INCORPERATING GENETICS AND GENETIC COUNSELING IN NEUROLOGY CLINICS: A SURVEY OF NEUROLOGISTS' PERCEPTIONS AND CURRENT PRACTICES

by

Madeline Gorman

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As members of the Master's Committee, we certify that we have read the thesis prepared by Maddie Gorman, titled *Incorporating Genetics and Genetic Counseling in Neurology Clinics: A Survey of Neurologists' Perceptions and Current Practices* and recommend that it be accepted as fulfilling the thesis requirement for the Master's Degree in Genetic Counseling.

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Final approval and acceptance of this thesis is contingent upon the candidate's submission of the final copies of the thesis to the Graduate College.

We hereby certify that we have read this thesis prepared under our direction and recommend that it be accepted as fulfilling the Master's requirement.

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LAND ACKNOWLEDGEMENT

We respectfully acknowledge the University of Arizona is on the land and territories of Indigenous peoples. Today, Arizona is home to 22 federally recognized tribes, with Tucson being home to the O'odham and the Yaqui. Committed to diversity and inclusion, the University strives to build sustainable relationships with sovereign Native Nations and Indigenous communities through education offerings, partnerships, and community service.

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Abstract

Neurogenetics has grown significantly in recent years, yet these advances have not always been fully incorporated into clinical practice. In previous studies, neurologists reported limited confidence in ordering and interpreting genetic tests, highlighting a gap between genetic advances and their application in neurology. Genetic counselors are uniquely trained to facilitate genetic testing, interpret results, and provide psychosocial support, ultimately improving patient care. This study aims to assess the current utilization of genetic testing and genetic counseling within neurology settings. Our study also looks at the association between genetic knowledge and neurologists' perception of value in access to genetic counseling. Through evaluating neurologists' perspectives on genetic testing and counseling, we aim to demonstrate the importance of integrating genetic counselors into neurology settings to improve patient outcomes. A cross-sectional survey was used to assess the utilization of genetic testing and genetic counseling in neurology clinics. Surveys were distributed to board-certified neurologists through email. Quantitative data was analyzed to identify associations between neurologists' experience and their use of genetic services. Out of the 28 completed responses, most respondents have been practicing neurology for more than 15 years and work in urban settings, yet 68% do not have a genetic counselor embedded into their clinics.

Overall, neurologists with more genetics training are more likely to order genetic testing and also feel more comfortable performing genetic counseling roles ($\rho = 0.739$, $p = 3.66 \times 10^{-5}$). Additionally, greater years of experience in practice correlated with increased comfort performing genetic counseling roles ($\rho = 0.519$, $p = 0.005$). However, we found no significant relationship between physicians' level of genetics training or comfort level performing genetic counseling roles and their perceived value of having a genetic counselor in neurology settings,

suggesting that the majority of neurologists value genetic counselors. These results highlight the need for increased integration of genetic counselors in neurology settings to enhance patient care for genetic services.

Introduction

Neurogenetics has experienced significant advances in recent years, however these genetic advances have not always been implemented in clinical settings. The discovery of novel genetic markers associated with both newly described and well characterized neurological conditions has resulted in an increased number of patients for whom genetic testing should be considered (Salm, 2014). Unfortunately, most non-geneticist physicians have limited education in medical genetics and do not have adequate experience with the complexities that are involved with genetic testing (Fellner, 2023). A recent study of non-genetic residents, including neurology residents, showed that over 90% of participants acknowledged a low level of confidence in recognizing genetic disorders and a need for additional medical genetics input (Mladenic, 2023). Similarly, a study of practicing neurologists looking at their attitudes towards genetic testing showed that although 76% had ordered a genetic test in the last 6 months, only 33% were confident on how and where to send genetic tests and a staggering 83.8% reported needing training on how to interpret results (Salm, 2014). This data demonstrates a clear gap in knowledge between genetics and neurology that needs to be filled to provide the best care to patients.

Genetic testing not only gives crucial diagnostic information, it is also becoming necessary for tailoring treatment plans to patients' specific conditions based on their genotype. For example, there are now FDA approved mutation specific therapies for Duchenne muscular dystrophy (DMD) and gene replacement therapies for Spinal Muscular Atrophy (SMA) (Aartsma-Rus, 2016; Ogbonmide, 2023). These new applications paired with decreasing costs and increasing accessibility, have grown public interest in pursuing genetic testing (Sadat, 2021). In a study of 1,215 patients evaluated by a neurologist and a genetic counselor at a University of

Pennsylvania neurogenetics clinic, 90.5% met genetic testing criteria and were offered testing (Guo, 2021). Within that group 30.7% received a positive genetic test result and thus a genetic diagnosis after their evaluation (Guo, 2021). This high yield of positive results exhibits the effectiveness that genetic testing can have when it is utilized in clinical settings, as well as the importance of having specific genetics professionals with expertise in the field. Physicians without training in genetics may lack the time and knowledge to address complex questions and issues regarding genetic testing, especially with how quickly the field is growing (Salm, 2014). Genetic counselors are individuals who can help bridge this gap. They are highly trained in roles and responsibilities such as interpreting genetic test results, facilitating communication and testing among patients and at-risk relatives, working to modify health behaviors and stigmas, and also engaging in psychotherapeutic counseling (Biesecker, 2020). For this reason, an increase in genetic professionals in a clinical neurology setting could have benefits throughout patient care. A collaborative relationship between neurologists and genetics professionals would ensure proper genetic testing and counseling for patients and families (Sadat, 2021).

Neurology itself covers an extensive range of conditions. Epilepsy, or the occurrence of two or more unprovoked seizures, is just one of those conditions. It is theorized that a large portion of unexplained epilepsies have a genetic cause (Smith, 2023). Given this high potential for genetic etiology, in 2022 the National Society of Genetic Counseling (NSGC) published practice guidelines regarding epilepsy care that were also endorsed by the American Epilepsy Society (Smith, 2023). The clinical recommendations encourage offering genetic testing and genetic counseling to all individuals with unexplained epilepsy, regardless of age. They also strongly emphasize the importance of a qualified provider, ideally a genetic counselor, being involved in all aspects of testing, including test selection, informed consent, interpretation, and

follow-up (Smith, 2023). Pre-test counseling should include a thorough informed consent process, particularly when more complex tests such as exome or genome sequencing are considered. Post-test counseling should address results, implications for patient care, and any incidental findings. The guidelines further highlight the need for multidisciplinary collaboration and suggest that integrating genetic counselors or support staff into neurology settings may help manage the increased scale of testing (Smith, 2023). Another condition which genetic counseling guidelines are being implemented in clinical settings is Amyotrophic Lateral Sclerosis (ALS) which is a progressive neurodegenerative disease. It is recommended that genetic testing is discussed with all ALS patients who have first-degree or second-degree relatives who are also affected with the disease (Chiò, 2014). It also strongly suggested that comprehensive counseling, including the psychological and social aspects of testing, be done by a trained professional to ensure the patient receives appropriate support and guidance (Chiò, 2014). These guidelines highlight not only the importance of identifying genetic causes of neurological conditions to provide more accurate diagnosis and treatments, but also the role that genetic counselors play in the process. Genetic counselors help patients and other providers understand results and diagnoses, possible family implications, and are highly trained in assisting in all aspects of the genetic testing and decision-making processes. Implementing these guidelines can drastically improve patient outcomes and care as well as provide a roadmap for integrating genetic counseling into neurology settings.

As genetic testing becomes integrated into patient care guidelines and recommendations it expands the already established need for genetics professionals in health care. The primary objective of this study aims to assess the current utilization of genetic testing and genetic counseling within neurology settings. By analyzing survey responses from neurologists, this

research explores both use of genetic services in clinical practice as well as the role of genetic counselors in these processes. Additionally, the study examines the extent of collaboration between neurologists and genetics professionals in their current practices.

Methods

Study Design

This study utilized a cross-sectional survey design to assess the current utilization of genetic testing and genetic counseling within neurology settings in the state of Arizona.

Participants and Recruitment

Participants in this study were board-certified neurologists currently practicing in Arizona. To recruit participants, a recruitment email was sent to various neurology professional organizations and institutions within the state, requesting their assistance in either distributing the survey to their members or providing a list of licensed neurologists in Arizona. The email outlined the purpose of the study, the importance of their participation, and the relevance of the study to the field. In addition, publicly available email addresses were used to distribute the survey. Members of the research team, including a practicing neurologist, sent the attached email to their professional contacts. Finally, a snowball approach was also utilized by asking individuals who received our recruitment email to also distribute this survey to their contacts, promoting additional recruitment.

Survey Instrument

A brief online survey was developed in Qualtrics to gather data on neurologists' current use of genetic testing and genetic counseling (Qualtrics, Provo, UT, 2024). The survey was designed to capture quantitative data, focusing on the frequency of genetic testing ordered, familiarity with genetic counseling services, confidence in interpreting genetic test results, and possible barriers to integrating genetic counseling into neurology practices.

The survey questions were influenced by existing literature and guidelines, particularly those from the National Society of Genetic Counseling (NSGC) and the American Epilepsy Society, which recommend genetic testing for unexplained epilepsy and comprehensive counseling for conditions such as Amyotrophic Lateral Sclerosis (ALS). These guidelines recommend that genetic testing include pre- and post-test counseling by qualified providers, ideally genetic counselors, and emphasize informed consent and psychosocial support throughout the entire testing process (Smith, 2023). Additionally, these trained professionals should provide counseling to address the many possible psychological aspects of testing to ensure proper patient guidance (Chiò, 2014).

Data Collection

The survey was distributed electronically through an email which contained a link to a secure online platform (Qualtrics) which ensured the anonymity of respondents. The survey was available for completion from December 9th, 2024 to January 13th, 2025, with reminders sent to at the 2 week and 4 week marks. The complete survey is available in the supplementary material.

Data Analysis

Quantitative data from the survey were analyzed using descriptive statistics in Excel to summarize the frequency and percentage distributions of responses. For more in depth analysis, the data was analyzed in R Studio (version 2023.12.1+402) and Pearson correlation coefficients were calculated. Likert scale responses were converted into numerical variables in order to examine associations between responses. In order to calculate correlations with respondents' perception of the value of a genetic counselor performing genetic counseling roles and their own comfort performing genetic counseling roles, we calculated composite scores to summarize each

of these questions. For each of these questions, respondents indicated either their perceived value of a genetic counselor performing eight genetic counseling roles or their comfort performing these roles themselves on a four-point Likert scale. These composite scores were calculated by converting Likert scale responses into numerical variables and then summing the scores across all eight genetic counseling roles included in each question.

Ethical Considerations

The study protocol was reviewed and approved by the Institutional Review Board (IRB) at the University of Arizona (STUDY00004930). Participation in the survey was voluntary, and informed consent was obtained electronically before participants began the survey. All data was anonymized to protect the identity of the participants.

Results

Demographics

A total of 33 board-certified neurologists participated in the study. Of these 33, 28 completed at least 75% of the survey and were included in the analysis. Respondents represented a variety of subspecialties within neurology, including pediatric neurology (16%), epilepsy (14%), headache medicine (14%), and general neurology (12%), among others (Table 1). Participants varied in years of experience, with 11% having practiced for fewer than five years, 21% practicing for five to fifteen years, and 68% having over fifteen years of experience (Table 1). Most respondents indicated they practice in an urban (86%) or suburban setting (11%), with no respondents reporting a rural practice setting (Table 1).

Table 1: Demographics

		Frequency (%)
Neurology subspecialty	Pediatric neurology	8 (16%)
	Epilepsy	7 (14%)
	Headache medicine	7 (14%)
	General neurology	6 (12%)
	Neuromuscular medicine	6 (12%)
	Neurocritical care	3 (6%)
	Vascular neurology	3 (6%)
	Clinical neurophysiology	2 (4%)
	Movement disorders	2 (4%)
	Other	5 (10%)
Years in practice	<5 years	3 (11%)
	5-15 years	6 (21%)
	>15 years	19 (68%)
Practice setting	Urban	24 (86%)
	Suburban	3 (11%)
	Rural	0 (0%)
	unsure	1 (4%)
Level of Training in Genetics	Very good	4 (17%)
	Good	5 (21%)
	Acceptable	11 (46%)
	Poor	4 (17%)

Source of Genetics Training	Formal coursework during medical school or residency	19 (38%)
	Continuing education coursework after medical school or residency	19 (38%)
	Genetic testing labs	7 (14%)
	Other	5 (10%)

Note: When asked about their subspecialty and source of genetics training participants could select more than one answer.

Genetics Training and Experience

Neurologists in this study reported a range of genetics training experiences. When asked to self-report their perceived level of genetics knowledge, 17% said it was 'very good', 21% said 'good', 48% said 'acceptable', and 17% of participants said their knowledge level was 'poor' (Table 1). No participants reported that their genetics knowledge level was 'very poor'. The majority of participants received their training through formal coursework in medical school or residency (38%) or continuing education courses (38%). Others reported that their genetics training was provided by genetic testing laboratories (14%) (Table 1).

Genetic Testing Practices

When asked about the frequency of genetic test ordering in clinical practice, 32% of neurologists reported ordering genetic testing weekly, 25% did so every few weeks, 32% ordered genetic testing every few months, and 11% reported never ordering testing (Table 2). The most commonly ordered genetic tests were gene panel testing (31%), followed by exome sequencing (17%), chromosomal microarray analysis (15%), and whole genome sequencing (13%) (Table 2). Less commonly used tests included single gene testing (9%), karyotyping (8%), and single site testing (5%). Participants reported utilizing a variety of laboratories, including commercial sendout genetic testing companies (53%), institutionally contacted laboratories (34%), and

university laboratories (13%) (Table 2). When selecting a genetic test, neurologists relied on different decision-making strategies. Most participants chose a pre-curated panel based on both the patient diagnosis and family history (52%) and a smaller proportion chose a pre-curated panel based solely on patient diagnosis (40%) (Table 2).

Table 2: Genetic testing practices

Genetic Testing Demographic		**Frequency (%)**
How often do participants order genetic testing?	Weekly	9 (32%)
	Every few weeks	7 (25%)
	Every few months	9 (32%)
	Never	3 (11%)
What kinds of genetic testing do participants order?	Gene panel test	23 (31%)
	Exome sequencing	13 (17%)
	Microarray	11 (15%)
	Whole genome sequencing	10 (13%)
	Single gene test	7 (9%)
	Karyotype	6 (8%)
	Single variant test	4 (5%)
	Unsure	1 (1%)
What labs do you order testing through?	Commercial sendoff laboratories (Ambry, Invitae, GeneDX, Athena Diagnostics, Fulgent, Prevention Genetics etc.)	20 (53%)
	Institutionally contact laboratories (LapCorp, Sonora Quest, Mayo Clinic, ARUP, etc.)	13 (34%)
	University Laboratory	5 (8%)
How do you choose which genetic test to order?	I choose a pre-curated panel based on BOTH patient diagnosis and family history'	13 (52%)
	I choose a pre-curated panel based on patient diagnosis'	10 (40%)
	Other	2 (8%)

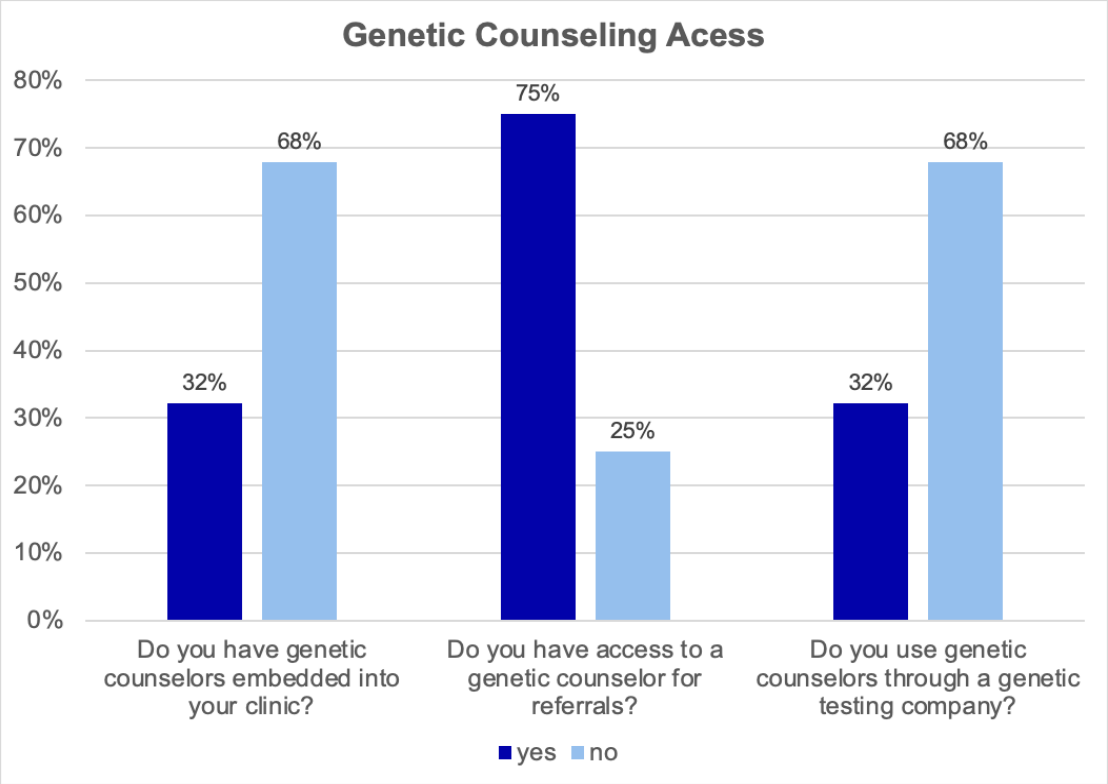
Note: For all questions except for how often they order genetic testing participants could select more than one answer.

When comparing a neurologist's level of genetics training to how frequently they ordered genetic testing, we found that neurologists with a higher level of genetics training were more likely to

order genetic tests regularly (ρ = .589, p = .002). This suggests that higher levels of genetics training may lead to greater incorporation of genetics into neurology practice.

The availability of genetic counseling services within neurology clinics was also assessed. Only 32% of participants reported having a genetic counselor embedded in their clinic, while 75% had access to a genetic counselor for referrals (Figure 1). Additionally, 32% of neurologists relied on genetic counselors provided through genetic testing companies (Figure 1). A notable 18% of participants reported that they did not have a genetic counselor embedded into their clinic, they did not have access to a genetic counselor for referrals, and that they also did not use genetic counselors through a testing company.

Figure 1. Access to Genetic Counselors in Neurology Clinics.



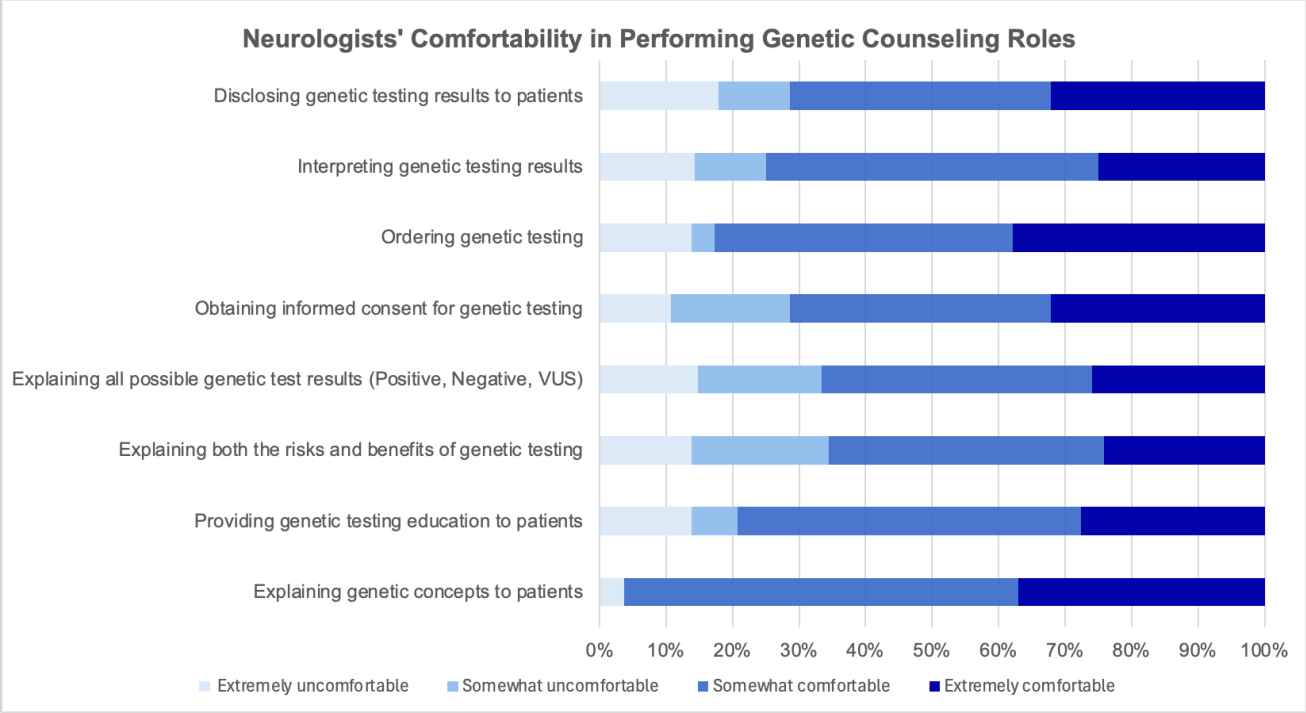
Neurologists' responses regarding their access to and use of genetic counselors in their current setting. The Y axis is the percentage of each response. The X axis displays the questions asked to neurologists regarding their access and use of genetic counselors. The dark blue bars indicate 'yes' responses, while the light blue bars indicate 'no' responses.

Genetic Counseling Integration

In our survey, we asked how comfortable practicing neurologists felt performing different genetic counseling roles in clinic. Overall, participants reported feeling most comfortable with explaining genetic concepts to patients, with 96% being at least somewhat comfortable with this role (Figure 2). We also found that 83% of surveyed neurologists reported they were at least somewhat comfortable with ordering genetic testing (Figure 2). However, participants expressed lower levels of comfort with some of the more detailed aspects of genetic counseling, including "explaining all possible genetic test results" where 34% of neurologists reported feeling at least

somewhat uncomfortable in this role (Figure 2). Similarly, 35% of participants reported some level of discomfort in explaining the risks and benefits of genetic testing (Figure 2). Interpreting and disclosing genetic test results followed similar patterns. While 50% of participants felt 'somewhat comfortable' with interpreting genetic test results, only 25% were 'extremely comfortable', and 25% reported discomfort (Figure 2). Comparably, 29% of neurologists reported feeling at least 'somewhat uncomfortable' with disclosing genetic test results to patients, despite 71% indicating some level of comfort (Figure 2).

Figure 2. Comfortability of Neurologists Performing Genetic Counseling Roles



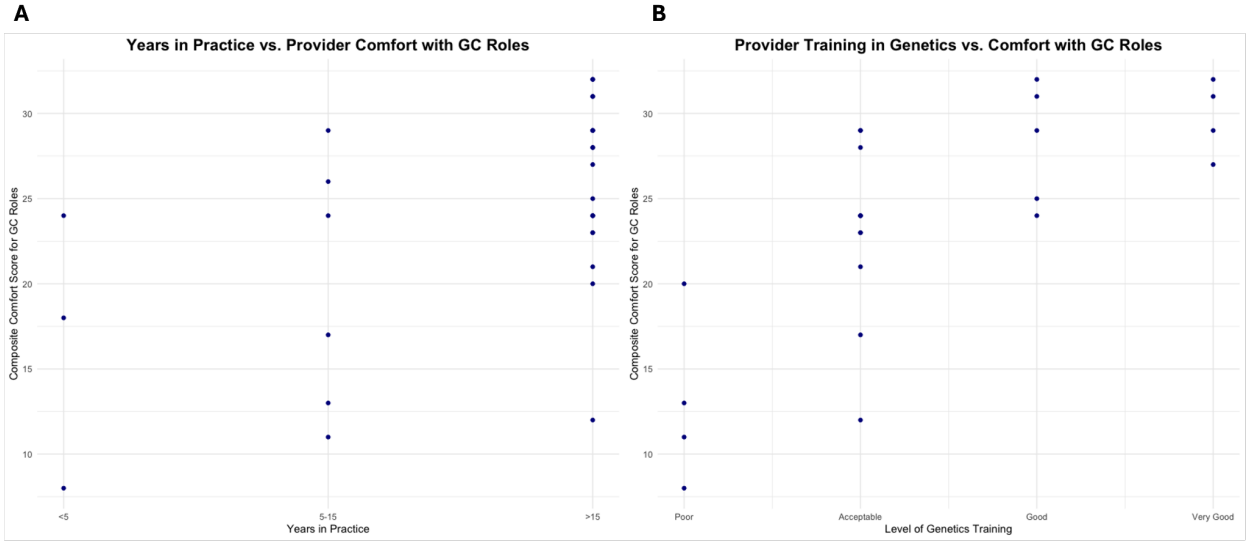
Neurologists responses regarding the level of comfort they have with performing eight different genetic counseling roles. The Y axis lists the various roles. The X axis shows the percentage of each response given. The bars are color-coded to reflect the percentage of responses in our sample, with darker colors used to show greater comfort performing the role.

We calculated a composite comfort score to represent neurologists' overall comfort level in performing genetic counseling roles. This score was derived by assigning numerical values to the Likert-scale survey responses, where respondents rated their comfort level performing each

genetic counseling role. The individual scores across each role were then summed to generate a composite measure of overall comfort. Our analysis of this composite score identified a significant positive correlation between neurologists' years of experience and their comfort performing genetic counseling roles ($\rho = 0.519$, p = 0.005) (Figure 3A). Neurologists with more years in practice reported greater comfort, while less experienced neurologists expressed a lower level of comfort performing the roles.

We also found a statistically significant positive correlation between the level of genetics training and neurologists' comfort in performing genetic counseling roles ($\rho = 0.739$, p = 3.66 x 10^{-5}) (Figure 3B). These findings suggest that the higher the genetics training level that participants reported, the more comfortable they are performing genetic counseling roles.

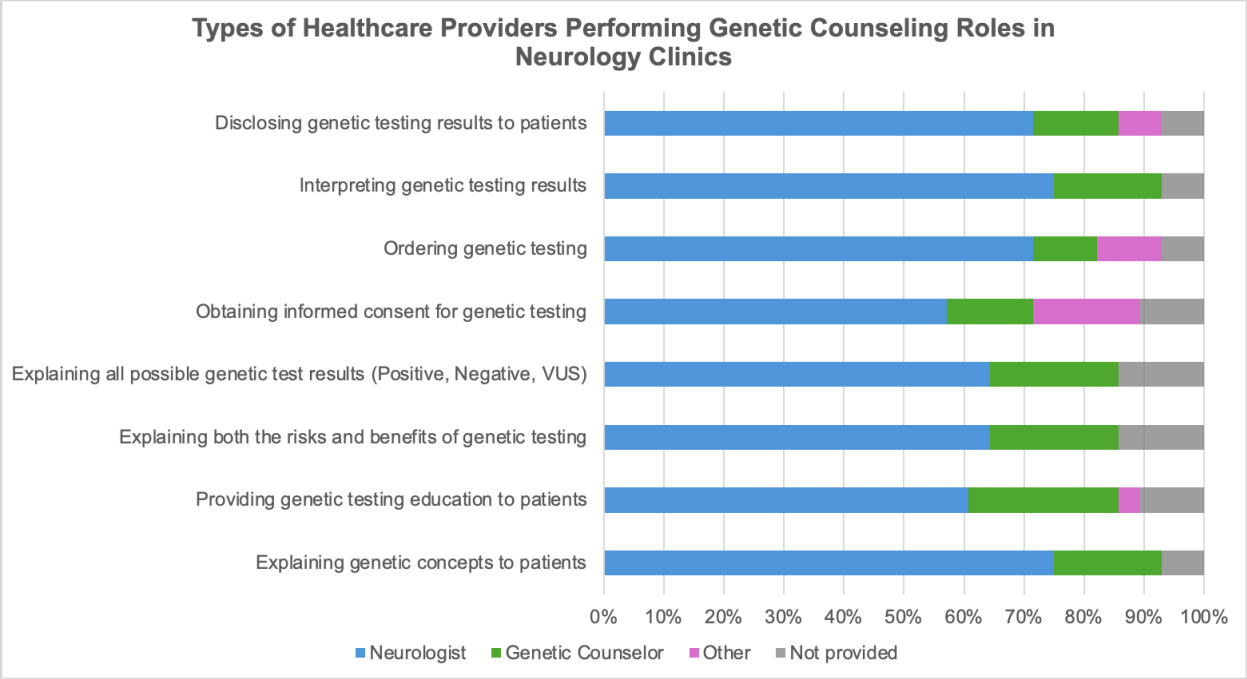
Figure 3: Provider Comfort with Genetic Counseling Roles Correlates with Years in Practice and Level of Genetics Training



Neurologists rated how comfortable they feel performing eight different genetic counseling roles using a likert scale from extremely uncomfortable to extremely comfortable. We generated a composite score by summing the responses for each respondent across all eight roles and found a significant correlation when compared to providers' years in practice (A; $\rho = 0.519$, p = 0.005) and their level of training in genetics (B; $\rho = 0.739$, p = 3.66 x 10^{-5}).

We also asked participants to describe which healthcare provider in their clinic performs each genetic counseling role. Our data indicates that neurologists are the primary providers of genetic counseling-related tasks in neurology clinical settings (Figure 4). Neurologists reported high involvement in all genetic counseling roles, with the highest percentages in 'explaining genetic concepts to patients' (75%), 'interpreting genetic test results' (75%), 'disclosing genetic test results to patients' (71%), and ordering genetic tests (71%) (Figure 4). Additionally, neurologists were also the predominant providers of risk-benefit discussions (64%), explanation of possible test results (64%), genetic test education (61%), and obtaining informed consent (57%) (Figure 4). The highest involvement of genetic counselors was reported in 'providing genetic testing education' (25%), 'explaining both the risks and benefits of testing' (21%) and 'interpreting genetic test results' (18%) (Figure 4). However, their participation in other areas, such as obtaining informed consent for testing (14%) and disclosing genetic testing results (14%), was low (Figure 4). Other healthcare providers (nurses, medical assistants, etc.) had minimal reported involvement in genetic counseling-related tasks, with the most common role of obtaining informed consent for genetic testing (18%) (Figure 4). Several respondents indicated that certain aspects of genetic counseling were not provided: 14% reported that 'explaining possible test results' is not addressed with patients, and 11% of neurologists indicated that informed consent for testing is not obtained (Figure 4). Of the 11% of participants who reported that informed consent was not obtained from patients, 66% claimed that they ordered genetic testing 'every few months' (Figure 4).

Figure 4. Performance of Genetic Counseling Roles in Neurology Clinics

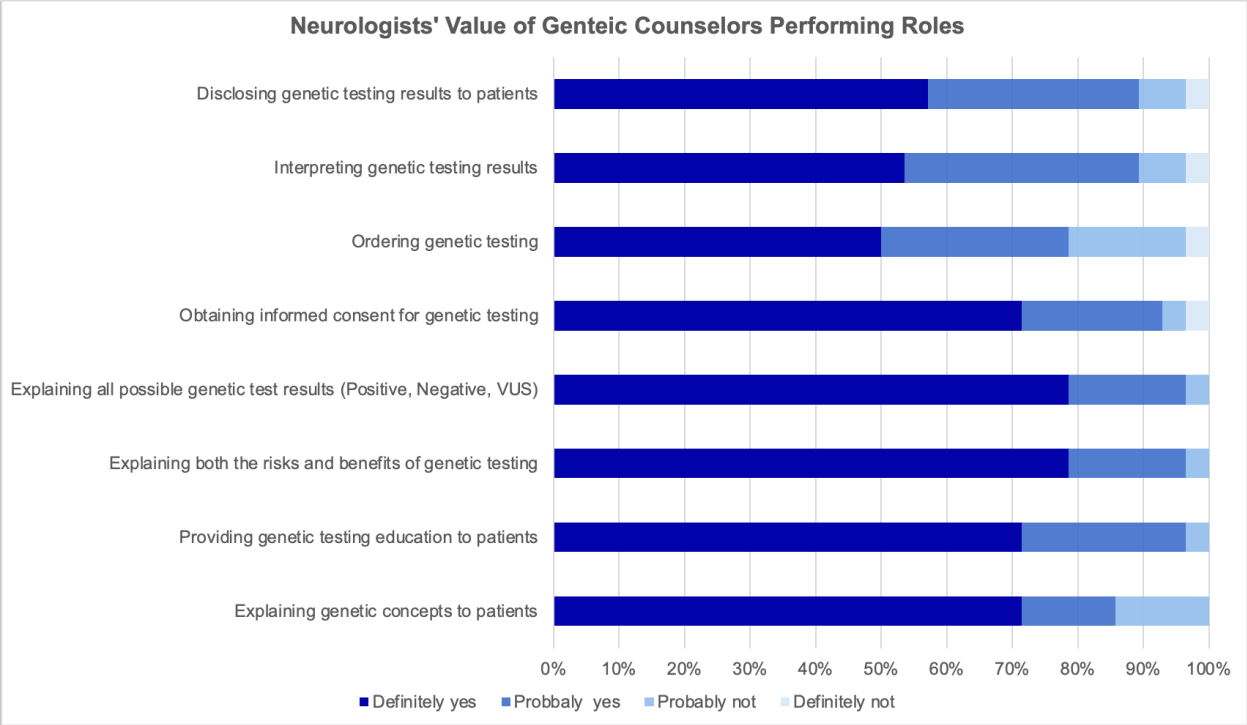


Neurologists were asked to describe which healthcare provider in their clinic typically performs eight different genetic counseling roles. The Y axis lists the various roles. The X axis shows the percentage responses for each role. The bars are color-coded to reflect the different categories of individuals who are typically performing the role.

Perceptions of Genetic Counselors in Neurology Settings

Participants were also asked about their perceptions of the value of genetic counselors performing traditional genetic counseling roles in clinics. The majority of neurologists assigned a high level of value for genetic counselors performing these roles. Over 90% of respondents indicated that they found value in genetic counselors providing genetic testing education, explaining the risks and benefits of testing, interpreting possible results (positive, negative, and VUS), and obtaining informed consent (Figure 5). Similarly, the majority of participants valued genetic counselors disclosing test results to patients (89%) (Figure 5). However, fewer neurologists viewed genetic counselors ordering genetic testing as valuable, with only 79% finding value (Figure 5).

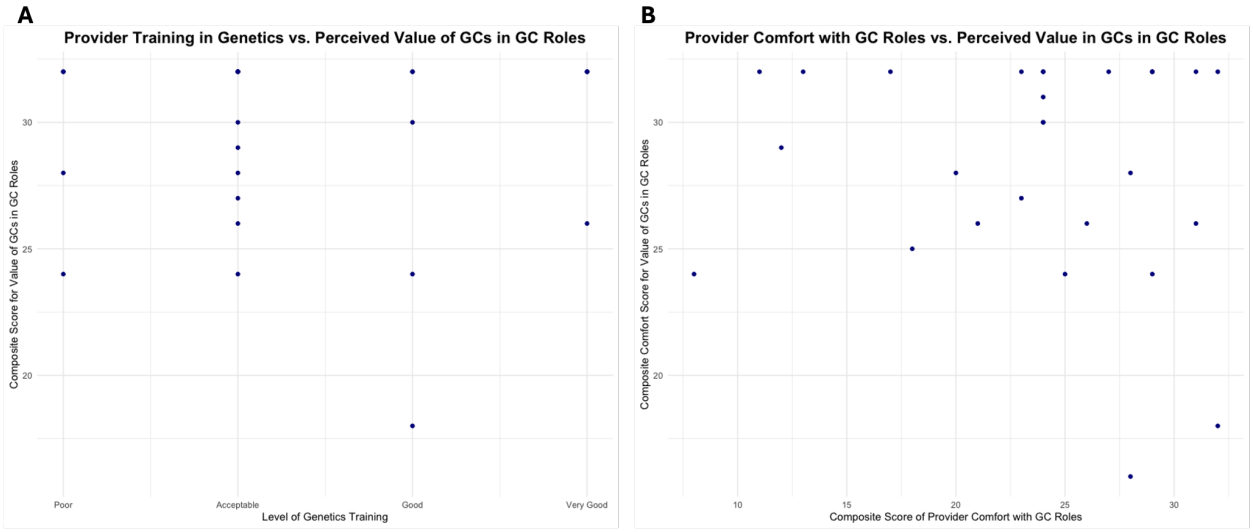
Figure 5: Neurologists' Perceptions of the Value of Genetic Counselors Performing Various Roles



Neurologists responses regarding if they find value in a genetic counselor performing eight different roles in a clinic. The Y axis lists the various roles. The X axis is the percentage of each response given. The bars are color-coded to reflect the level of value that was indicated with increasingly lighter color for less value found.

Interestingly, no statistically significant relationship was found between neurologists' perceived value of genetic counselors performing genetic counseling roles and their level of genetics training ($\rho = 0.027$, $p = 0.907$) (Figure 6A). There was also no significant correlation between neurologists' perceived value of genetic counselors performing genetic counseling roles and their comfort with performing genetic counseling roles in clinics themselves ($\rho = -0.1087077$, $p= 0.582$) (Figure 6B). These data suggest that neurologists recognize the value of genetic counselors, regardless of their own level of expertise in genetics, and that there is value to having a genetic counselor completing genetic counseling roles, regardless of how they feel about performing these roles themselves.

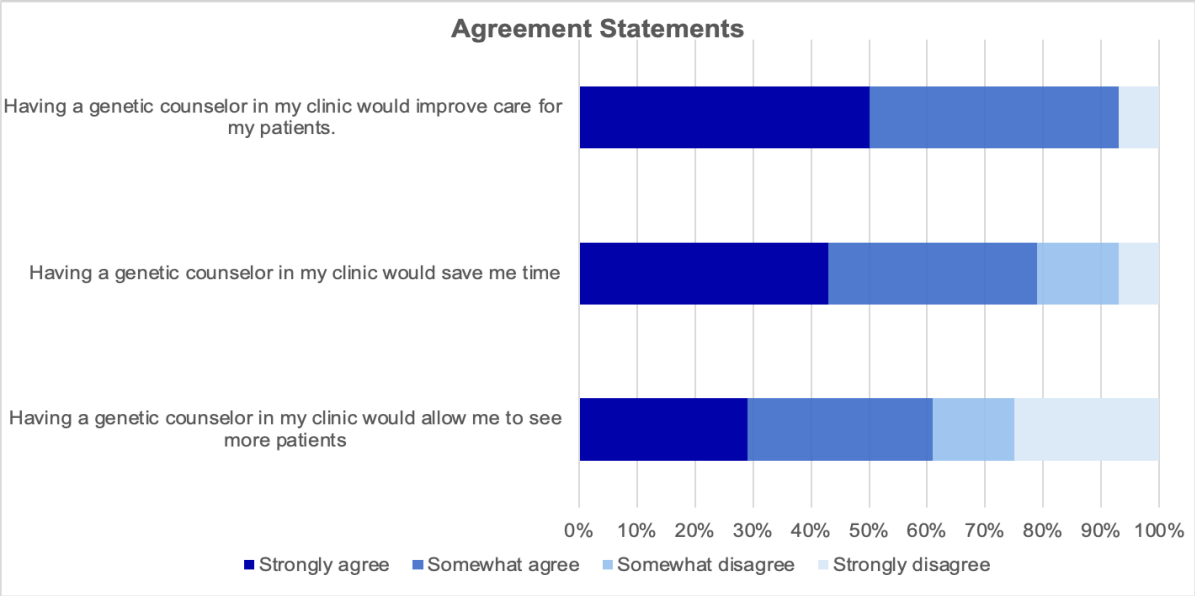
Figure 6: Providers' Perceived Value of genetic Counselors in Genetic Counseling Roles Correlates with Level of Genetics Training and Provider Comfort with Genetic Counseling Roles



Neurologists rated how much value they find in a genetic counselor performing eight different genetic counseling roles using a likert scale from definitely yes valuable to definitely not valuable. We generated a composite score by summing the responses for each respondent across all eight roles and found no significant correlations when compared to providers' level of training in genetics or their own comfort with the genetic counseling roles.

The survey also explored neurologists' perceptions of potential benefits of having a genetic counselor in their clinic. Findings indicated that 61% of neurologists agreed that a genetic counselor would allow them to see more patients (Figure 7). Most respondents (79%) felt that having a genetic counselor would save them time (Figure 7). Finally, nearly all respondents (93%) agreed that having a genetic counselor in their clinic would improve care (Figure 7). These findings suggest that neurologists recognize the potential value of incorporating genetic counselors into their practice, not only for their specialized genetic expertise, but also for their contribution to the overall efficiency and administrative functioning of the clinic.

Figure 7: Neurologists' Level of Agreement Regarding Statements About Having a Genetic Counselor in Clinic



The Y axis shows the statements that neurologists' were asked to indicate their level of agreement. The X axis is the percentage of each response given. The bars are color-coded to reflect the level of agreement that was indicated with increasingly lighter color for less agreement with each statement.

Neurologists' Involvement with and Value of Genetic Counselors in Clinical Roles

Given the high level of value of genetic counselors perceived by neurologists, we next wanted to assess whether the type of healthcare provider performing traditional genetic counseling roles in these clinics impacted the neurologists' comfort performing certain clinical roles or their perceived value of having a genetic counselor perform these traditional genetic counseling roles. To do this, we analyzed respondents' value in a genetic counselor performing genetic counseling roles and comfort performing these roles themselves stratified by who typically performs genetic counseling roles in these neurology clinics.

For several genetic counseling roles, neurologists acted as the primary providers while still expressing strong support for genetic counselors performing these roles in clinics. Notably,

among the 75% of participants who reported personally explaining genetic concepts to patients, 85% of them still valued a genetic counselor performing this role (Figure 8A). A similar trend was observed for genetic testing education, with all of the 61% of neurologists who provided this service themselves, also valuing a genetic counselor's involvement (Figure 8A). Likewise, roles involving test interpretation and results disclosure were primarily performed by neurologists, yet over 85% of those who provided these services themselves still saw value in having a genetic counselor involved (Figure 8A).

Interestingly, while most neurologists who obtained informed consent for genetic testing (85%) supported genetic counselor involvement (89%), fewer neurologists who ordered genetic testing (71%) saw value in a genetic counselor performing this task (75%) (Figure 8A). This suggests that neurologists may perceive test ordering aligning with their own scope of practice, although most still value a genetic counselor in this role.

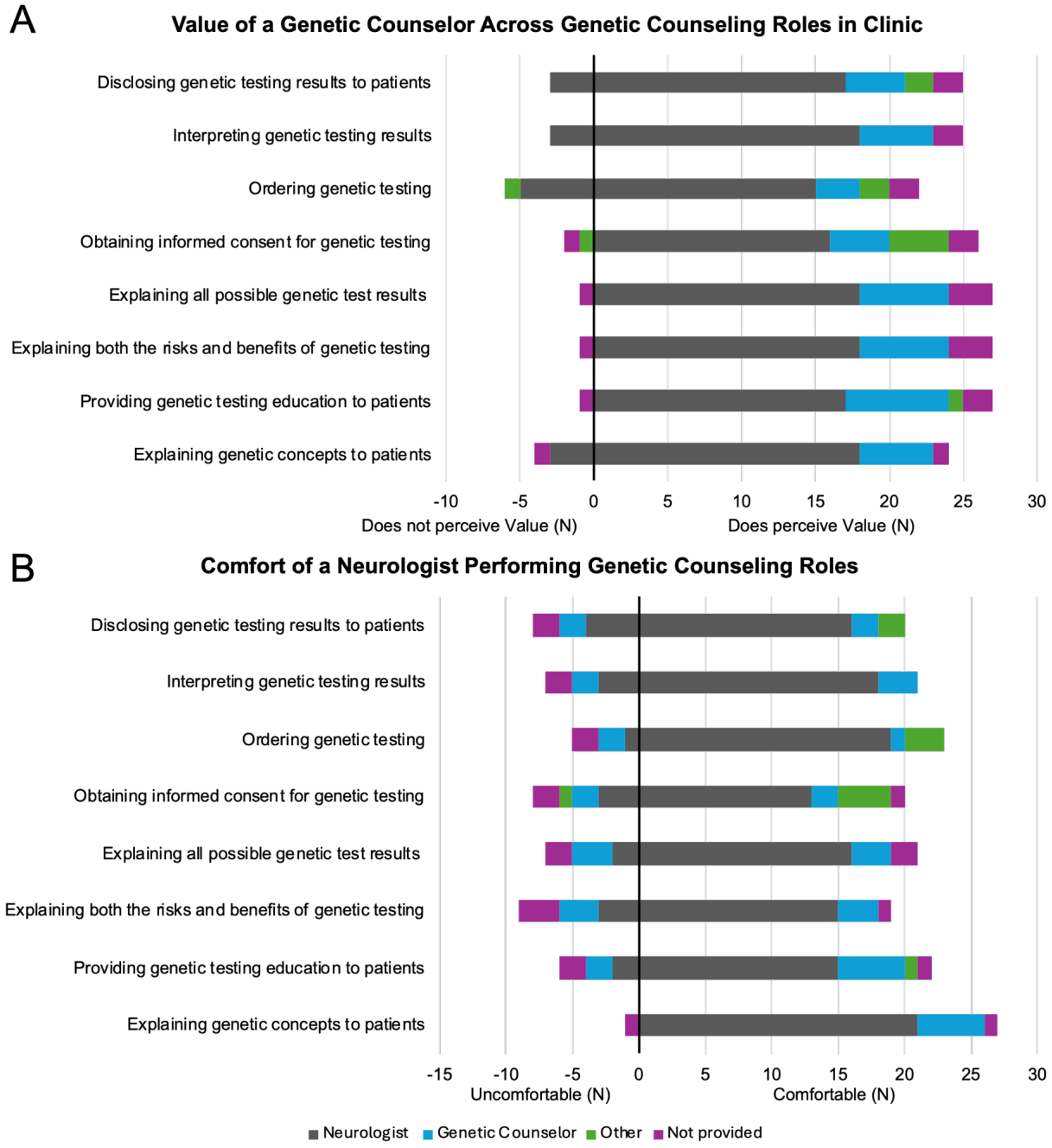
Of the respondents, three reported that they had a genetic counselor performing all eight genetic counseling roles on the survey. All of them said that they valued a genetic counselor performing all of these roles in a clinic reinforcing the perceived value of genetic counseling services by a genetic counselor in neurology settings. (Figure 8A).

Neurologists' Involvement with and Comfort with Genetic Counseling Roles

The survey results reveal a general alignment between neurologists' comfort with genetic counseling roles and their actual involvement in providing these services. However, there are notable patterns where neurologists provide the roles despite reporting some level of discomfort. Most neurologists (75%) reported that they are the ones explaining genetic concepts to patients, and all of them feel comfortable doing so (Figure 8B). The 71% of neurologists who actually did the role of ordering genetic testing also reported very high comfort levels with the task (95%)

(Figure 8B). However, when it comes to some of the intricacies of genetic testing, like those who provide genetic testing education (61%) or explain the risks and benefits of genetic testing (64%) in their clinic, some express discomfort (12% and 17%, respectively) (Figure 8B). Neurologists also commonly perform the roles of explaining genetic test results (64%) and obtaining informed consent (57%), but 11% and 19%, respectively, report discomfort performing these roles (Figure 8B). Notably, when genetic counselors were reported to do these tasks in clinics, neurologists were even more likely to report discomfort performing them themselves, suggesting an awareness of the potential complexities involved in these roles and the value of genetic counselors' specialized training in the area. This trend also applies to interpreting results and disclosing genetic test results with patients. Of the neurologists who reported performing the roles in clinic (75%, 71%), some still said they had discomfort with the tasks (14%, 20%) (Figure 8B).

Figure 8: Neurologists Report Value in a Genetic Counselor Performing Genetic Counseling Roles Despite Comfortably Performing Many Genetic Counseling Roles Themselves



A) Neurologists' responses to whether or not they value a GC performing GC roles stratified by who in their clinic typically performs these roles. Positive values on the x-axis indicate responses valuing a GC performing these roles. Negative values on the x-axis indicate responses that do not find value in a GC performing these roles. B) Neurologists' responses to whether or not they are comfortable performing GC roles themself stratified by who in their clinic typically performs these roles. Positive values on the x-axis indicate responses where the neurologist is comfortable

performing these roles. Negative values on the x-axis indicate responses where the neurologist is uncomfortable performing these roles.

Discussion

In this study, we explored the role of genetic testing and genetic counselors in neurology settings. Our results reveal several important trends regarding neurologists' involvement in genetic testing, genetic counseling, their comfort performing genetic counseling roles, and their acknowledgment of the value of genetic counselors.

Genetic Counseling Integration and Clinic Practices

Despite acknowledging the value of genetic counselors in neurology, the integration of these providers seems to be limited. Only 32% of respondents reported having a genetic counselor embedded in their clinic, while 75% had access to one through referrals. This suggests that while genetic counselors are available in some neurology clinics, their presence is not widespread. The limited access and integration of genetic counselors may contribute to neurologists taking on more specialized genetic counseling responsibilities. The data shows that neurologists frequently participate in genetic counseling roles in their clinics, even when they express some level of discomfort with some of the more complex roles. This highlights potential areas where genetic counselors could fill in gaps in patient care.

Neurologists' Comfort with Genetic Counseling Roles

Our findings revealed that while a majority of neurologists expressed comfort in explaining genetic concepts, this level of comfort did not necessarily align with their self-reported knowledge of genetics. This discrepancy between comfort and knowledge raises an interesting question: how do neurologists, who perceive their genetic knowledge as poor, still feel comfortable explaining genetic concepts to patients? One possibility is that neurologists may feel

confident in conveying basic information about genetics, even if they lack in-depth understanding of complex concepts or testing. This suggests that comfort with explaining genetic concepts may be more related to the familiarity of the topic or confidence in communicating general knowledge, rather than a deep technical understanding of genetics.

Additionally, while the vast majority of neurologists report feeling comfortable ordering genetic testing and explaining genetic concepts, the same level of comfort does not extend to skills pertaining to the complexities of genetic testing and informed consent. Higher levels of discomfort were reported in explaining both the risks and benefits of testing, all possible test results, and obtaining informed consent for genetic testing. These roles are integral to the genetic counseling and genetic testing processes so that patients can make autonomous decisions about their health (Ormond, 2021). They require careful communication to ensure that patients have a well-rounded understanding of the implications of genetic testing and the impacts it could have on themselves and their families. If patients are receiving inadequate counseling due to low knowledge of genetics or discomfort with communicating these concepts this could lead to misinterpretation or even harm.

Notably, 25% of participants were also uncomfortable interpreting genetic test results, and even more, 29%, felt uncomfortable in communicating those results to patients. This finding underscores a crucial gap in the genetic testing process, which includes the elements of test analysis, disclosure, and explanation (Faucett, 2019). It is logical that if neurologists were uncomfortable with interpreting the results themselves, they would also find discomfort in explaining them to their patients. This highlights a need for increased education and resources to support neurologists in genetic result interpretation and patient communication as well as the integration of genetic counselors into neurology clinics. Neurologists may struggle more with the

nuances of genetic testing that require specialized knowledge and skills that are typically more within the scope of genetic counselors. Overall, this suggests that while neurologists are confident in initiating genetic testing and providing broad, foundational explanations, the complexities of genetic counseling remain challenging. The gaps in comfortability highlight the importance of additional training for neurologists to help bridge the knowledge and communication divide, as well as greater collaboration with genetic counselors. To do so would enhance the quality of patient care, especially in complex cases where genetic testing results can have significant implications for diagnosis and treatment.

Perceived Value of Genetic Counselors

Despite their comfort in explaining genetic concepts, neurologists in this study consistently recognized the value of genetic counselors, particularly in tasks like interpreting test results, discussing risks and benefits, and obtaining informed consent. Over 90% of respondents acknowledged the value of having a genetic counselor take on these roles, reflecting a general consensus that genetic counselors' specialized training would improve patient care. This perception is aligned with prior research, which suggests that neurologists appreciate the expertise genetic counselors bring to genetic counseling, particularly in areas that go beyond basic genetic information (Purdy et al., 2022).

The survey data demonstrates a consistent trend: neurologists frequently provide genetic counseling services in their clinics, yet still perceive significant value in having a genetic counselor fulfill these roles. Importantly, even among neurologists who reported comfort with genetic counseling tasks, the majority still believed genetic counselors added value, particularly in areas requiring specialized expertise. This perceived value does not appear to be influenced by

neurologists' own comfort levels or training in genetics. Instead, their responses suggest a recognition of the distinct and complementary expertise that genetic counselors contribute to patient care. While neurologists can function as the default providers in the absence of genetic counselors, they recognize the added benefit of having a provider with specific training and expertise performing the roles. Overall, the responses highlight a clear need for greater collaboration between neurologists and genetic counselors to ensure that patients receive the most comprehensive and accurate genetic information possible.

Limitations

There are several limitations to this study. First, the sample size was relatively small, with only 28 neurologists participating. Additionally, the study relied on self-reported data, which may be subject to bias or inaccuracies. Future studies with larger, more diverse samples and objective measures of neurologists' comfort and knowledge could provide stronger insights into the integration of genetic counselors into neurology settings.

Conclusions

Our findings suggest that the integration of genetic counselors into neurology clinics could significantly benefit both neurologists and patients. Given the increasing role of genetic testing in neurology, it is crucial to ensure that neurologists have access to genetic counseling professionals who can help interpret results, discuss risks, benefits, and results of testing with patients, and provide comprehensive support in areas such as psychosocial counseling and informed decision making. This collaboration could help alleviate neurologists' discomfort with complex genetic counseling tasks, improve patient care, and enhance the overall integration of genetic testing into neurology practice.

Future research should explore the effectiveness of different models for integrating genetic counselors into neurology clinics, particularly looking at the impact on neurologists' confidence, patient satisfaction, and clinical outcomes. Additionally, investigating barriers to genetic counselor integration—such as funding, logistical challenges, and lack of awareness—could help identify strategies to improve the availability and accessibility of genetic counseling services in neurology.

Supplemental 1.

Neurologist Questionnaire

Start of Block

Q1 • Are you a licensed neurologist in the state of Arizona?

- ○ *Yes (1)*
- ○ *No (2)*

Skip To: End of Survey If • Are you a licensed neurologist in the state of Arizona? = No

Q2 Which subspecialty of neurology do you practice? (select all that apply)

- ☐ *Autonomic Disorders (1)*
- ☐ *Behavioral Neurology (2)*
- ☐ *Clinical Neurophysiology (3)*
- ☐ *Epilepsy (4)*
- ☐ *General Neurology (5)*
- ☐ *Geriatric Neurology (6)*
- ☐ *Headache Medicine (7)*

☐ *Neural Repair and Rehabilitation (8)*

☐ *Neurocritical Care (9)*

☐ *Neuro-Oncology (10)*

☐ *Neuromuscular Medicine (11)*

☐ *Pediatric Neurology (12)*

☐ *Vascular Neurology (13)*

☐ *other (14)* _____

Q3 How long have you been practicing Neurology?

○ *<5 years (1)*

○ *5-15 years (2)*

○ *>15 years (3)*

Q4 Do you practice in an urban, suburban, or rural setting?

○ *Urban (1)*

○ *Suburban (2)*

○ *Rural (3)*

○ *Unsure (4)*

End of Block: Default Question Block

Start of Block: Block 1

Q5 How would you describe your current level of genetics training?

	Very Poor (1)	*Poor (2)*	*Acceptable (3)*	*Good (4)*	*Very Good (5)*
Genetics training level (1)	○	○	○	○	○

Q6 How did you receive your genetics training? (select all that apply)

☐ *Formal coursework during medical school or residency (1)*

☐ *Continuing education coursework after medical school or residency (2)*

☐ *Genetic testing labs (3)*

☐ *Other, please describe (4)*

_____

Q7 How often do you order genetics testing?

	Never (1)	*Every few months (2)*	*Every few weeks (3)*	*Weekly (4)*
Ordering genetic tests (1)	○	○	○	○

Skip To: Q10 If How often do you order genetics testing? = Ordering genetic tests [Never]

Q8 What kinds of genetic testing do you order?

- ☐ *Single Variant Test (1)*
- ☐ *Single Gene Test (2)*
- ☐ *Gene Panel Test (3)*
- ☐ *Exome Sequencing (4)*
- ☐ *Whole Genome Sequencing (5)*
- ☐ *Karyotype (6)*
- ☐ *Microarray (7)*
- ☐ *Unsure (8)*

Q9 What labs do you order testing through? (select all that apply)

☐ *Commercial sendoff laboratories (Ambry, Invitae, GeneDX, Athena Diagnostics, Fulgent, Prevention Genetics, etc.) (1)*

☐ *Institutionally contact laboratories (LapCorp, Sonora Quest, Mayo Clinic, ARUP, etc.) (2)*

☐ *Direct to consumer (23andMe, Color Genomics, ect.) (3)*

☐ *University Laboratory (4)*

☐ *Unsure (5)*

Q10 How do you choose which genetic test to order?

○ *I choose a pre-curated panel based on patient diagnosis (1)*

○ *I choose a pre-curated panel based on patient family history (2)*

○ *I choose a pre-curated panel based on BOTH patient diagnosis and family history (3)*

○ *I custom design a panel (4)*

○ *I contact the genetic testing lab (5)*

○ *Other, please describe (6)* _____

Q11 How comfortable are you with these parts of genetic counseling and testing?

	Extremely uncomfortable (1)	*Somewhat uncomfortable (2)*	*Somewhat comfortable (3)*	*Extremely comfortable (4)*
Explaining genetic concepts to patients (1)	○	○	○	○
Providing genetic testing education to patients (2)	○	○	○	○
Explaining both the risks and benefits of genetic testing (3)	○	○	○	○
Explaining all possible genetic test results (Positive, Negative, VUS) (4)	○	○	○	○
Obtaining informed consent for genetic testing (5)	○	○	○	○
Ordering genetic testing (6)	○	○	○	○

Interpreting genetic testing results (7)	○	○	○	○
Disclosing genetic testing results to patients (8)	○	○	○	○

End of Block: Block 1

Start of Block: Block 2

Q12 Who in your clinic typically provides these aspects of genetic counseling?

	You (1)	*Nurse (2)*	*Genetic Counselor (3)*	*Medical Assistant (4)*	*Other (5)*	*Not provided (6)*
Explaining genetic concepts to patients (1)	○	○	○	○	○	○
Providing genetic testing education to patients (2)	○	○	○	○	○	○
Explaining both the risks and benefits of genetic testing (3)	○	○	○	○	○	○

Explaining all possible genetic test results (Positive, Negative, VUS) (4)	○	○	○	○	○	○
Obtaining informed consent for genetic testing (5)	○	○	○	○	○	○
Ordering genetic testing (6)	○	○	○	○	○	○
Interpreting genetic testing results (7)	○	○	○	○	○	○
Disclosing genetic testing results to patients (8)	○	○	○	○	○	○

Q13 Do you have genetic counselors embedded into your clinic?

○ *Yes (1)*

○ *No (2)*

Q14 Do you have access to a genetic counselor for referrals?

○ *Yes (1)*

○ *No (2)*

Q15 Do you use genetic counselors through a genetic testing company?

○ *Yes (1)*

○ *No (2)*

Q16 For each of the statements below, indicate your agreement using the scale provided.

	Strongly Disagree (1)	*Somewhat disagree (2)*	*Somewhat agree (3)*	*Strongly agree (4)*
Having a genetic counselor in my clinic would allow me to see more patients. (1)	○	○	○	○
Having a genetic counselor in my clinic would save me time. (2)	○	○	○	○

Having a genetic counselor in my clinic would improve care for my patients. (3)	○	○	○	○

Q17 Do you find value in a genetic counselor performing the following roles?

	Definitely not (1)	*Probably not (2)*	*Probably yes (3)*	*Definitely yes (4)*
Explaining genetic concepts to patients (1)	○	○	○	○
Providing genetic testing education to patients (2)	○	○	○	○
Explaining both the risks and benefits of genetic testing (3)	○	○	○	○
Explaining all possible genetic test results (Positive, Negative, VUS) (4)	○	○	○	○
Obtaining informed consent for	○	○	○	○

genetic testing (5)				
Ordering genetic testing (6)	○	○	○	○
Interpreting genetic testing results (7)	○	○	○	○
Disclosing genetic testing results to patients (8)	○	○	○	○

End of Block: Block 2

Supplemental 2.

Participant consent form found at the start of the survey

"You are invited to participate in a research study. Your participation in this research study is voluntary, and you do not have to participate. This document contains important information about this study and what to expect if you decide to participate. Please consider the information carefully. Feel free to ask questions before making your decision on whether or not to participate.

Study Purposes and Procedures: This study aims to collect information about the current utilization of genetic testing and genetic counseling within neurology settings in the state of Arizona. The secondary objective of this research is to identify any existing gaps between recommended guidelines of genetic testing/counseling and clinical practice that may impact patient care. By shedding light on the practical application of genetic testing and genetic counseling in neurology, we aim to demonstrate the importance of genetic counselors within neurology settings.

The survey is administered online through a program called Qualtrics and should take 15 minutes of your time to complete.

Once you have submitted the survey, your responses cannot be withdrawn due to the anonymous nature of the survey. Up to the point of submitting, you have the right to withdraw by exiting the survey. Surveys which are 75% complete or more will be included in data analysis.

This study will be written up as part of the PI's master's thesis and will be freely available via the University of Arizona's thesis repository.

Risks and Benefits: Your participation may benefit you individually as you are able to share your personal experiences and allow your voice to be heard. Your participation may benefit society in raising awareness of these issues. There is also a risk that your confidentiality may be breached through participation in this study.

Confidentiality: The information that you give in the study will be anonymous. Your name will not be collected for research purposes or linked to your answers. Because of the nature of the data, it may be possible to deduce your identity; however, there will be no attempt to do so and your data will be reported in a way that will not identify you. Identifiable research data will be encrypted and password protected. Information collected about you will not be used or shared for future research studies.

The information that you provide in the study will be handled confidentially. However, there may be circumstances where this information must be released or shared as required by law. The University of Arizona Institutional Review Board may review the research records for monitoring purposes.

Contact: For questions, concerns, or complaints about the study you may contact the study Principal Investigator, Maddie Gorman, at mmgorman2@arizona.edu or the research advisor, Valerie Schaibley, at vschaibley@arizona.edu.

For questions about your rights as a participant in this study, or to discuss other study-related concerns or complaints with someone who is not part of the research team, you may contact the Human Subjects Protection Program Director at 520-626-8630 or online at https://research.arizona.edu/compliance/human-subjects-protection-program. By clicking Next, you are allowing your responses to be used for research purposes."

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