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The challenges and opportunities of offering and integrating training in clinical molecular genetics and clinical cytogenetics: A survey of LGG Fellowship Program Directors

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Conflict of Interest

The authors declare no conflicts of interest.

Ethics Declaration

This study was assessed by the UCLA Institutional Review Board and was determined to be exempt.

Additional Information

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Abstract

Purpose: The specialty of Laboratory Genetics and Genomics (LGG) was created in 2017 in an effort to reflect the increasing convergence in technologies and approaches between clinical molecular genetics and clinical cytogenetics. However, there has not yet been any formal evaluation of the merging of these disciplines and the challenges faced by Program Directors (PDs) tasked with ensuring the successful training of laboratory geneticists under the new model.

Methods: An electronic multi-question Qualtrics survey was created and was sent to the PD for each of the Accreditation Council for Graduate Medical Education–accredited LGG fellowship programs at the time. The data were collected, and the responses were aggregated for each question.

Results: All of the responding PDs had started training at least 1 LGG fellow. PDs noted challenges with funding, staff shortages, molecular/cytogenetics content integration, limited total training time, increased remote work, increased sendout testing, and a lack of prior cytogenetics knowledge among incoming fellows.

Conclusion: This survey attempted to assess the challenges that LGG PDs have been facing in offering and integrating clinical molecular genetics and clinical cytogenetics fellowship training. Common challenges between programs were noted, and a set of 6 concluding comments are provided to facilitate future discussion.

Keywords

ACGME; Cytogenetics; Fellowship; Molecular genetics; Training

Introduction

Different options for clinical fellowship training in molecular genetics and/or cytogenetics have coexisted for several years. Before 2017, the American Board of Medical Genetics and Genomics (ABMGG) accredited fellowship training sites in clinical molecular genetics and/or cytogenetics and offered certification examinations to individuals with an MD and/or

PhD completing a 2-year program in one or the other discipline. Dual certification was accomplished following a combined 3 years of training, generally done in tandem with 2 years in 1 area and 1 year in the second area, although some programs enforced 2 years of training in each specialty when performed back to back. In addition, for pathologists who are board certified in anatomic and/or clinical pathology, as well as MD medical geneticists, the American Board of Pathology (along with the ABMGG) offers certification in Molecular Genetic Pathology (MGP), which is a 1-year Accreditation Council for Graduate Medical Education (ACGME)–accredited fellowship program.[1–3] MGP training includes germline and molecular infectious disease testing, with an emphasis on somatic hematopoietic neoplasms and solid tumor testing.[4]

In 2017, the ABMGG combined the 2 specialties into a single new specialty named Laboratory Genetics and Genomics (LGG) in an effort to reflect the increasing convergence in technologies and approaches that had been occurring in routine clinical laboratory practice for quite some time.[5,6] This new fellowship was accredited by the ACGME, and existing ABMGG programs were given several years to transition to ACGME guidelines and oversite. As of 2023, all of the LGG programs have transitioned the accreditation of their fellowship training; however, there has not as of yet been any formal evaluation of the merging of these disciplines and the challenges faced by Program Directors (PDs) tasked with achieving established milestones and ensuring successful training of laboratory geneticists under the new model.

Therefore, an electronic survey was created and sent to LGG PDs to evaluate specific aspects associated with these training programs, such as fellow recruitment, program funding, and content integration. Although many of these topics may have originally been considered by the PDs at the time that they submitted their initial application for accreditation, the purpose of this survey was to assess how well their conceptualized training program is working in practice as well as document any additional unanticipated challenges that they may have encountered since LGG fellow training was initiated at their institution. A set of 6 concluding comments related to the training of future LGG fellows was formulated based on these results.

Materials and Methods

Survey questions were initially developed and presented to the Lab Directors' Special Interest Group of the Association of Professors of Human and Medical Genetics (APHMG) for further development, clarity, questions, and comments at the APHMG 2022 Annual Meeting (Palm Springs, CA). Following this initial discussion, Qualtrics was used to create an electronic multi-question survey that was certified to be exempt from IRB review per 45 CFR 46.104 category 2. A list of ACGME-accredited LGG fellowship programs and contact information for each of the PDs was retrieved from the ACGME website in November 2022 (there were 39 total accredited programs at the time). An email invitation was sent to each of the PDs, and following a positive initial response, a personalized link to the Qualtrics survey then followed. Once the responses had been received, the data were collected and reviewed to ensure that only 1 set of responses was received from each program, and the responses

were aggregated for each question for the purpose of this study. The initial survey results were presented at the APHMG 2023 Annual Meeting (Kiawah Island, SC).

Results

Invitations were sent to a total of 39 LGG PDs, and positive initial responses were received from 36 of them. Surveys were sent to those 36 PDs, with 1 PD declining to complete the survey because that particular program had not yet had an LGG fellow and thus felt unable to appropriately respond to the survey questions. Complete responses to the survey were received from the remaining 35 PDs for a response rate reflecting about 90% of LGG training sites (35/39). Of the PDs who provided responses, almost half of them (43%) had started training their first LGG fellow in 2017 (Figure 1A) when LGG training was initiated and had thus already acquired more than 5 years of experience in directing an LGG training program and in training LGG fellows. Other programs had started training fellows in the subsequent years, with some having only started within the past year (2022). However, all of the responding PDs had started training at least 1 LGG fellow.

The required length of the ACGME-accredited LGG fellowship training is 24 months, and 25 of 35 (71%) programs indicated that they were providing 2 total years of training (Figure 1B). The remaining 10 of 35 (29%) indicated that they were providing more than 2 years of training. A 2-year training format was offered primarily because PDs indicated that (1) their understanding was that ACGME-accredited LGG training was required to be 2 years in duration (22%), (2) 2 years of training was all that their institution would financially support (13%), and/or (3) they thought LGG fellows could be sufficiently trained in 2 years (9%) (Figure 1C). For PDs who opted instead for a 3-year format, the rationale included that (1) 3 years was in keeping with the duration of the previous ABMGG-accredited combined training (16%), (2) additional dedicated research time was preferred and thus 3 years was the only way to accomplish this (8%), and/or (3) they thought LGG fellows cannot be sufficiently trained in 2 years (17%). Additional comments from PDs regarding their justifications for a 2- or 3-year program are provided in Supplemental Table 1.

Approximately 63% of the programs had a total complement of 1 or 2 fellows at a time (Supplemental Figure 1A), with 80% of the programs consistently able to fill all of their available positions every year (Supplemental Figure 1B). Of the programs who were not able to consistently fill their available positions, 50% indicated that for any given year, there were a sufficient number of applicants but that funding to support fellow training was not always available (Supplemental Figure 1C). A smaller percentage (20%) indicated that they received a sufficient number of applicants but that they were often not of adequate quality to be offered a position (ie, those PDs would rather have their positions go unfilled). The majority of PDs (68%) expected their LGG fellow applicants to have a solid background only in molecular genetics, with 24% of PDs expecting their LGG fellow applicants to have a solid background in both molecular genetics and cytogenetics (Figure 2A). One program indicated that a solid background in one or the other discipline was expected. From the survey, programs who expected applicants to have a solid background in both disciplines only provided 2 years of training (unpublished). In practice, 88% of PDs indicated that the majority of their incoming fellows had a solid background only in molecular genetics,

whereas only 9% of PDs indicated that the majority of their incoming fellows had a solid background in both molecular genetics and cytogenetics (Figure 2B).

In determining what proportion of the total training time should be devoted to one specialty or the other, 80% of PDs indicated that molecular genetics training should potentially comprise 26% to 50% of the training, whereas 63% indicated that cytogenetics training should potentially comprise the same proportion of the total time (Figure 3). In addition, only 14% of PDs indicated that molecular genetics training should potentially comprise up to 75% of the training time, whereas 37% of PDs responded similarly in relation to cytogenetics; overall, PDs seemed to indicate that more time should be spent on cytogenetics training. Two PDs also thought that molecular genetics training potentially only needed to comprise ≤25% of the total training time, although only 1 of them indicated that their incoming trainees generally had a solid background in molecular genetics. Regarding how PDs organized their training programs, approximately 49% chose to provide rotations that were more than 2 months in duration, 20% chose to provide rotations that were 2 months in duration, and 29% chose to provide rotations that were only 1 month in duration (Figure 4A). Only 1 PD offered rotations that were less than 1 month long. In an attempt to integrate training between molecular genetics and cytogenetics through those rotations across the duration of the fellowship time, 43% of PDs alternated rotations between the 2 specialties and repeated rotations as much as possible (Figure 4B). In contrast, 23% of PDs chose to alternate rotations but only provide 1 instance of each rotation. Approximately 35% of PDs attempted to integrate molecular genetics and cytogenetics training within the same rotation, choosing to either offer each rotation once (26%) or several times (9%) throughout the training period.

Because clinical laboratory training does not typically involve direct interaction with patients, given the nature of the recent SARS-CoV-2 pandemic, there has also been a tendency toward increased virtual training for LGG fellows. During March and April of 2020, almost half of the programs (46%) indicated that they switched to at least 50% virtual training, and since then, 60% of the programs had opted to retain virtual training for 25% of the total time, with 36% of the programs retaining virtual training for at least 50% of the total training time (Supplemental Figure 2). Interestingly, in March and April of 2020, 20% of the programs indicated that fellowship training remained 100% in-person with no virtual training options available. Finally, LGG PDs were given an opportunity to enter a free-text response regarding the biggest challenge they had faced thus far in attempting to successfully train LGG fellows. Responses were varied, but multiple PDs noted challenges with many of the items previously mentioned, including funding, staff shortages, molecular/cytogenetics content integration, limited total training time, increased remote work, increased sendout testing, and a lack of prior cytogenetics knowledge among incoming fellows (Figure 5). The full set of PD responses is provided in Supplemental Table 2.

Discussion

This survey attempted to capture information regarding the challenges that LGG PDs have been facing in offering and integrating clinical molecular genetics and clinical cytogenetics

fellowship training. Because approximately 90% of PDs provided responses to this survey, this data set provides a reasonably comprehensive assessment regarding the current status of LGG training. Although almost half of the programs have been training LGG fellows for more than 5 years, more than half of the programs only have 1 or 2 total positions available at any 1 time. Therefore, the majority of this experience may only be based on a few initial trainees for a given program, which may make it challenging for some PDs to thoroughly evaluate the success of their curriculum thus far, and they may also be less inclined to revise their overall training program structure so early in its life span.

The responses from many PDs indicated a concern regarding whether 2 years of total training was sufficient when training in both specialties previously required at least 3 years. The majority of PDs had chosen to restrict their LGG training to 2 years for various reasons, although less than 10% of PDs indicated that 2 years of training seemed sufficient; several PDs also noted that they felt that their training had to be restricted to 2 years, but that they wished it could be 3 years instead. Although it was not specifically mentioned by any of the LGG PDs, providing a sufficient amount of training time in both germline and somatic approaches across both specialties within a 2-year program also presents a challenge, and this may have contributed to some PDs choosing to extend their training programs beyond the accredited 24 months. There is clearly not a consensus yet among PDs regarding the most appropriate total amount of LGG fellowship training necessary.

Multiple PDs indicated experiencing some form of funding/financial challenge in attempting to successfully train LGG fellows, although, unfortunately, the funding source(s) used by each of the individual LGG training programs were not collected as a part of the survey. This may be partially responsible for the generally low complement observed among programs because they may not have sufficient funding to support additional trainees. The majority of PDs indicated that they were typically able to fill their available positions; therefore, it appears that the total complement reported by programs is an accurate reflection of the total number of fellows they are able to train (for financial or other reasons). Funding challenges may also be contributing to why many programs have opted for only 2 total years of training (as was noted by some PDs) because adding a third year of training would increase the total cost to train each fellow. Therefore, some programs may generally be opting to train as many fellows as possible vs training fewer fellows for longer periods of time given their potential budget constraints.

Providing sufficient cytogenetics training was also noted to be a challenge, although this may have been somewhat expected because it was generally felt to be true even when cytogenetics and molecular genetics were offered as separate fellowship training programs. However, it remains as an important professional issue for cytogenetics laboratories needing to recruit additional directors because many newly certified laboratory geneticists may still require additional cytogenetics experience (which may or may not be available) before becoming fully proficient in that area. Almost all of the PDs expected incoming trainees to have prior molecular genetics experience, whereas only 30% of PDs expected incoming trainees to have some prior cytogenetics experience. However, in reality, only 12% of PDs were finding that their incoming trainees had prior cytogenetics experience. Responses from PDs indicated that they may be attempting to address this discrepancy using multiple

approaches including extending their training program to 3 years, further integrating cytogenetics training into more of the rotations, and/or weighting their overall fellowship training time more heavily toward cytogenetics than molecular genetics. Programs can also recommend that trainees take advantage of a number of available cytogenetics didactic resources through organizations such as the American College of Medical Genetics and Genomics (ACMG) and the Association for Molecular Pathology. However, given a general shift toward more molecular-oriented approaches in the field, full training in all of the current cytogenetics techniques may eventually no longer be required by the ACGME and/or necessary for certification in LGG by the ABMGG.

Integration of molecular genetics training and cytogenetics training was attempted by LGG PDs in different ways, with approximately two-thirds of the programs alternating between molecular genetics and cytogenetics rotations without attempting to combine them based on unifying themes. Although this survey did not specifically address the reasons for the different approaches, some of the practical considerations behind the differences in integration strategies may be due to tests being performed in different locations, tests being performed by different groups of technologists, tests being signed out by different individuals, tests being reported in different laboratory information systems and/or different locations within the same electronic medical record, and/or ease of training in each area with focused time initially devoted to one discipline or the other before a more integrated approach later in training. Further research may be needed to determine whether alternating or combining rotations is most suitable for LGG fellow learning.

The increase in virtual learning/training was also noted to be a challenge by multiple PDs, and most of the programs had retained at least some virtual training options at the time that the survey was administered. Although this survey did not attempt to collect any baseline data on pre-pandemic virtual training patterns, it is likely that the current amount of virtual training is higher than it was during the first few years of LGG fellowship training (2017–2019). This likely poses a challenge for LGG PDs who may not have anticipated this change, although this challenge is not specific to LGG training, as evidenced by the increased incorporation of virtual education into clinical care.[7,8] However, as laboratory genetics training becomes more focused on the analysis and interpretation of larger data sets, the challenge for LGG PDs may no longer be how best to incorporate virtual training into their fellowship program but instead how best to retain an appropriate amount of in-person training for hands on troubleshooting, technical expertise, and personnel management skills to achieve competency of LGG fellows.

Finally, several PDs noted that increasing numbers of tests were being sent out from their institution to larger reference laboratories, namely germline molecular genetic tests (eg, gene panels, and exomes). Although trainees may still be able to shadow providers in clinic where these tests are being discussed and ordered and where patients are being consented, and although trainees may still be able to participate in conferences where these results are being discussed with the clinical team, trainees may not be able to observe, perform, analyze, and/or draft reports for these tests, which are all critical for their training. Decisions regarding in-house vs sendout testing may be beyond the control or purview of specific LGG PDs; therefore, alternative approaches (such as the incorporation of additional external

rotations or the creation of new educational modules) may be necessary for those programs to maintain the breadth of training experiences needed for LGG fellow education. Almost 75% of the LGG programs indicated having some required external rotations already in place and almost 50% of the programs indicated having some optional external rotations already in place (Supplemental Figure 3), so perhaps additional external rotations may only be necessary for certain programs.

Taking everything together, several concluding comments related to LGG fellowship training are provided below:

1. The optimal duration for LGG fellowship training is uncertain. Additional research and further discussion is needed in this area as programs continue to obtain experience with training LGG fellows. A correlation with certification examination performance and years of fellowship training and observed shortages of ABMGG-certified workforce may shed light on the appropriate number of years required for training.[9,10]
2. Because clinical genetics workplace shortages currently exist, LGG PDs may want to consider working with their institutions to obtain funding for additional LGG fellowship positions. External sources of funding to support laboratory training (such as those available from the ACMG Foundation) should be considered.
3. Because applicants generally have prior molecular genetics experience, LGG PDs may want to consider weighting their total training time more heavily in favor of cytogenetics over molecular genetics or maintain flexibility in their fellowship training curriculum to accommodate the strengths and weakness of each candidate to obtain competency in both areas for all trainees. A transition to competency-based training may assist PDs in better addressing a trainee's initial and ongoing abilities in clinical molecular genetics and clinical cytogenetics.
4. LGG PDs should continue to explore ways to further integrate molecular genetics and cytogenetics training as much as possible (eg, based on specific clinical themes) to maximize learning during the allotted fellowship period.
5. Because virtual training has increased in recent years, LGG PDs should continue to explore novel mechanisms to engage fellows in virtual training where necessary but should also continue to facilitate in-person interactions with laboratory staff and other genetics personnel (clinical geneticists, genetic counselors, variant scientists, etc) as much as possible.[11]
6. If many of the relevant tests are increasingly being sent out from an individual institution, LGG PDs may want to consider establishing external rotations with other laboratories who may be performing those tests in-house to complement and enhance the training of their LGG fellows. PDs may also want to work to bring additional testing in-house that can be used for LGG training, although those decisions may be beyond their control. Finally, PDs may want to create additional educational modules (using historical cases, for example) that address methods and testing that their laboratory may no longer perform or for which the

current volume and/or breadth of cases is insufficient to support fellow training (eg, cytogenetics cases involving rare abnormalities).

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

Data Availability

The deidentified data used to create the figures and tables are available upon request from jdeignan@mednet.ucla.edu.

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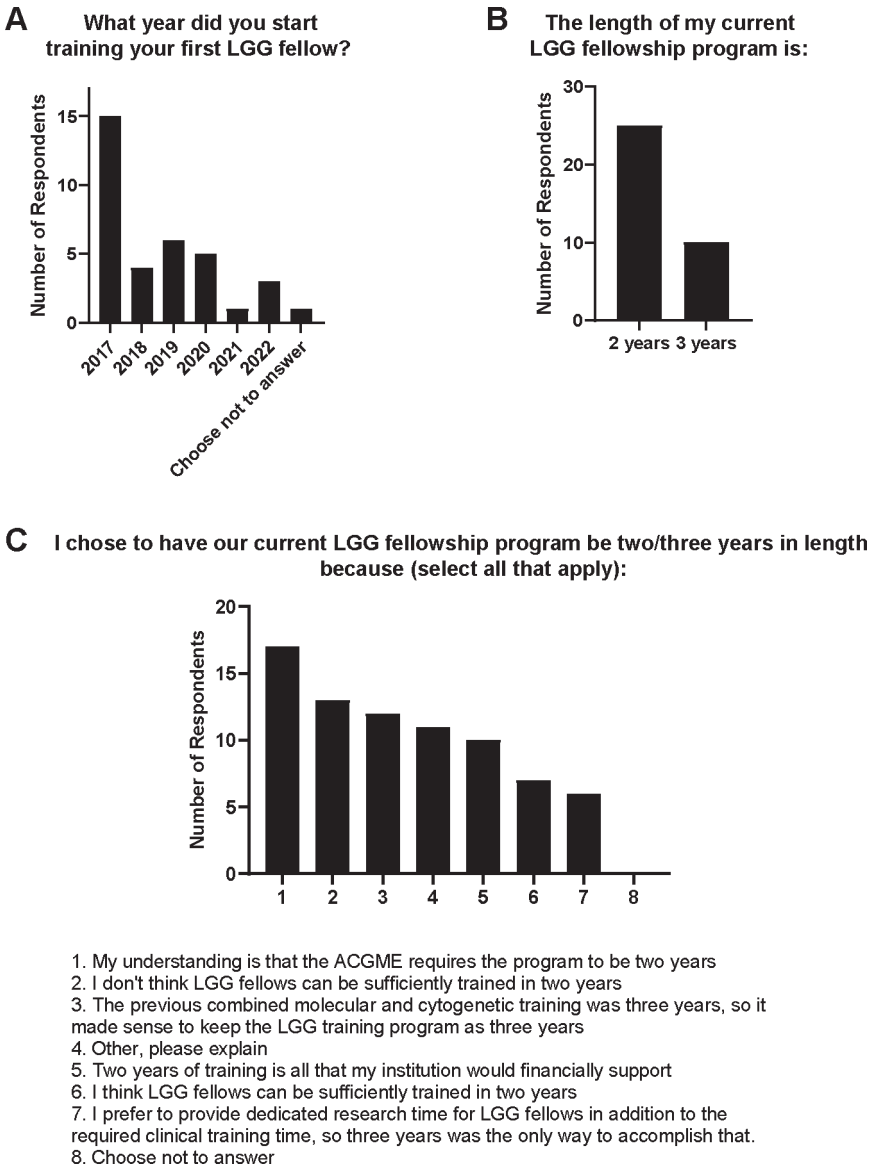

Figure 1. Fellowship program length.

A. Fellowship starting year. B. Current length of fellowship. C. Justification for fellowship length. ACGME, Accreditation Council for Graduate Medical Education; LGG, Laboratory Genetics and Genomics.

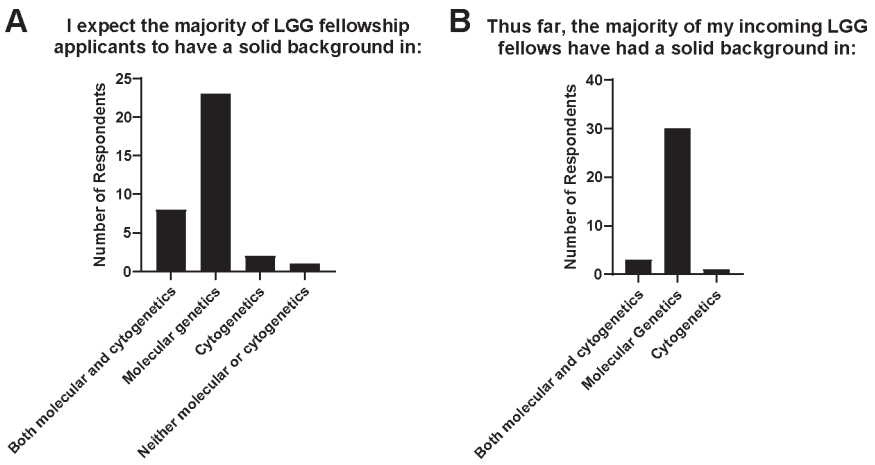

Figure 2. Fellow background.

A. Expectations for fellow applicants. B. Observations from accepted fellows. LGG, Laboratory Genetics and Genomics.

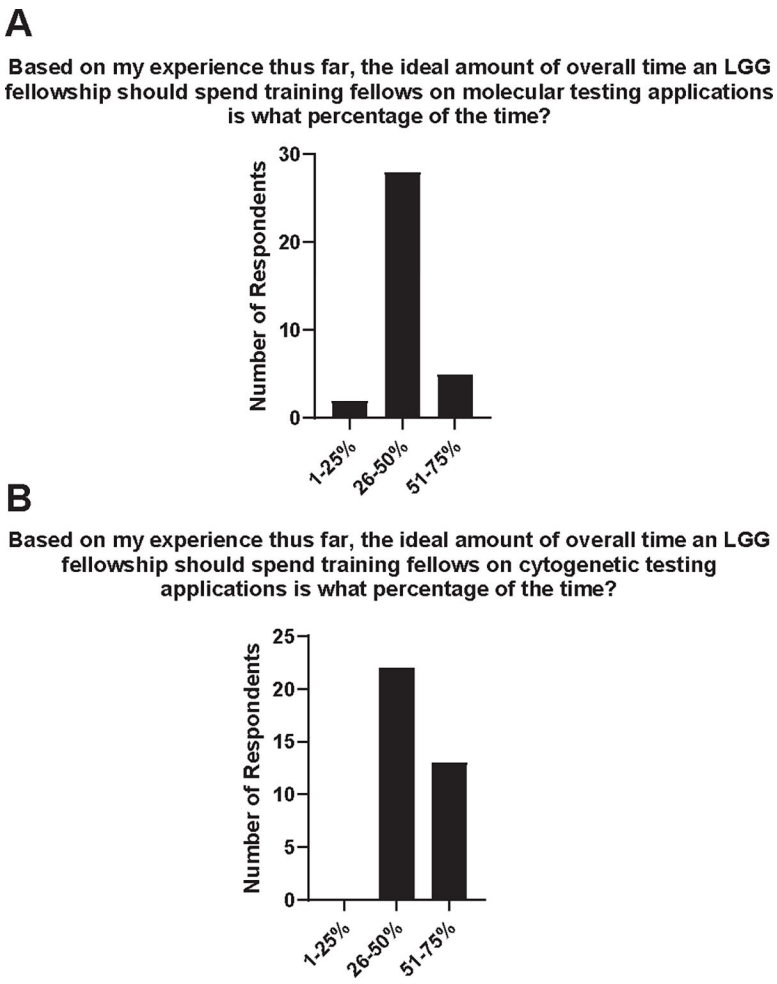

Figure 3. Molecular and cytogenetics training time.

A. Ideal proportion of molecular training time. B. Ideal proportion of cytogenetics training time. LGG, Laboratory Genetics and Genomics.

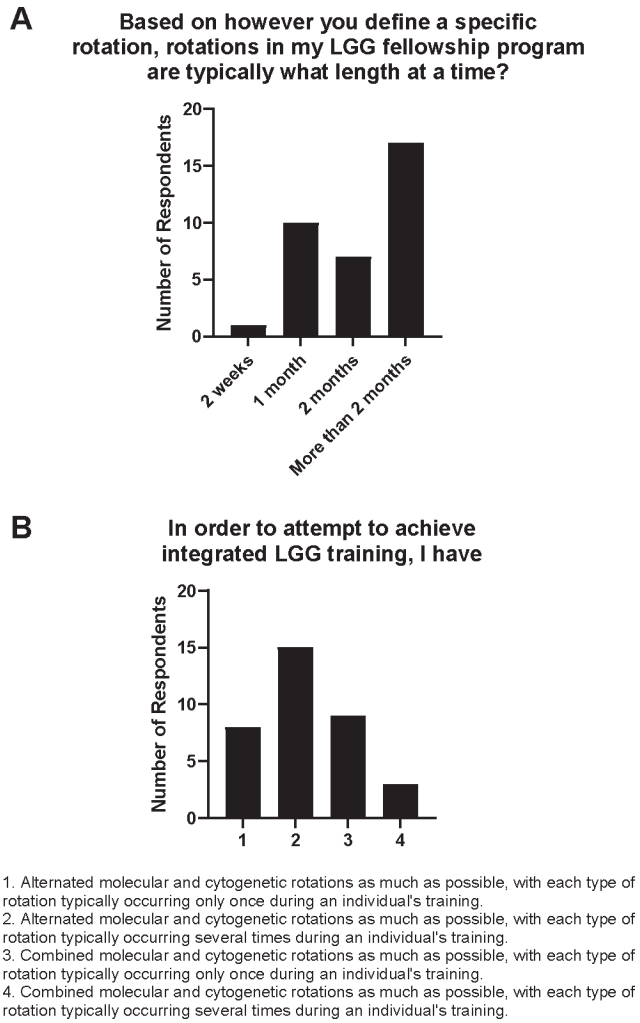

Figure 4. Rotation length and integration.

A. Rotation length. B. Current mechanism for integration. LGG, Laboratory Genetics and Genomics.

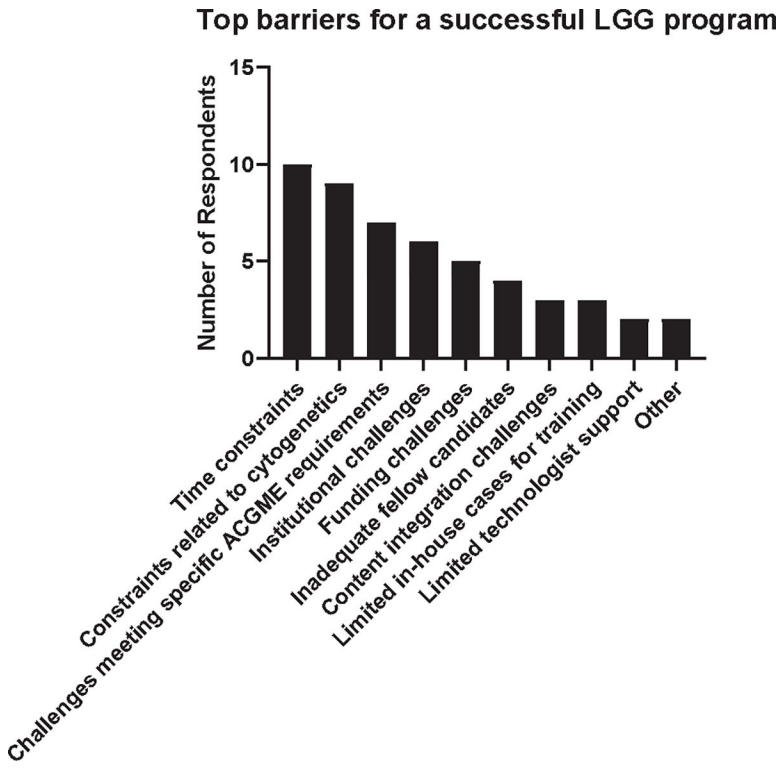

Figure 5. Top barriers for a successful Laboratory Genetics and Genomics (LGG) program.
ACGME, Accreditation Council for Graduate Medical Education.