)</td>
</tr>
<tr>
<td>Other</td>
<td>13 (33.3)</td>
</tr>
</tbody>
</table>

*11 respondents were dual-certified in Cytogenetics and Molecular genetics*
### Table 2. Laboratory Characteristics

<table>
<thead>
<tr>
<th></th>
<th>GC (n=20)</th>
<th>No GC (n=19)</th>
<th>Total (n=39)</th>
</tr>
</thead>
<tbody>
<tr>
<td>% of total services for acquired mutation testing</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>&lt;1%</td>
<td>3 (15.0)</td>
<td>2 (10.5)</td>
<td>5 (12.8)</td>
</tr>
<tr>
<td>1-25%</td>
<td>10 (50.0)</td>
<td>4 (21.1)</td>
<td>14 (35.9)</td>
</tr>
<tr>
<td>26-50%</td>
<td>1 (5.0)</td>
<td>6 (31.6)</td>
<td>7 (18.0)</td>
</tr>
<tr>
<td>&gt;51%</td>
<td>6 (30.0)</td>
<td>6 (31.6)</td>
<td>12 (30.8)</td>
</tr>
<tr>
<td>I don’t know</td>
<td>0 (0)</td>
<td>1 (5.3)</td>
<td>1 (2.6)</td>
</tr>
<tr>
<td># Orders Received for Acquired mutation Testing Per Month</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>&lt;50</td>
<td>9 (45)</td>
<td>8 (42.1)</td>
<td>17 (43.6)</td>
</tr>
<tr>
<td>50-100</td>
<td>3 (15)</td>
<td>4 (21.1)</td>
<td>7 (18.4)</td>
</tr>
<tr>
<td>101-200</td>
<td>2 (10.0)</td>
<td>3 (15.8)</td>
<td>5 (12.8)</td>
</tr>
<tr>
<td>201-500</td>
<td>5 (25.0)</td>
<td>1 (5.3)</td>
<td>6 (15.4)</td>
</tr>
<tr>
<td>&gt;500</td>
<td>1 (5.0)</td>
<td>2 (10.5)</td>
<td>3 (7.7)</td>
</tr>
<tr>
<td>Laboratory Certifications</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>CLIA</td>
<td>20 (100)</td>
<td>19 (100)</td>
<td>39 (100)</td>
</tr>
<tr>
<td>CAP</td>
<td>19 (95.0)</td>
<td>14 (73.7)</td>
<td>33 (84.6)</td>
</tr>
<tr>
<td>ABMG Training Site</td>
<td>19 (95.0)</td>
<td>7 (36.8)</td>
<td>26 (66.7)</td>
</tr>
<tr>
<td>Patient Population for Acquired Mutation Testing</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Pediatric Only</td>
<td>2 (10.0)</td>
<td>1 (5.3)</td>
<td>3 (7.7)</td>
</tr>
<tr>
<td>Adult Only</td>
<td>1 (5.0)</td>
<td>1 (5.3)</td>
<td>2 (5.1)</td>
</tr>
<tr>
<td>Adult &amp; Pediatric</td>
<td>17 (85.0)</td>
<td>17 (89.5)</td>
<td>34 (87.2)</td>
</tr>
<tr>
<td>Median (range)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Number of Individuals</td>
<td>35 (6-100)</td>
<td>11.5 (5-60)</td>
<td>20 (5-100)</td>
</tr>
<tr>
<td>Employed by Laboratory</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Table 3. Proportion of Time GCs Dedicate to Acquired Mutation Testing as Perceived by the Laboratory Director (n=20)

<table>
<thead>
<tr>
<th>% Total Time</th>
<th>n (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>&lt;1%</td>
<td>5 (25.0)</td>
</tr>
<tr>
<td>1-25%</td>
<td>13 (65.0)</td>
</tr>
<tr>
<td>≥ 26%</td>
<td>1 (5)</td>
</tr>
</tbody>
</table>
Table 4. Estimated Number of Incidental Reportable Constitutional Abnormalities During Acquired Mutation Testing Per Year (n=39)

<table>
<thead>
<tr>
<th>Response Option</th>
<th>Frequency n (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>0</td>
<td>8 (20.5)</td>
</tr>
<tr>
<td>1-5</td>
<td>22 (56.4)</td>
</tr>
<tr>
<td>6-10</td>
<td>3 (7.7)</td>
</tr>
<tr>
<td>11-15</td>
<td>1 (2.6)</td>
</tr>
<tr>
<td>16-20</td>
<td>1 (2.6)</td>
</tr>
<tr>
<td>≥21</td>
<td>3 (7.7)</td>
</tr>
</tbody>
</table>
Table 5. Proportion of Time (% effort) Lab GCs Spend on the Common Roles Currently Performed Related to Acquired Mutation Testing (n=20)

<table>
<thead>
<tr>
<th>Role</th>
<th>Percent Effort Reported, n (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Act as Ordering Provider Resource</td>
<td>11 (55) 5 (25) 0 (0)</td>
</tr>
<tr>
<td>Discuss Incidental Findings</td>
<td>12 (60) 2 (10) 1 (5)</td>
</tr>
<tr>
<td>Clarify Test Order</td>
<td>11 (55) 5 (25) 0 (0)</td>
</tr>
<tr>
<td>Call Out Results to Provider</td>
<td>14 (70) 1 (5) 1 (5)</td>
</tr>
<tr>
<td>Collect Clinical Data Related to Case</td>
<td>11 (55) 3 (15) 2 (10)</td>
</tr>
<tr>
<td>Develop Patient Materials</td>
<td>12 (60) 3 (15) 1 (5)</td>
</tr>
<tr>
<td>Coordinate Research Studies</td>
<td>13 (65) 3 (15) 0 (0)</td>
</tr>
<tr>
<td>Administrative Duties</td>
<td>14 (70) 1 (5) 0 (0)</td>
</tr>
<tr>
<td>Marketing</td>
<td>11 (55) 2 (10) 1 (5)</td>
</tr>
</tbody>
</table>
Table 6. Most and Least Important Topics for GCs to Learn About to Expand Their Role In Acquired Mutation Testing (n=38)

<table>
<thead>
<tr>
<th>Topic</th>
<th>GC</th>
<th>No GC</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Most Important (Rank 1)</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Interpretation of pathology reports related to acquired mutation testing</td>
<td>4</td>
<td>5</td>
<td>9</td>
</tr>
<tr>
<td>Psychosocial skills</td>
<td>6</td>
<td>3</td>
<td>9</td>
</tr>
<tr>
<td>Natural history of cancer</td>
<td>1</td>
<td>7</td>
<td>8</td>
</tr>
<tr>
<td>Clarification of Sample Type</td>
<td>5</td>
<td>1</td>
<td>6</td>
</tr>
<tr>
<td><strong>Least Important (Rank 10)</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Patents for acquired mutation testing</td>
<td>5</td>
<td>5</td>
<td>10</td>
</tr>
<tr>
<td>Flow cytometry</td>
<td>3</td>
<td>3</td>
<td>6</td>
</tr>
<tr>
<td>Psychosocial skills</td>
<td>3</td>
<td>3</td>
<td>6</td>
</tr>
<tr>
<td>Bench protocols for carrying out acquired mutation analysis</td>
<td>2</td>
<td>3</td>
<td>5</td>
</tr>
</tbody>
</table>

*One respondent did not provide data for this question.*
Table 7. Additional Themes Related to Training for GCs to Enhance their Involvement in Acquired Mutation Testing (n=10)

<table>
<thead>
<tr>
<th>Training Theme</th>
<th>Frequency Reported</th>
</tr>
</thead>
<tbody>
<tr>
<td>More education in genetics/genomics</td>
<td>4</td>
</tr>
<tr>
<td>Teaching/Interactions with other HCPs</td>
<td>4</td>
</tr>
<tr>
<td>Attend Conferences</td>
<td>3</td>
</tr>
<tr>
<td>On the job training + additional master’s coursework</td>
<td>2</td>
</tr>
</tbody>
</table>

*Three respondents indicated more than one theme; thus the total frequency reported is n>10*
### Table 8. Most Frequent Reasons Why a GC is Not Currently Employed by the Laboratory (n=19)

<table>
<thead>
<tr>
<th>Reason</th>
<th>Frequency n(%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lack of funding for a GC position</td>
<td>9 (47.4)</td>
</tr>
<tr>
<td>Collaborate with GCs in other departments</td>
<td>8 (42.1)</td>
</tr>
<tr>
<td>Limited role for a GC</td>
<td>7 (36.8)</td>
</tr>
</tbody>
</table>

*Respondents could select all applicable response options*
Figure 1. Common Roles Currently Performed by Laboratory GCs Related to Acquired Mutation Testing (n=20)

Figure 1 demonstrates roles that ≥50% of respondents indicated were currently performed by GCs in their laboratory related to acquired mutation testing.
Figure 2. Common Expanded Roles for Laboratory GCs Related to Acquired Mutation Testing (n=39)

Figure 2 demonstrates roles that ≥45% of respondents indicated were not currently performed, but could be appropriate for laboratory GCs to perform related to acquired mutation testing. The grey bars indicate the number of responses for laboratory directors who employ a GC and the black bars indicate the number of responses for laboratory directors who do not employ a GC.
Figure 3. Common Roles Reported as Not Relevant for GCs to Perform Related to Acquired Mutation Testing (n=39)

Figure 3 demonstrates roles that ≥50% of respondents indicated were not relevant for laboratory GCs to perform related to acquired mutation testing.
Figure 4 demonstrates the best modes of training to prepare GCs for a role in acquired mutation testing as perceived by laboratory directors. Other training modes included were described as a combination of on the job training and additional master’s coursework.
Figure 5. Level of Agreement Regarding GCs Role in Acquired Mutation Testing between Directors Who Employ a GC vs. Directors Who Do Not

<table>
<thead>
<tr>
<th>Employ GC</th>
<th>N</th>
<th>Median Level of Agreement</th>
<th>Range</th>
<th>P-value</th>
</tr>
</thead>
<tbody>
<tr>
<td>No</td>
<td>19</td>
<td>5</td>
<td>1-10</td>
<td>0.050*</td>
</tr>
<tr>
<td>Yes</td>
<td>20</td>
<td>8</td>
<td>1-10</td>
<td></td>
</tr>
</tbody>
</table>

Figure 5 compares the level of agreement on the role of GCs in acquired mutation testing between laboratory directors who employ a GC as compared to laboratory directors who do not employ a GC. Non-parametric analysis (Kruskall-Wallis Test, 1-way analysis) was performed to assess statistically significant differences between the two groups.
APPENDIX B. Survey for Laboratory Directors who work with a Laboratory GC

Eligibility Assessment

1. Professional Title
   - Laboratory Director
   - Associate Laboratory Director
   - Assistant Laboratory Director
   - Other type of Laboratory Director, specify: _____________________
   - I am not a Laboratory Director (Exclusion criteria)

2. Does your laboratory perform genetic testing for acquired mutations?
   - Yes
   - No (Exclusion criteria)
   - I don’t know (Exclusion criteria)

Section I. Role of Genetic Counselors (GCs) in your Laboratory

3. Does your laboratory employ a genetic counselor (GC)?
   - Yes. If yes, how many GCs work in your laboratory? _____
   - No (Respondents would continue to APPENDIX C)

4. What percent of the GCs’ time in your laboratory is dedicated to acquired mutations?
   - <1%
   - 1-25%
   - 26-50%
   - 51-75%
   - 76-100%
   - I don’t know

5. What percent of the GC’s time in your laboratory is dedicated to constitutional (germline) mutations?
   - <1%
   - 1-25%
   - 26-50%
   - 51-75%
   - 76-100%
   - I don’t know

6. Please indicate your level of agreement with the following statement: I think lab GCs have a role in genetic testing for acquired mutations.
   - 1…
   - 2…
   - 3…
   - 4…
   - 5…
   - 6…
   - 7…
   - 8…
   - 9…
   - 10
   (Strongly disagree) (Strongly agree)
7. Do GCs in your lab currently perform the following roles related to acquired mutation testing?

<table>
<thead>
<tr>
<th>Role</th>
<th>Yes</th>
<th>No, I feel this is not a relevant role for GCs in acquired mutation testing.</th>
<th>No. However, I feel this could be a role for lab GCs in acquired mutation testing.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Collection of clinical data related to the case</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Clarification of sample type</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Clarification of a test order</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Acting as a resource for healthcare providers during test ordering</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Interpretation of test results</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Creation of laboratory reports</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Discussing abnormal constitutional findings with healthcare providers</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Administrative duties</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Calling out test results to ordering provider</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Signing laboratory reports</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Coordination of research studies</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Marketing</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Management of projects</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Development of patient materials</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Other, please specify:</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
8. Of the time your lab GCs spend on **acquired** mutation testing, what percent effort most accurately reflects the time they spend on each of the following roles?

<table>
<thead>
<tr>
<th>Role</th>
<th>1%-25%</th>
<th>26%-50%</th>
<th>51%-75%</th>
<th>76%-100%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Collection of clinical data related to the case</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Clarification of sample type</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Clarification of a test order</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Acting as a resource for healthcare providers during test ordering</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Interpretation of test results</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Creation of laboratory reports</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Discussing abnormal constitutional findings with healthcare providers</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Administrative duties</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Calling out test results to ordering provider</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Signing laboratory reports</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Coordination of research studies</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Marketing</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Management of projects</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Development of patient materials</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Other, please specify:</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**Section II. Knowledge/training for Genetic Counselors**

9. What skills/knowledge do GCs bring to your laboratory? (*Check all that apply*)

- [ ] Ability to **interpret** complex genetic information/results
- [ ] Ability to **explain** complex genetic information/results
- [ ] **Detailed** training in acquired mutations and oncogenesis
- [ ] **General** training in acquired mutations and oncogenesis
- [ ] Ability to assimilate evolving technology/testing
- [ ] Training in hereditary cancer syndromes
- [ ] Clinical perspective on oncology patients
- [ ] Clinical perspective on acquired mutations
- [ ] Other, please specify:_________________________________________
10. In your opinion, how important is it for GCs to learn about the following topics to expand their role in genetic testing for **acquired** mutations? Please rank the following items 1-10 (1 being **most** important and 10 being **least** important).

- Understanding of lab methodologies for detecting acquired mutations
- Bench protocols for carrying out acquired mutation analysis
- Understanding of sample types that are amenable to testing
- Interpretation of pathology reports as related to acquired tests
- Flow cytometry
- Natural history of cancer
- Treatment of cancer
- Test development for acquired mutations
- Patents for acquired testing
- Psychosocial skills

11. What is the best way for GCs to learn more about the topics above?

- Training during genetic counseling master’s degree
- On the job training
- Certificate training
- Advanced research degree (i.e., PhD)
- Other, please specify: _____________________________

12. In the space below, please indicate other roles, training content, etc... that you think could enhance GCs involvement in genetic testing for acquired mutations:

______________________________________________________________________________
______________________________________________________________________________
______________________________________________________________________________

**Section III. Laboratory Testing Volume and Demographics**

13. In your laboratory, genetic testing for **acquired** mutations accounts for what percentage of services?

- <1%
- 1-25%
- 26-50%
- 51-75%
- 76-100%
- I don’t know
14. **Per month**, how many orders does your laboratory receive for **acquired** mutation testing?

- □ <50
- □ 50-100
- □ 101-200
- □ 201-300
- □ 301-400
- □ 401-500
- □ >500

15. **Per year**, how many times does your laboratory find an incidental reportable **constitutional** abnormality when testing for acquired mutations?

- □ 0 (*Skip question 16*)
- □ 1-5
- □ 6-10
- □ 11-15
- □ 16-20
- □ ≥21

16. What procedure(s) does your laboratory follow after detecting an incidental reportable **constitutional** abnormality during testing for acquired mutations? (*Check all that apply*)

- □ Not report the incidental constitutional abnormality
- □ Report it as an abnormal constitutional result
- □ Request additional sample for confirmation of constitutional result
- □ Have Lab Director call out result to ordering physician
- □ Have Laboratory GC call out result to ordering physician
- □ Have a different laboratory staff member to call out the result, please specify: ______
- □ Other, specify: __________________________________________

17. Please provide a brief explanation of why you would not report an incidental constitutional abnormality: ________________________________________________

18. What are the certifications currently held by your laboratory? (*Check all that apply*)

- □ Clinical Laboratory Improvement Amendments (CLIA)
- □ College of American Pathologists (CAP)
- □ Fellowship training site for the American Board of Medical Genetics (ABMG)
- □ Other(s), please specify: ______________________________________

19. How many individuals are employed by your laboratory? ____________

20. From which of the following patient populations does your laboratory receive requests for **acquired** mutation testing?

- □ Pediatric
- □ Adult
- □ Both
Section IV. Participant Demographics

21. Are you the primary decision maker for staffing and hiring in your laboratory?
   ☐ Yes
   ☐ No

22. What is your specialty (Check all that apply)
   ☐ Cytogenetics
   ☐ Molecular genetics
   ☐ Biochemical genetics
   ☐ Other, specify: ____________________

23. How long have you been board-certified in your specialty (e.g., FACMG)?
   ☐ <5 years
   ☐ 6-10 years
   ☐ 11-15 years
   ☐ >15 years
   ☐ Not applicable
      24. What is your age (in years)? ______

25. What is your gender?
   ☐ Male
   ☐ Female

26. What is your race? (Check all that apply)
   ☐ African American
   ☐ American Indian or Alaska Native
   ☐ Asian
   ☐ Black or African American
   ☐ Native Hawaiian or other Pacific Islander
   ☐ White

27. What is your ethnicity?
   ☐ Non-Hispanic
   ☐ Hispanic
APPENDIX C. Survey for Laboratory Directors who do not work with a Laboratory GC

Eligibility Assessment

1. Professional Title
   - □ Laboratory Director
   - □ Associate Laboratory Director
   - □ Assistant Laboratory Director
   - □ Other type of Laboratory Director, specify: _____________________
   - □ I am not a Laboratory Director (Exclusion criteria)

2. Does your laboratory perform genetic testing for acquired mutations?
   - □ Yes
   - □ No (Exclusion criteria)
   - □ I don’t know (Exclusion criteria)

Section I. Genetic Counselors (GCs) in your Laboratory

3. Why does your laboratory not employ a GC? (Check all that apply)
   - □ Limited role for a GC in the laboratory
   - □ Lack of funding for a position
   - □ Type of testing offered does not require GC services
   - □ Unsure how to use GC’s skills in the laboratory
   - □ Other, specify: _____________________

4. Please indicate your level of agreement with the following statement: I think lab GCs have a role in genetic testing for acquired mutations.
   1…………….2…………..3…………..4…………..5…………..6…………..7…………..8…………..9…………..10
   (Strongly disagree)  (Strongly agree)
5. Which of the following roles do you think laboratory GCs could perform related to genetic testing for acquired mutations: *(Check all that apply)*

<table>
<thead>
<tr>
<th>Role</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Collection of clinical data related to the case</td>
<td></td>
</tr>
<tr>
<td>Clarification of sample type</td>
<td></td>
</tr>
<tr>
<td>Clarification of a test order</td>
<td></td>
</tr>
<tr>
<td>Acting as a resource for healthcare providers during test ordering</td>
<td></td>
</tr>
<tr>
<td>Interpretation of test results</td>
<td></td>
</tr>
<tr>
<td>Creation of laboratory reports</td>
<td></td>
</tr>
<tr>
<td>Discussing abnormal constitutional findings with healthcare providers</td>
<td></td>
</tr>
<tr>
<td>Administrative duties</td>
<td></td>
</tr>
<tr>
<td>Calling out test results to ordering provider</td>
<td></td>
</tr>
<tr>
<td>Signing laboratory reports</td>
<td></td>
</tr>
<tr>
<td>Coordination of research studies</td>
<td></td>
</tr>
<tr>
<td>Marketing</td>
<td></td>
</tr>
<tr>
<td>Management of projects</td>
<td></td>
</tr>
<tr>
<td>Development of patient materials</td>
<td></td>
</tr>
<tr>
<td>Other, please specify:</td>
<td></td>
</tr>
</tbody>
</table>

**Section II. Knowledge/training for Genetic Counselors**

6. What skills/knowledge do you feel GCs could bring to your laboratory? *(Check all that apply)*

- [ ] Ability to *interpret* complex genetic information/results
- [ ] Ability to *explain* complex genetic information/results
- [ ] *Detailed* training in acquired mutations and oncogenesis
- [ ] *General* training in acquired mutations and oncogenesis
- [ ] Ability to assimilate evolving technology/testing
- [ ] Training in hereditary cancer syndromes
- [ ] Clinical perspective on oncology patients
- [ ] Clinical perspective on acquired mutations
- [ ] Other, please specify: ___________________________________________
7. In your opinion, how important is it for GCs to learn about the following topics to expand their role in genetic testing for **acquired** mutations? Please rank the following items 1-10 (1 being **most** important and 10 being **least** important).

_____Understanding of lab methodologies for detecting acquired mutations
_____Bench protocols for carrying out acquired mutation analysis
_____Understanding of sample types that are amenable to testing
_____Interpretation of pathology reports as related to acquired tests
_____Flow cytometry
_____Natural history of cancer
_____Treatment of cancer
_____Test development for acquired mutations
_____Patents for acquired testing
_____Psychosocial skills

8. What is the best way for GCs to learn more about the topics above?

☐ Training during genetic counseling master’s degree
☐ On the job training
☐ Certificate training
☐ Advanced research degree (i.e., PhD)
☐ Other, please specify: __________________________

9. In the space below, please indicate other roles, training content, etc... that you think could enhance GCs involvement in genetic testing for acquired mutations:

__________________________________________________________

______________________________________________________________________________
______________________________________________________________________________

**Section III. Laboratory Testing Volume and Demographics**

10. In your laboratory, genetic testing for **acquired** mutations accounts for what percentage of services?

☐ <1%
☐ 1-25%
☐ 26-50%
☐ 51-75%
☐ 76-100%
☐ I don’t know
11. **Per month**, how many orders does your laboratory receive for **acquired** mutation testing?
   - ☐ <50
   - ☐ 50-100
   - ☐ 101-200
   - ☐ 201-300
   - ☐ 301-400
   - ☐ 401-500
   - ☐ >500

12. **Per year**, how many times does your laboratory find an incidental reportable **constitutional** abnormality when testing for acquired mutations?
   - ☐ 0 (Skip question 16)
   - ☐ 1-5
   - ☐ 6-10
   - ☐ 11-15
   - ☐ 16-20
   - ☐ ≥21

13. What procedure(s) does your laboratory follow after detecting an incidental reportable **constitutional** abnormality during testing for **acquired** mutations? *(Check all that apply)*
   - ☐ Not report the incidental constitutional abnormality
   - ☐ Report it as an abnormal constitutional result
   - ☐ Request additional sample for confirmation of constitutional result
   - ☐ Have Lab Director call out result to ordering physician
   - ☐ Have Laboratory GC call out result to ordering physician
   - ☐ Have a different laboratory staff member to call out the result, please specify: _______
   - ☐ Other, specify: ________________________________

14. Please provide a brief explanation of why you would not report an incidental constitutional abnormality: ________________________________

15. What are the certifications currently held by your laboratory? *(Check all that apply)*
   - ☐ Clinical Laboratory Improvement Amendments (CLIA)
   - ☐ College of American Pathologists (CAP)
   - ☐ Fellowship training site for the American Board of Medical Genetics (ABMG)
   - ☐ Other(s), please specify: ________________________________

16. How many individuals are employed by your laboratory? _____________
17. From which of the following patient populations does your laboratory receive requests for acquired mutation testing?

- Pediatric
- Adult
- Both

Section IV. Participant Demographics

18. Are you the primary decision maker for staffing and hiring in your laboratory?
- Yes
- No

19. What is your specialty (Check all that apply)
- Cytogenetics
- Molecular genetics
- Biochemical genetics
- Other, specify: ________________

20. How long have you been board-certified in your specialty (e.g., FACMG)?
- <5 years
- 6-10 years
- 11-15 years
- >15 years
- Not applicable

21. What is your age (in years)? _____

22. What is your gender?
- Male
- Female

23. What is your race? (Check all that apply)
- African American
- American Indian or Alaska Native
- Asian
- Black or African American
- Native Hawaiian or other Pacific Islander
- White

24. What is your ethnicity?
- Non-Hispanic
- Hispanic