

# Further Defining the Role of the Laboratory Genetic Counselor

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**Abstract** Laboratory genetic counseling is becoming increasingly common as a result of increased laboratory services and genetic testing menus, as well as growing job responsibilities. Christian et al. (2012) provided the first quantitative data regarding the roles of the laboratory-based genetic counselor (LBGC) finding that two of the most prevalent roles are as customer liaisons and communicators of test results. The goal of the present study was to further delineate the role of the LBGC by addressing specific tasks that LBGCs are involved with on a day-to-day basis. A survey was designed to expand upon themes identified in the Christian et al. (2012) study by querying specific tasks performed in several categories of potential LBGC job duties. An invitation for LBGCs to participate was distributed via email to the membership of the National Society of Genetic Counselors (NSGC) and the Canadian Association of Genetic Counsellors (CAGC). We identified 121 genetic counselors who primarily work in the

laboratory setting or whose job role includes a laboratory component. Almost all respondents performed customer liaison/case coordination (95 %), and interpretation and result reporting (88 %). The most frequently performed tasks within these categories involved addressing questions from clients, making phone calls with genetic testing results, obtaining clinical or family history information for results interpretation, and composing case-specific interpretations for unique results and/or obtaining literature references to support interpretations. The study results also point to trends of expanding roles in sales and marketing, variant interpretation and management responsibilities. Results of this study may be useful to further define the full scope of practice of LBGCs, aid in the development of new LBGC positions and expand current positions to include roles related to test development, research, and student supervision. It may also aid in curriculum updates for training programs to increase exposure to LBGC roles.

**Keywords** Laboratory · Genetic counselor · Non-clinical · Roles

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## Introduction

According to data from the National Society of Genetic Counselors (NSGC) Professional Status Survey (PSS), laboratory genetic counseling is becoming increasingly common. The percentage of genetic counselors citing diagnostic laboratories as their primary work setting increased from 6 % in the 2002 PSS to 16.8 % in the 2014 PSS (National Society of Genetic Counselors 2002, 2014). This data may underrepresent genetic counselors whose roles included both

clinical and non-clinical components, as respondents to the 2014 NSGC PSS were only able to select one primary role (clinical or non-clinical).

The expanding number of laboratory genetic counselors is a result of increased laboratory services and genetic testing menus, as well as growing job responsibilities (Zetzsche et al. 2013). The role of the laboratory-based genetic counselor (LBGC) was first described by Amos and Gold in 1998 as “clinical genetics educators who provide physicians with genetics expertise that they would not otherwise be able to access” (Amos and Gold 1998, p.294). This is accomplished by answering specific questions about tests offered by the laboratory, gathering clinical information essential for test interpretation, providing referrals to local clinical genetic counseling services, performing risk recalculation based on additional information obtained, and explaining the impact and limitations of genetic test results. Through a series of case studies, Scacheri et al. explored the role of LBGCs in improving patient care in the context of molecular diagnostic laboratories (Scacheri et al. 2008). The authors commented on the importance of the LBGC in assessing orders for appropriateness of testing, test prioritization, and providing recommendations for follow-up testing. Zetzsche et al. explored the history of laboratory genetic counseling roles through semi-structured interviews of LBGCs with up to 24 years of experience (Zetzsche et al. 2013). The interviews highlighted the versatility of genetic counselors and the evolution of laboratory-based roles alongside the growth, commercialization, and specialization of genetic testing laboratories.

Christian et al. (2012) provided the first quantitative data regarding the role of LBGCs by administering a survey to laboratory genetic counselors in North America. They assessed the proportion of time spent on various roles as well as areas of job satisfaction. Their data corroborated the earlier findings of Scacheri et al. (2008) by demonstrating that two of the most prevalent roles for LBGCs are as customer liaisons and communicators of test results. Respondents in this study reported high levels of satisfaction with the support they received from laboratory directors and their interactions with genetic counselors and other clinicians. While the Christian et al. (2012) study focused on broad categories of roles performed by LBGCs, it did not address specific tasks that LBGCs are involved with on a day-to-day basis. The goal of the present study was to further delineate the role of the LBGC. This study was designed to capture the broad range and versatility of genetic counseling services in the laboratory. An additional aim was to elucidate any differences in the types of roles performed by LBGCs in various types of clinical laboratories.

## Methods

### Participants and Instrumentation

The study used a survey designed by the authors, who represented four institutions, two countries, and a variety of different laboratory settings. The survey was designed using Survey Monkey (SurveyMonkey.com, LLC, Palo Alto, CA, USA) and included a combination of multiple choice, matrix/rating scale, and open-ended questions. The survey was composed of 38 questions which addressed demographics and work environment, specific tasks performed by laboratory genetic counselors, percent of time involved in these tasks, and overall job satisfaction. Questions regarding the tasks performed by LBGCs were organized into 10 categories: 1) Customer Liaison, 2) Interpretation and Result Reporting, 3) Test Development and Test Performance Issues, 4) Research and Publications, 5) Website and Database Support, 6) Sales and Marketing, 7) Insurance and Billing, 8) Education and Supervision of Students, 9) Management Duties, and 10) Clinical Counseling. The categories were developed based on the Christian et al. (2012) study and roles currently performed by the authors. The survey was piloted on a small group of LBGCs and based on their feedback revisions to word choice and organization of survey questions were made. The survey was estimated to take 30 min to complete. Approval was obtained for exempt status through the Institutional Review Board at the Mayo Clinic.

The target population was genetic counselors working in a laboratory setting or whose role included a laboratory component and this criteria was stated in the invitation as the requirement for participation. An invitation specifically addressed to LBGCs was circulated by email to general memberships of the National Society of Genetic Counselors (NSGC) ( $n=2806$ ), the Canadian Association of Genetic Counsellors (CAGC) ( $n=320$ ), and the NSGC Industry Special Interest Group ( $n=167$ ). As some genetic counselors belong to more than one of these organizations, duplicate invitations would have been received. This research participation invitation was sent to these groups June 3, 2013. A reminder email was sent 2 weeks after the initial invitation. A total of 130 surveys were returned. Of these, 8 respondents only completed the demographic questions and were thus excluded from analysis. One respondent only completed a third of the survey before discontinuing and was also excluded from analysis. The remaining 121 respondents completed most or all survey items and composed the final sample. Based on the 2014 PSS, it is estimated that there are 471 genetic counselors who are members of NSGC and whose primary role is in a diagnostic laboratory (16.8 % of 2806 members), therefore, the estimated

response rate from eligible participants for this study was 26 % (121/471). This response rate may be an underrepresentation of participation as 16.8 % is the percentage of genetic counselors from the 2014 PSS who reported their *primary* role as a diagnostic laboratory, whereas, we invited participation of all genetic counselors whose role included a laboratory component.

### Data Analysis

Data analysis was performed using JMP software (JMP® 10.0.0 ©2012 SAS Institute Inc., Cary, NC, USA, <http://www.jmp.com/>). Mean, percentage and total number of respondents were used to summarize the responses. Matrix of choice questions had five response options: very often, often, sometimes, rarely, and never. To simplify the descriptive narrative, when analyzing the *frequency* with which a task was performed, the very often and often responses have been combined and are referred to as ‘frequently’ in the remainder of the paper. When analyzing the number of respondents that performed a task within a given category, regardless of frequency, responses were categorized into two groups: the responses of very often, often, sometimes, and rarely were combined to show the total sum of respondents who perform the task at all as opposed to the respondents who do not perform the task (those who responded never). Responses to the open-ended questions were reviewed for common themes by two authors independently. Themes and words used in more than one response were identified as potential domains.

## Results

### Demographics and Work Setting

Demographic and work setting data are shown in Table 1. The majority of respondents reported working for a commercial laboratory (57 %), followed by university or academic laboratory (20.7 %) and hospital-based laboratory (11.6 %). Of the respondents, The majority of respondents reporting working in a molecular genetics laboratory (81.8 %), followed by cytogenetics laboratories (38 %) and biochemical laboratories (14.9 %); 45 (37.2 %) reported working in multiple laboratories, e.g., molecular genetics and cytogenetics laboratories. The mean genetic counseling experience was 7.7 years and the mean time employed as a LBGC was 4.3 years. Almost one fifth of respondents (18.2 %) indicated that they were the first genetic counselor to hold a laboratory based role in their laboratory.

### Job Activities

#### *Percentage of Time Devoted to Each Category*

Overall, customer liaison/case coordination and interpretation/result reporting accounted for more than 50 % of respondents’ time (Table 2). The remaining eight categories each accounted for less than 10 % of respondents’ time, on average. Table 3 depicts the percentage of respondents by type of laboratory who perform each of the role categories at some point, regardless of the frequency (the respondents who indicated they performed the role either very often, often, sometimes, or rarely). This figure shows that respondents in the majority of subspecialties perform most role categories to some extent.

#### *Customer Liaison and Case Coordination*

Almost all respondents, regardless of type of laboratory, reported performing duties within the customer liaison and case coordination category, and the average percentage of time devoted to this category was 30.2 % (Tables 2 and 3). The most frequently performed task within the category was related to addressing questions from clients. Respondents frequently addressed questions regarding test algorithms/strategies (78.5 %), interpretation of test results (76.9 %), and logistics of testing, such as turn-around-time or specimen requirements (72.7 %) (Table S-I). Only 21.5 % of LBGCs frequently answered questions directly from patients. Other frequently performed tasks included determining the appropriateness of testing (66.1 %) and managing high priority cases and specimens (62.8 %).

Facilitating sending specimens to other laboratories was performed infrequently, with 58.7 % rarely or never performing this task. Respondents who facilitated send-outs represented all types of laboratories, but this task was performed most frequently by respondents working in a hospital send-out laboratory (80 %), and infrequently by respondents working at free fetal DNA, maternal serum screening, and newborn screening laboratories (20 %, 9.1 %, 0 %) (Table 4). Among respondents who reported facilitating specimen send outs, 19.1 % frequently established utilization testing criteria and 17 % selected the referral laboratory. While some LBGCs frequently resolved specimen handling issues, just as many reported infrequently performing this task. By type of laboratory, this task was frequently performed by more LBGCs in hospital send-out (60 %) and cytogenetics (50 %) laboratories than in other types of laboratories (Table 4).

#### *Interpreting and Reporting Results*

The average percentage of time devoted to tasks related to interpretation and result reporting was 26.1 %, with 80–100 % of respondents across laboratory types performing at

**Table 1** Demographics and work setting

Variable	<i>n</i>	%	Mean	Median (range)
Gender				
Female	115	95.0		
Male	6	5.0		
Age				
18–24	2	1.7		
25–34	68	56.1		
35–44	44	36.3		
45–54	6	5.0		
55–64	1	0.8		
Race				
White	107	88.4		
Asian	9	7.4		
Black/African American	2	1.7		
Other/no response	3	2.5		
Country of practice				
US	106	87.6		
Canada	14	11.6		
Outside US and Canada	1	0.8		
Region				
1: CT, MA, ME, NH, RI, VT, CN Maritime Provinces	19	15.7		
2: DC, DE, MD, NJ, NY, PA, VA, WV, PR, Quebec	23	19.0		
3: AL, FL, GA, KY, LA, MS, NC, SC, TN	7	5.8		
4: AR, IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, OK, SD, WI, Ontario	33	27.3		
5: AZ, CO, MT, NM, TX, UT, WY, Alberta, Manitoba, Sask.	20	16.5		
6: – AK, CA, HI, ID, NV, OR, WA	17	14.0		
Other	2	1.7		
Years GC experience			7.7	6 (.17–26)
Years as laboratory-based GC			4.3	3 (.17–26)
Years working in current position			3.6	3 (.1–26)
Primary role				
Laboratory genetic counselor	83	68.6		
Sales/marketing/medical specialist representative	17	14.0		
Management	7	5.8		
Clinical genetic counselor	4	3.3		
Research/study coordinator	3	2.5		
Teaching/supervising students	0	0		
Other	7	5.8		
Laboratory setting				
Commercial laboratory	69	57.0		
University/academic laboratory	25	20.7		
Hospital-based laboratory	14	11.6		
Provincial/regional health service	10	8.3		
State/public health laboratory	0	0		
Other/no response	3	2.5		
Type of laboratory <sup>a</sup>				
Molecular laboratory	99	81.8		
Cytogenetics laboratory	46	38.0		
Biochemical laboratory	18	14.9		
Maternal serum screening laboratory	11	9.1		

**Table 1** (continued)

Variable	<i>n</i>	%	Mean	Median (range)
Newborn screening laboratory	10	8.3		
Hospital send-out laboratory	5	4.1		
Free fetal DNA laboratory	5	4.1		
Direct to consumer testing laboratory	0	0		
Other	2	1.7		
Number of other genetic counselors				
Within respondent's laboratory			11.1	4 (0–80)
Within respondent's institution			17.3	15 (0–80)

<sup>a</sup> Respondents could choose multiple options

least one task in this area (Tables 2 and 3). The most frequently performed tasks were making phone calls with genetic testing results (56.2 % performed frequently), obtaining clinical or family history information for use in result interpretation (52.5 %), and composing case-specific interpretations for unique results and/or obtaining literature references to support interpretations (51.2 %) (Table S-II). Other frequently performed tasks included drafting standard reporting comments (47.1 %), investigating clinical significance of variants (42.1 %), and reviewing results data (41.3 %) (e.g., sequencing results, prenatal screen results, etc.).

A minority of respondents signed genetic test reports; 24.8 % of respondents indicated they did this frequently, while 64.5 % indicated that they never sign reports. The vast majority of respondents who signed reports did so in conjunction with a laboratory director (94.9 %) (Fig. 1). Respondents working in cytogenetics, molecular and maternal serum screening laboratories frequently signed results more so than respondents in other types of laboratories (26.3, 26.1 and 18.2 % respectively) (Table 4). It was common for these LBGCs to sign normal, abnormal, and uncertain or equivocal test results in conjunction with a laboratory director (74.4, 76.9, and 74.4 % respectively) (Fig. 1). A minority of survey

respondents reported that they signed test reports without a laboratory director's signature; typically for canceled test reports. Only two individuals (5.1 %) reported signing abnormal or normal test reports without a director.

The majority (87.6 %) of respondents called out some type of final test results (Table S-II). Only 10 % of respondents called out normal results, while greater than 50 % of respondents called out abnormal, prenatal, complex and other results (Fig. 2). Results were most commonly called out to clinicians, followed by laboratory staff, and only 4 % of respondents reported calling results to patients (unless specifically requested, then 5.7 % of LBGCs reported calling patients with results).

### Sales and Marketing

The sales and marketing category accounted for 9.8 % of respondents' time (Table 2). The sales and marketing task that respondents performed most frequently was creating, editing, and/or reviewing marketing materials (46.3 %) (Table S-III). The majority of respondents never or rarely met with clients or clinicians (52.1 %) or contacted clients or clinicians to promote testing (62.8 %). Staffing booths at conferences was

**Table 2** Time devoted to LBGC role categories

Role category	Mean % of time spent performing roles/tasks	Most frequently performed role/task within category
Customer liaison/case coordination	30.2	Answer client/clinician questions regarding test algorithms and strategies
Interpretation and result reporting	26.1	Make phone calls with genetic test results
Sales and marketing	9.8	Create/edit/review marketing materials
Counseling patients	7.3	
Research and publications	6.9	Analyze research data
Management duties	5.2	Involvement in laboratory staffing decisions
Test development and performance	5.0	Develop new protocols/procedures
Website and database support	4.2	Maintain external laboratory websites (e.g., online test catalogs)
Education and supervision of students	3.3	Educate other healthcare providers (e.g., lectures, seminars)
Insurance and billing	2.1	Communicate insurance/billing information to clinicians

**Table 3** Percentage of LBGs performing a job role (very often/often/sometimes/rarely) by type of laboratory

	Molecular	Cytogenetics	Biochemical	Maternal serum	Newborn screening	Hospital send-out	Free fetal DNA
Customer liaison/case coordination	99.0 %	100 %	100 %	100 %	100 %	100 %	100 %
Interpretation/result reporting	97.0 %	97.8 %	100 %	90.9 %	90.0 %	100 %	80.0 %
Sales/marketing	89.9 %	84.8 %	66.7 %	70.0 %	63.6 %	80.0 %	80.0 %
Research/publication	88.9 %	82.6 %	77.8 %	54.6 %	90.0 %	100 %	60.0 %
Test development/performance	93.9 %	95.7 %	88.9 %	90.9 %	100 %	100 %	80.0 %
Website and database support	69.7 %	76.1 %	88.9 %	63.6 %	70.0 %	80.0 %	40.0 %
Education/supervision of students	100 %	100 %	100 %	100 %	100 %	100 %	100 %
Insurance/billing	83.8 %	84.8 %	66.7 %	72.7 %	60.0 %	80.0 %	60.0 %

more evenly split, with 41.7 % indicating they never or rarely performed this job, 35.8 % indicating they sometimes performed this job, and 22.5 % indicating that they frequently performed this job. By laboratory type, LBGs in free fetal DNA laboratories reported the most frequent performance of tasks in sales and marketing in sharp contrast to LBGs in hospital-based send out laboratories (Table 4).

#### Research and Publication

The majority of LBGs across different types of laboratories reported performing some tasks within research and publication (Table 3), accounting for an average of 6.9 % of their time (Table 2). The two tasks performed most frequently were collecting and analyzing data (20.0 and 24.5 %, respectively) (Table S-IV). LBGs in university/academic laboratory settings more frequently wrote IRB protocols, recruited and consented research participants, collected and analyzed data, submitted data to public databases for research, and wrote publications (Fig. 3). In contrast, hospital-based LBGs more frequently drafted consent forms than both university/academic and commercial laboratory genetic counselors. Of the 106 individuals engaging in research, 53 % have held a research title of primary investigator, co-primary investigator, and/or project or study coordinator. The most common title was project or study coordinator, held by 41.2 % of LBGs who performed research.

#### Management Duties

The average percentage of time spent on management duties was 5.2 % (Table 2). Respondents were asked whether or not they were involved in different management duties, rather than how frequently they were involved in these activities. LBGs reported management duties that included involvement in laboratory staffing decisions (38.8 %), recruiting employees (37.2 %), supervising other employees (32.2 %), and writing

performance evaluations of employees (30.6 %). The least frequently performed task related to management duties was involvement in laboratory purchasing decisions (9.1 %).

#### Test Development and Test Performance

On average, respondents spent 5.0 % of their time on test development and test performance (Table 2). The most frequently performed tasks were developing new protocols/procedures (37.5 %) and assisting in decisions regarding what tests should be developed (36.4 %) (Table S-V). While tasks related to test development and performance were not frequent roles for LBGs, greater than 80 % of LBGs across all types of laboratories reported some level of involvement (Table 3). The majority of LBGs indicated that they were rarely or never involved with the validation of new tests or test performance, but those in molecular laboratories assisted with test validation more frequently than other LBGs, with 19.2 % reporting frequent involvement in this role (Table 4). The development of new protocols and procedures related to test development was a more common role in maternal serum screening, newborn screening and hospital send-out laboratories.

#### Website and Database Support

The average percentage of time spent on website and database support was 4.2 % (Table 2). Tasks related to laboratory website and database support were performed with similar frequency, with 22.3 to 27.3 % of respondents performing maintenance of laboratory-specific computer systems/external laboratory websites and contributing to public websites, respectively (Table S-VI). Tasks related to website and database support were less often performed by LBGs in free fetal DNA laboratories, and more commonly reported by LBGs in biochemical genetics laboratories (Table 3).

**Table 4** Frequently (very often/often) performed tasks by type of laboratory

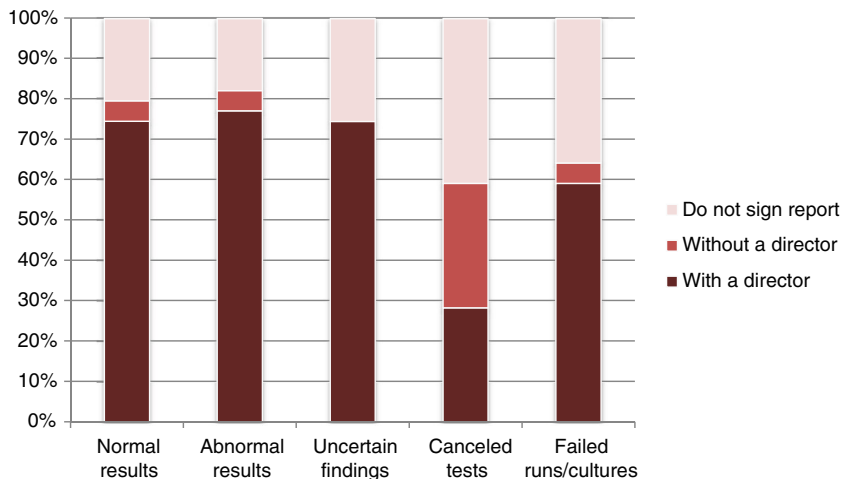
	Molecular	Cytogenetics	Biochemical	Maternal serum	Newborn screening	Hospital send-out	Free fetal DNA
<b>Customer liaison/case coordination tasks</b>							
Resolve specimen handling issues	34.3 %	50.0 %	27.8 %	20.0 %	20.0 %	60.0 %	20.0 %
Answer questions directly from patients	22.2 %	13.0 %	5.6 %	9.1 %	10.0 %	0 %	0 %
Facilitate send-outs	18.2 %	28.3 %	22.2 %	9.1 %	0 %	80.0 %	20.0 %
<b>Interpretation/reporting tasks</b>							
Investigate variants	45.5 %	56.5 %	11.1 %	0 %	20.0 %	20.0 %	0 %
Review result data	44.4 %	45.7 %	33.3 %	54.6 %	30.0 %	20.0 %	60.0 %
Sign reports	26.3 %	26.1 %	5.6 %	18.2 %	0 %	0 %	0 %
<b>Sales/marketing tasks</b>							
Meet with client	30.3 %	10.9 %	5.6 %	27.3 %	30.0 %	0 %	60.0 %
Call/email client	19.2 %	13.0 %	11.1 %	36.4 %	30.0 %	0 %	40.0 %
Staff booths	25.5 %	10.9 %	5.6 %	9.1 %	0 %	0 %	40.0 %
<b>Test development/test performance tasks</b>							
Assist with validation	13.4 %	6.8 %	5.9 %	0 %	0 %	0 %	0 %
Involvement in test performance	19.2 %	17.4 %	16.7 %	18.2 %	20.0 %	20.0 %	20.0 %
Test development decisions	40.4 %	34.8 %	11.1 %	18.2 %	10.0 %	40.0 %	40.0 %
Develop protocols	36.4 %	31.1 %	50.0 %	63.6 %	60.0 %	60.0 %	20.0 %
<b>Education/supervision tasks</b>							
Genetic counseling students	22.2 %	15.2 %	11.1 %	9.1 %	0 %	0 %	0 %
Other students	38.4 %	28.3 %	33.3 %	18.2 %	10.0 %	20.0 %	0 %
Laboratory staff	28.3 %	26.1 %	5.6 %	9.1 %	30.0 %	0 %	20.0 %

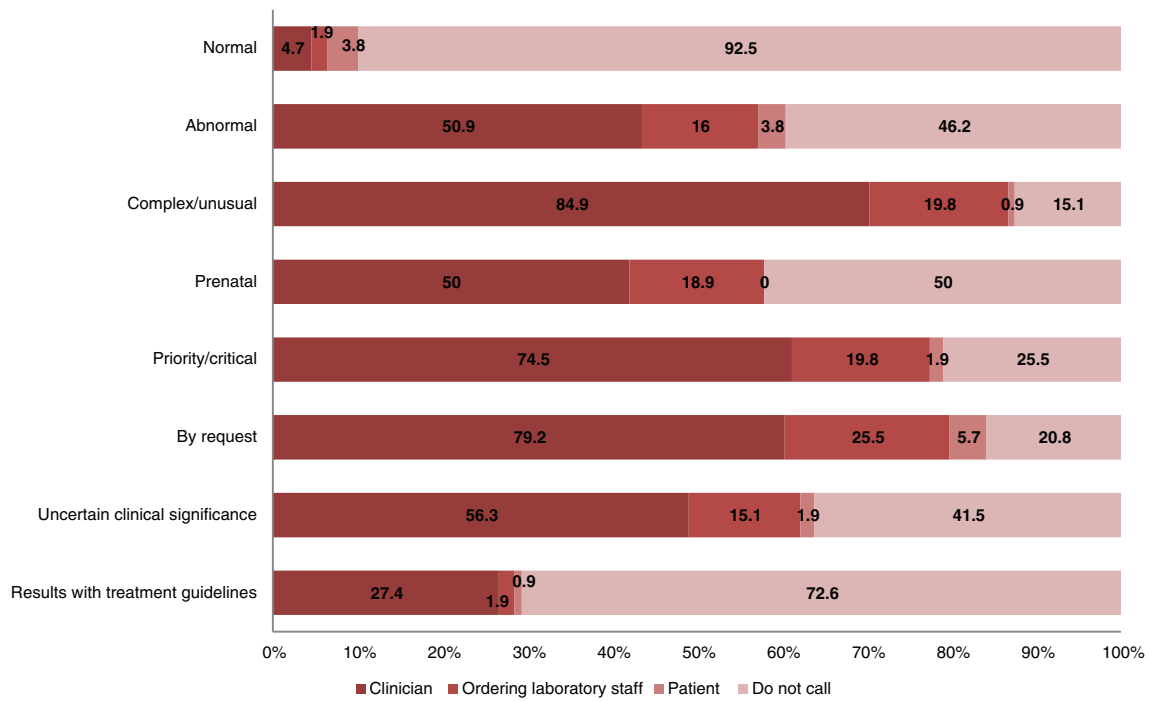
*Education and Supervision of Students*

All respondents indicated performing at least one of the surveyed tasks in education and student supervision, but this accounted for just 3.3 % of their time (Table 2). The educational tasks performed most frequently were education of other healthcare providers through lectures or seminars (39 %), education of students (33.9 %), and education of other laboratory staff

28.9 % (Table S-VII). Specifically related to supervising genetic counseling students, 66.9 % of LBGCS reported some level of involvement, but less than a quarter (18.2 %) reported frequent supervision. By laboratory setting, LBGCS in university or academic settings were most likely to supervise genetic counseling students, with 40 % reporting frequent involvement with this task. Interestingly, by laboratory type, no LBGCS in newborn screening, hospital send-out or free fetal

**Fig. 1** LBGCS signing test results, with and without a laboratory director, by result type (n = 43)





**Fig. 2** LBGCs calling test results to clinician, laboratory staff and/or patient, by result type (n = 106)

DNA settings reported supervising genetic counseling students (Table 4).

*Insurance and Billing*

Respondents spent the least amount of time on insurance and billing issues (2.1 %) (Table 2). The majority of respondents were never or rarely involved with tasks related to insurance and billing. Only 1.7 % of respondents were frequently involved in obtaining insurance preauthorization for testing, with 93.4 % indicating they never performed this task (Table S-VIII). Only 4.1 % of respondent were frequently involved in communicating insurance/billing information to patients, with 81.0 % indicating they never performed that task.

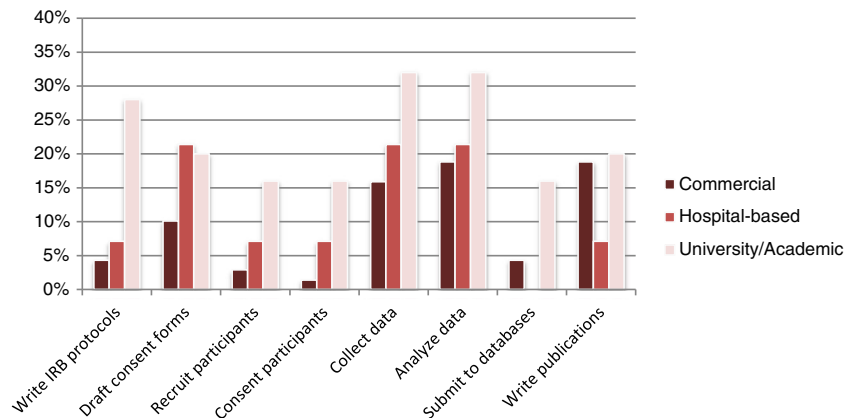
*Counseling Patients*

The average percentage of time spent counseling patients face-to-face in a clinical setting was 7.3 % (Table 2), with 24.0 % of respondents indicating that they performed this task. LBGCs reported a wide spectrum of settings in which they provided clinical services, including prenatal, pediatric, cancer, and adult specialty clinics.

*Additional Roles*

The respondents were asked to comment on any additional tasks they performed that were not identified throughout the survey. The free text responses were consolidated and only a few unique tasks were identified. These additional tasks included: biobank management, College of American

**Fig. 3** Frequently (very often/often) performed research/publication tasks by laboratory setting





Pathologists (CAP) and Clinical Laboratory Improvement Amendments (CLIA) inspection preparedness, market analysis, and policy making.

### Job Satisfaction

Overall, LBGCs reported a high level of job satisfaction, with 52.1 % ( $n=63$ ) of respondents indicating they were ‘satisfied’ and 44.6 % ( $n=54$ ) indicating they were ‘very satisfied’. Five respondents commented on autonomy positively affecting satisfaction and four noted that they were highly satisfied with the variety of roles and opportunities their current position provided. Five counselors indicated that their role in the laboratory allowed them to feel important and to have a broad impact on patient care. The level of flexibility and feeling of being valued were other recurrent themes.

“I love what I do. I like coming into work not knowing what the day will bring, it keeps you on your toes. I like being able to help with MANY cases in 1 day—instead of meeting intensively with a handful of patients in a day, I might help 20! .... I feel like I’m making a difference—if these patients were my own family members I would want someone like me making sure everything goes smoothly.”

“Although I am not counseling patients I have found that I like the independence and autonomy that this position has provided me. I feel like I am a valuable member of the team and that I contribute to proper functioning of the laboratory and provincial testing program.”

Only four respondents reported being “dissatisfied” or “very dissatisfied” with their current positions (3.3 %). Respondents identified several factors as negatively affecting job satisfaction: staffing issues and interpersonal issues (3 respondents), too many responsibilities (4 respondents) and feeling under-valued (2 respondents). One respondent noted that the counselor role is new at many companies and it may take additional time for the true value and appropriate use of genetic counselors to be completely realized and embraced.

### Discussion

The purpose of this study was to further describe the role of the laboratory genetic counselor by building on the data published by Christian et al. (2012). In addition to reevaluating the time devoted to the roles identified in Christian et al.’s study, the current study also further delineated these roles, querying specific tasks within each category. An additional aim was to evaluate whether laboratory settings had a significant impact on the types of tasks being performed by LBGCs.

Considering the rapid growth of this field, comparison of the data between the previous and current studies provides insight into the similarities, differences, and possible trends in the roles of LBGCs. The data highlights the wide variety of roles performed and elucidates a few major roles performed frequently by a majority of respondents which account for a significant proportion of time. It also revealed common roles, those accounting for a small percentage of time but performed by the majority of respondents. Minor roles were those with a low percentage of time and those performed infrequently or not at all by most LBGCs.

### Major Roles

Consistent with the Christian et al.’s (2012) study, results of the current study indicate that the most frequently performed roles for LBGCs are customer liaison/case coordination and interpretation/result reporting. The most frequently performed role in the current study was customer liaison/case coordination. Commonly performed tasks within this role were consistent with Scacheri et al.’s (2008) previous description of the LBGC role, including “[being] available to discuss specific testing strategies, assist in pedigree assessment, discuss the benefits and limitations of genetic testing for the patient, describe the experience the laboratory has with that test, and discuss ethical and other considerations with the clinical professional” (Scacheri et al. 2008).

This study also addressed LBGCs in test utilization management roles, which was not previously addressed in the Christian et al. (2012) study. Utilization management (UM) is the evaluation of the medical necessity, appropriateness, and efficiency of the testing in an effort to help improve patient care and reduce healthcare spending, two issues of increasing focus in the current era of healthcare reform (Dickerson et al. 2014; Kotzer et al. 2014; Miller et al. 2014). Results indicated that UM efforts are a common LBGC role, as a majority of respondents frequently determined test appropriateness and corrected ordering errors. The present study found that this was a particularly important role for hospital-based LBGCs, who indicated that performing test UM activities, such as resolving specimen handling issues and facilitating send-outs, were the most frequent tasks in customer liaison and case coordination roles. UM roles for LBGCs are likely to continue to expand in light of recent publications highlighting the integral role of genetic counselors in laboratory test UM initiatives to increase the value of laboratory testing and reduce the cost of healthcare spending. (Dickerson et al. 2014; Kotzer et al. 2014; Miller et al. 2014).

The frequency of specific tasks in certain categories differed depending on the type of laboratory, perhaps reflecting some unique aspects of different laboratory settings. For example, within the customer liaison/case coordination category, resolving specimen handling issues was more frequently

performed by LBGCs in cytogenetics laboratories than in other types of laboratories. In cytogenetics labs, the complexity of cell culturing and the need for further molecular studies may account for some of the increased involvement of LBGCs in specimen handling issues.

As was seen in the Christian et al. (2012) study, a second major role category of LBGCs was interpretation and result reporting. The most frequently performed task within this role was making phone calls with genetic test results. Consistent with previous studies, results of the current study indicate that the laboratory genetic counselor most commonly communicated abnormal, complex/unusual, or critical results to clinicians (Amos and Gold 1998; Scacheri et al. 2008; Zetzsche et al. 2013).

Investigating clinical significance of variants and reviewing result data (such as sequencing results) was a frequent role for 42.1 % of respondents. These tasks were performed most often by cytogenetic and molecular LBGCs. As chromosomal microarray has become routine and the availability of large-scale sequencing panels has exponentially increased over the last several years, the need for variant interpretation has also greatly expanded. These results show a trend of genetic counselors filling this interpretative role.

The proportion of LBGCs who reported signing reports decreased from 44.2 % in the Christian et al. (2012) study to 35.5 % of respondents in this study. The majority (64.5 %) in this study responded that they never sign reports. In relation to genetic counseling licensure efforts, the College of American Pathologists recently published an Issue Brief indicating that they do not support the scope of practice of genetic counselors to include the interpretation of genetic test results (CAP 2014), which could have implications for LBGCs' ability to sign genetic test results, since an individual's signature on a test result is a statement of 'interpretation by.' However, it is unclear if CAP guidelines would prevent LBGCs from signing reports in collaboration with a laboratory MD/PhD director, or from independently signing canceled test reports.

### Common Roles

While roles in sales and marketing were not performed frequently amongst respondents, the majority of respondents across laboratories reported performing some aspects of this role. The percentage of time devoted to sales and marketing (9.8 %) more than doubled from the amount of time reported by LBGCs in the Christian et al. (2012) study (4.07 %) but still accounts for less than 10 % of time. This may indicate an increase in sales and marketing of genetic testing in general, as well as an increasing role of LBGCs in this area, in which they previously had little or no involvement. Amongst laboratory types, LBGCs in free fetal DNA laboratories stood out as having the largest role in sales and marketing, which may reflect the newness of this technology, the current high

commercial marketability of the testing and/or that LBGCs in free fetal DNA laboratories are perhaps hired specifically for sales and marketing roles.

Test development and performance was another common role in the current study, with the majority of LBGCs reporting some role in this category despite accounting for only 5.0 % of LBGCs' time on average. Test development activities were widely reported as duties "sometimes" performed, including decisions regarding development of new tests, creation of test algorithms, and development of new protocols or procedures. LBGCs in molecular laboratories reported assisting with test validation more frequently than other LBGCs, perhaps reflecting the tremendous growth in molecular testing options due to next-generation sequencing technology. Test development is a potential area for increased involvement of LBGCs, as their clinical expertise and work with clients and clinicians provides valuable perspective for the test development process.

Though 100 % of respondents in this study reported some role in teaching and supervising students, this remained a minor or infrequent role for most LBGCs, as noted by Christian et al. in (2012). It is not surprising that LBGCs in university or academic settings were most likely to supervise genetic counseling students, with 40 % reporting frequent involvement in this role. Interestingly, no LBGCs in newborn screening, hospital send-out or free fetal DNA settings reported supervising genetic counseling students, although newborn screening and free fetal DNA LBGCs reported a relatively high frequency of educating other laboratory staff and hospital send-out LBGCs played a role in the education of other students. Given the apparent lack of genetic counseling student supervision within newborn screening, hospital send-out, and free fetal DNA laboratories and the growing number of genetic counselors in these settings, this may be an area for graduate school programs to explore offering rotations in the future.

### Minor Roles

Similar to the Christian et al. (2012) study, this study confirmed that management, website and database support, and research are performed less frequently by LBGCs. Additionally, although Christian et al. did not specifically ask about LBGC roles related to billing and insurance, the current study found that the majority of LBGCs were never or rarely involved in these roles. Since many genetic testing laboratories currently offer third party billing and preauthorization services, this suggests that this role is being performed by other laboratory personnel, likely those with specialized training in coding and billing.

Of note, almost one-third of respondents indicated that they supervise other employees and this was not unique to any type of laboratory. This is approximately twice the number of

clinical genetic counselors who reported having managerial responsibilities or direct reports (17.6 %) (National Society of Genetic Counselors 2014). This may point to LBGCs having career growth opportunities that are less likely in clinical settings. It would be interesting to explore this role to further understand the nature of the role and the type of direct reports LBGCs are supervising.

The time spent on research and publication activities increased from 2.7 % in the Christian et al. (2012) study to 6.9 % in this study. Compared to all 2014 PSS survey respondents, LBGCs in this study reported more involvement in research compared to the genetic counseling community overall (88.4 versus 49.8 %) (National Society of Genetic Counselors 2014). Of respondents, 38.8 % reported writing IRB protocols compared to 24.6 % of all genetic counselors. Research and publication may be areas for growth and career development for LBGCs. Opportunities for research topics are plentiful in the laboratory setting: genetic testing on specific disorders, research on genetic databases and biobanks, utilization efforts, and the field of laboratory genetic counseling are just some examples of research topics LBGCs may explore.

### **Job Satisfaction for Laboratory-Based Genetic Counselors**

Results of this study demonstrated a high level of job satisfaction among respondents (52 % and 45 % of respondents indicated they were 'satisfied' or 'very satisfied,' respectively), consistent with results of the 2014 NSGC Professional Status Survey (PSS) where 50.7 % of non-clinical genetic counselors reported being 'satisfied' and 38.0 % reported being 'very satisfied' with their jobs. Based on comments received from respondents, clinical laboratory settings appear to provide many opportunities for professional growth and development of genetic counselors outside of traditional clinical roles. When asked to comment on job satisfaction in their current role, autonomy was the most common theme reported in relation to a high level of job satisfaction. These results are in agreement with the 2014 NSGC PSS, which reported that 92.1 % of non-clinical genetic counselors were 'satisfied' or 'very satisfied' with the level of autonomy they maintain in their position (National Society of Genetic Counselors 2014). Interestingly, the PSS also indicated that non-clinical counselors expressed higher levels of satisfaction than their clinical peers in many aspects of their jobs.

### **Professional Background of Laboratory-Based Genetic Counselors**

Among the respondents of this survey, 58 % were practicing genetic counselors prior to taking a laboratory position. This

could suggest that previous clinical experience provides an advantage to genetic counselors interested in laboratory positions, or that newer graduates are more likely to gravitate to clinical genetic counseling positions. Alternatively, it may suggest that experienced genetic counselors may be seeking laboratory positions. This may be especially true as more flexible work arrangements, including remote positions, are being offered for LBGC positions. There have been several reasons reported for clinical genetic counselors moving to laboratory-based positions, including work/family balance, desire for new opportunities, financial, career advancement, and compassion fatigue/burnout (Dickerson et al. 2015). Additional studies could examine whether this appears to be a national trend in the genetic counseling profession.

### **Study Limitations**

This study utilized a self-created survey that, while piloted with practicing LBGCs, was not formally validated. Due to the cross-sectional nature of this study, results describe the practices of the specific survey respondents at the time the survey was administered, and may therefore not be widely generalizable to all potential work environments and job duties for LBGCs. In particular, since the creation of this survey, there seems to be an increasing number of LBGCs working remotely. This was not queried in the survey so it is unknown whether these roles differ from on-site LBGCs and whether working off-site impacts job satisfaction. Also, the implications of certain work environments on the frequency of corresponding LBGC roles may not have been adequately captured in this study. For example, in laboratories in which job duties are divided among multiple LBGCs (as indicated by two-thirds of respondents), or for respondents who work in more than one type of laboratory (as indicated by over one-third of respondents), it was difficult to accurately assess the frequency of each task as it pertained to a specific laboratory. Additionally, laboratories with multiple LBGCs may have been over-represented as they may have been more likely to hear about the study from colleagues, whereas LBGCs in subspecialties, i.e., hospital send-out laboratories and free fetal DNA laboratories, may be underrepresented or data may not accurately reflect their roles because of the lower number of participants from these subspecialties. Finally, responses may be influenced by differing interpretations of questions where terminology may not have been clearly defined, in particular pertaining to the section of the survey regarding signing laboratory reports. It was unclear whether respondents who indicated signing laboratory reports were implying that they were interpreting results in those cases.

## Conclusion

The number of practicing LBGCs continues to grow, in parallel with tremendous development in the genetic testing industry. There is increasing evidence of the benefit of LBGCs, both to the genetic testing laboratory and patient care (Miller et al. 2014; Scacheri et al. 2008). This study highlights the variety of roles performed by laboratory genetic counselors and confirmed previous descriptions of the LBGCs primary roles in customer liaison, case coordination, and interpretation and result reporting. The study results also point to trends of expanding roles in sales and marketing, variant interpretation and management responsibilities. Results of this study may be useful for further definition of the full scope of practice of LBGCs. This may aid in the development of new LBGC positions or expansion of current positions, including growth of roles related to test development, research, and student supervision.

## Training Implications

The authors purport that this data may be helpful in the creation of tools for genetic counseling training programs, for practicing genetic counselors to prepare for a role in the laboratory, and for laboratory genetic counselors to understand where other LBGCs in specific subspecialties are focusing their time. Swanson et al. discussed the future growth of and need for laboratory genetic counseling in parallel to advancements and clinical applications possible with next generation sequencing (Swanson et al. 2014). They suggested that professional training should adapt in order to prepare genetic counselors for roles in variant curation and integration of clinical information into customized interpretations and reports, a role that our survey results indicated is expanding. Current standards for genetic counseling graduate programs require students to have instruction in, and exposure to, genetic laboratories but the extent and variety of this exposure is variable (Accreditation Council for Genetic Counseling 2013). As the number of LBGC positions is growing, perhaps program directors and LBGCs can work together to create more opportunities for students to work in laboratory settings and establish some criteria for proficiency. This may also warrant exploration of whether the ABGC Practice-Based Competencies should be expanded to incorporate skills commonly used by LBGCs, such as variant interpretation and test utilization management. Additional research may be beneficial to assess the genetic counseling graduate programs regarding LBGC skill development opportunities including exposure to LBGC supervisors, rotations in genetic laboratories, and practice in drafting test interpretations and reviewing variants for clinical significance. Further research could explore how remote LBGC positions differ from on-site positions, especially regarding specific job

roles, clinician versus direct patient contact, and job satisfaction.

## Compliance with Ethical Standards

**Conflict of Interest** Lindsey Waltman, Cassandra Runke, Jessica Balcom, Jacquelyn D. Riley, Margaret Lilley, Susan Christian, Lindsay Zetzsche, and McKinsey L. Goodenberger declare that they have no conflict of interest.

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

**Animal Studies** No animal studies were carried out by the authors for this article.

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