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AN ASSESSMENT OF KNOWLEDGE AND ATTITUDES OF GENETIC COUNSELING SERVICES IN U.S. HTCs

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Hemophilia is a hereditary bleeding disorder which requires lifelong specialized care. A network of Hemophilia Treatment Centers (HTCs) exists to meet the medical needs of patients affected by hemophilia. Genetic counseling services are an integral part of the HTC model of care; however, many HTCs do not have genetic counselors on staff. As a result, the duty to provide these services must fall to other healthcare providers within the HTC. To assess the knowledge and attitudes of these providers we developed a 49 question survey that was distributed electronically to hematologists and nurses at U.S. HTCs. The survey consisted of a three sections: demographic information, knowledge of hemophilia genetics, and attitudes towards genetic services. A total of 111 complete responses were received and analyzed. The average knowledge score among all participants was 74.8% with a total of 81 participants receiving a passing score of 70% or above. Thirty participants scored below 70% in the knowledge section. In general, attitude scores were high indicating that the majority of hematologists and nurses in HTCs feel confident in their ability to provide genetic counseling services. Over 90% of participants reported that they have some form of access to genetic counseling services at their center.

Hematologists and nurses practicing in U.S. HTCs demonstrate sufficient knowledge of the genetics of hemophilia, and they generally feel confident in their ability to provide genetic counseling services to their patients. While their knowledge is sufficient, the average knowledge score was lower than 75%. Certain questions covering new genetic technologies and testing practices were more commonly missed than questions asking about more basic aspects of hemophilia genetics, such as inheritance and carrier testing. Finally, many clinics report having access to a counselor, but it is oftentimes a hematologist or nurse who is providing genetic counseling services to patients. Given the inconsistency in knowledge among providers coupled with the high confidence in one's ability to counsel patients, it leaves room to question whether information about the genetics of hemophilia is being communicated to patients in the most appropriate and accurate manner.

TABLE OF CONTENTS

LIST OF FIGURES

LIST OF TABLES

ABBREVIATIONS

CDC = Center for Disease Control and Prevention

CPHS = Committee for the Protection of Human Subjects

HRSA = Health Resources and Services Administration

HTC = Hemophilia Treatment Center

PGD = Preimplantation genetic diagnosis

Background

Hemophilia is a rare hereditary bleeding disorder characterized by a deficiency of either coagulation factor VIII or IX with bleeding symptoms that range in severity from mild to severe (1). Complications associated with hemophilia include hemarthrosis, hematomas of the soft tissues, intracranial hemorrhages, prolonged bleeding, poor wound healing, prolonged oozing following minor injuries, epistaxis, and ecchymosis (2). Due to the chronic nature and specialized treatment of the disorder, the Health Resources and Services Administration (HRSA) began funding a network of Hemophilia Treatment Centers (HTCs) in 1975 which employ a multidisciplinary approach to the management of hemophilia (3).

There are over 140 registered HTCs across the United States (4) and each center is comprised of a core staff that includes a medical director, nurse coordinator, psychosocial professional, and physical therapist. Some centers have additional providers on staff such as genetic counselors, orthopedists, and dentists (3). The goals of HTCs are outlined by both the HRSA include: the provision of comprehensive care to patients and families affected by hereditary bleeding disorders, such as hemophilia; the provision of services to women, minorities, adolescents, the uninsured/underinsured, and people living in geographically underserved regions; the provision of outreach and education services; education emphasizing the importance of prevention of bleeding episodes; collaboration with other healthcare entities; and connection of patients to primary care physicians. The Centers for Disease Control and Prevention (CDC) performs surveillances of outcomes and monitors blood safety in patients with hemophilia seen at HTCs.

HTCs rely on a multidisciplinary approach to patient management which incorporates diagnosis, treatment, prevention and family education into its model of care. HTCs provide healthcare services to 70-80% of individuals with hemophilia in the United States; and, in 2004, 27,662 patients received services from HTCs in the United States, 15,224 of whom had hemophilia (5).

Genetic counseling services are an essential component of the multidisciplinary care for patients with hemophilia. Genetic counseling services at HTCs mainly consist of genetic education about the etiology, inheritance, recurrence risk, carrier screening, and genetic testing of hemophilia. Among the 142 registered HTCs, the CDC reports that only 12 (8.5%) centers have a genetic counselor on staff (4). Without a genetic counselor, the responsibility of providing genetic education to patients would likely fall to a physician, nurse, or social worker at the center.

While several studies have assessed the knowledge and attitudes of healthcare providers toward genetic services, (6-11) there are no studies that have measured the genetics knowledge

of physicians and nurses in HTCs. The objective of this study is to determine the level of genetics knowledge among physicians and nurses in HTCs, assess how genetic education is being provided to patients, and identify the attitudes towards and barriers for patients surrounding genetic counseling services in HTCs.

Materials and Methods

We developed a questionnaire, with permission, that was modeled after a validated tool originally created by Hofman *et al.* and used to assess the knowledge of genetics and genetic testing among family medicine physicians practicing in the United States (7). The questionnaire used in this study was estimated to take 15-20 minutes to complete and was comprised of 49 questions divided into three sections: 1) demographic information (14 questions), 2) knowledge of hemophilia genetics (14 questions), and 3) attitudes towards genetic services in an HTC (21 questions). The knowledge section consisted of multiple choice and true/false questions covering subjects such as heredity, genetic testing, prenatal/reproductive issues, and carrier screening as they apply specifically to hemophilia A. In addition, an optional free response section was provided at the end of the questionnaire for participants who wished to include other comments. The questionnaire was generated using the web-based REDCap software (12), and it was only available in electronic format. The questionnaire and study design were approved by the Committee for the Protection of Human Subjects (CPHS) at the University of Texas Health Science Center in Houston, Texas prior to data collection.

Requests for survey participation were sent by email to hematologists and nurses in U.S. HTCs. Email addresses for potential participants were obtained from the online HTC Directory maintained by the CDC. In total, 570 email addresses were obtained from the directory. Only email addresses for individuals listed as pediatric and/or adult hematologists, nurses, and nurse practitioners in the directory were used. Of the 570 email addresses, 45 were returned as undeliverable. In addition, 9 healthcare providers requested to be excluded from the study. As a result, the total number of invited participants was 516.

Data collection occurred between September 2012 and February 2013. All responses were submitted anonymously, and there were no incentives offered to survey participants. Complete survey responses were compiled and analyzed in STATA statistical software (version 10, College Station, TX). Following completion of the questionnaire, individual participants were given a knowledge score based on their responses in the knowledge section. Scores were generated by calculating the number of correct responses divided by the total number of questions. In the attitudes section, participants ranked their response using a 5 point Likert scale with 1 representing those who strongly disagree and 5 representing those who strongly

agree. For the purposes of analysis Likert scale responses were grouped together into three categories: 1-2, 3, and 4-5 which correspond to the thematic categories: 1)Disagree 2)Agree 3)Strongly agree. Similar categorization was done for the second part of the attitude section where Likert responses were grouped together into three categories: 1-2, 3, and 4-5 which correspond to the thematic categories: 1)Not confident 2)Neither confident nor unconfident 3)Confident. Comparisons between demographic features and knowledge and attitudes were made using chi-square and one-sided t-tests. A cut-off p-value of 0.05 or less was used to determine statistical significance. Overall demographic characteristics and study findings that were statistically significant are reported.

Results

A total of 117 complete responses and 27 partial responses were received for an overall response rate of 22.7%. This is comparable to response rates in similar studies (9,11). Partial responses were excluded from the analysis. Of the 117 complete responses, 4 were from genetic counselors and 2 were from other allied healthcare professionals. These responses were analyzed separately since they were not part of the targeted population. Of the remaining 111 complete responses, 51 (46.0%) were from hematologists and 60 (54.1%) were from nurses. Overall, there were 27 male participants (25.5%) and 79 female participants (74.5%) with an average age of 60.0 years among the total cohort. Other demographic characteristics are summarized in Table 1.

A total knowledge score was generated for each participant by dividing the correct number of responses by the total number of questions with the highest possible score being 14 out of 14. The average score among all participants was 10.5 out of 14 (74.8%). A minimum of 10 correct responses was required to achieve a passing score. All questions were weighted equally. A total of 81 participants (73.0%) passed the knowledge section with a score of 70% or higher and 30 participants (27.0%) scored below 70% in the knowledge section. Thirty-eight out of 51 (74.5%) physicians and 43 out of 61 (71.7%) nurses achieved passing scores of 10 out of 14 (71.4%) or more. The difference in pass rates between physicians and nurses (see Table 2) was not statistically significant ($p = 0.737$).

While the overall scores were not significantly different between physicians and nurses there were three questions for which the correct response rate was significantly different between the two groups. Question 1, which asked about the detection rate of *F8* genetic testing in males with severe hemophilia, and question 11, which asked about the best method for determining whether or not a female is a carrier of hemophilia, were more frequently missed by

nurses than physicians. Alternatively, question 10, which asked about the risk of recurrence for a family with a simplex case, was more frequently missed by the physician group (Table 3).

Although knowledge scores were favorable overall, some topics were commonly missed in this section by both physicians and nurses. Specifically, three questions were consistently answered incorrectly by over 50% of the study population: questions 4, 6, and 14 (see Table 4). These questions covered topics including: preimplantation genetic diagnosis (PGD), inheritance, and genetic testing.

The attitudes section consisted of two parts. In the first part participants were asked about their attitudes regarding genetic counseling services in clinical practice. The statements in this section covered subjects including the provider's perceived value of genetic counseling services, the provider's comfort level with respect to certain aspects of genetic counseling services, and referral practices. The breakdown of questions is summarized in Table 5. Question 4, which focused on provider referral practices, indicated that physicians (60.8%) were much more likely to refer patients for genetic counseling compared with nurses (36.7%, $p = 0.027$).

The second part of the attitudes section asked participants how confident they felt about providing individual aspects of genetic counseling services. Specifically, these questions included details regarding genetic testing, psychosocial counseling, and insurance issues. Participants ranked how confident they felt in their ability to provide information about these topics using a 5 point Likert scale where 1 was not at all confident and 5 was very confident. The results from this section are summarized in Table 6. Only question 13 showed a statistically significant difference in responses between physicians and nurses. Responses to this question demonstrated that physicians feel more confident than nurses in their ability to discuss insurance issues that may arise in the context of genetic testing ($p = 0.026$).

Finally, a comparison of knowledge and attitude questions which covered the same themes was performed. There were three major themes present in both the knowledge and attitudes sections: inheritance, genetic testing, and carrier testing. Specifically, among providers that incorrectly answered question 6, which asked about the risk of recurrence for a carrier female, 79.0% agreed with the statement, "I feel comfortable educating a patient about X-linked inheritance" ($p = 0.393$). Also, 57.6% of providers who incorrectly answered question 14, which asked about the best individual to offer genetic testing, agreed with the statement, "I would feel comfortable explaining the benefits and limitations of genetic testing for hemophilia" ($p = 0.052$). In addition, 72.7% of providers who incorrectly answered question 14 felt confident in their ability to help a patient decide whether to be tested ($p = 0.748$).

Lastly, our study assessed the type of access HTCs have to genetic counseling services. The majority of participants (91.6%) reported that they have some form of access to genetic

counseling services at their center. A follow-up question gave participants the ability to specify what type of access they have (see Table 7). The most common form of access reported was having a genetic counselor that was available as needed (34.7%). Another 24.5% reported having a genetic counselor that attends every clinic. Still, another 28.6% of participants reported having other types of access. Participants who selected other were asked to specify their access. Responses were categorized and are presented in Box 1. The most common form of access specified by participants was a referral to a separate clinic outside of the HTC (84.4%). Many of these clinics are within the same institution as the HTC, but a separate appointment must be made. Also of note, the majority of providers (n = 83, 75.5%) agreed that their patients would benefit from meeting with a genetic counselor as part of their care.

Box 1 Other forms of access to genetic counseling services

By referral within institution (75.0%)

By referral outside of institution (9.4%)

Services provided by other healthcare providers, not genetic counselors (9.4%)

Limited access/vacancies (6.3%)

Discussion

This study provides a glimpse into the level of knowledge and the attitudes of providers in HTCs across the United States. The majority of providers have knowledge scores that demonstrate an above average level of understanding of the genetics of hemophilia. In addition, the majority of providers report that they feel comfortable providing basic genetic counseling services to patients treated at HTCs. However, newer genetic technologies, such as preimplantation genetic diagnosis (PGD), and issues of insurance discrimination are areas in which providers not specifically trained in genetic counseling feel less comfortable.

Knowledge

When asked about PGD, the majority of physicians and nurses (59.5%) incorrectly believed that PGD can be useful even if the familial gene mutation is not known. By definition, PGD is used to detect the presence or absence of a mutated gene that is known to

cause a genetic disease in a single cell isolated from an embryo (13). Without having the known familial mutation it is not possible to diagnose an embryo prior to implantation. That being said, PGD is a specialized service that is not considered standard of care for patients with hemophilia. As a result, it is reasonable to say that complex issues in genetic medicine, such as PGD, lie outside the scope of practice of hematologists and nurses in HTCs; and, therefore, the discussion of such issues with patients fall to other specially trained healthcare providers. Of note, 3 out of the 4 genetic counselors that completed the survey answered this question correctly. Out of all of the healthcare providers in HTCs it would seem most likely that patients wishing to discuss PGD further should be referred to a genetic counselor when available.

Another question that was commonly answered incorrectly asked about the chance that, in any given pregnancy, a female carrier would have a male with hemophilia. The majority (55.9%) of participants answered incorrectly with the majority selecting 1 in 2 live births as their answer. The correct response is 1 in 4 live births which takes into account both the chance for a male baby that also inherits the disease-causing hemophilia mutation. It is possible that responders made the assumption that the affected fetus is male, and thus selected the answer choice that corresponded to the 50-50 chance in each pregnancy of a carrier female giving birth to a male with hemophilia. It is likely that this is the case since the majority of participants answered the other questions concerning the heredity of hemophilia correctly. By comparison, though, all four of the genetic counselors that responded answered this question correctly. While there is most likely not a deficiency in provider knowledge in regards to the inheritance of hemophilia, it is important for providers to accurately communicate the risk of recurrence of hemophilia to carrier females.

Lastly, the question which was answered incorrectly most often (61.3%) was question 14 in the knowledge section. This question asked which family member would be the best person to offer genetic testing. It is generally agreed that, whenever possible, genetic testing should only be offered if the results of the test can be adequately interpreted. For that reason it is best to begin testing in an affected individual (or index case) before proceeding to test seemingly unaffected family members in order to eliminate the possibility of receiving an uninformative negative test result (14,15). In our study, the majority of physicians (52.9%) and nurses (50.9%) did not choose to offer genetic testing to an affected individual first, but rather chose to offer it to the sister of an affected male. While there is a 50% chance that this individual could be a carrier, knowing the affected son's genetic mutation would be important to avoid the possibility of an uninformative negative test result in the sister. Another 7.8% of physicians and 8.8% of nurses chose the mother of an affected son as the best candidate for testing. Offering genetic testing to the mother poses the same risk to receive an uninformative negative test result as with the sister of an affected male. Only after the causative mutation in

the family has been identified can genetic screening to other family members, whether they be unaffected relatives or obligate carriers, be most informative. It is worthwhile to point out that all four genetic counselors in the study chose to offer testing to the affected individual first. Our finding is not surprising, and similar studies have demonstrated a lack of consistency in the practices of providers in the context of genetic testing in individuals with a family history of a genetic condition. For example, a study performed by Mehnert *et al.* found that approximately 50% of the gynecologists they surveyed did not recognize the importance of having the genetic test results of an index patient when interpreting the test results of an unaffected individual (11). Without confirmation of a known familial mutation, negative genetic testing in an unaffected individual is not informative because it cannot rule out the possibility of an inherited mutation that was undetectable by the testing methodology used.

Attitudes

Our study aimed to determine the attitudes of providers toward genetic counseling services as well as their overall level of confidence in providing these services to patients. Previously, a study by Hunter *et al.* showed that the majority of Canadian physicians felt that they possessed adequate genetic knowledge; however, less than 50% of them felt comfortable discussing information about genetic services with their patients (6). Since healthcare providers working in HTCs see a large number of patients with hereditary bleeding disorders we wanted to assess their attitudes toward educating patients about genetics. Overall, attitude scores among physicians and nurses in our study were high indicating that most hematologists and nurses in HTCs feel confident their ability to provide genetic counseling services. Questions in this section focused on common aspects of hemophilia genetics such as counseling a patient about genetic testing and helping a patient decided whether or not to pursue genetic testing. Other attitude questions aimed to identify the provider's perceived value of genetic counseling services. The majority of participants indicated that they see value in genetic counseling services, especially for first-degree female relatives of affected males.

In general, physicians felt more comfortable discussing possible insurance implications with patients than nurses did. A study by Acton *et al.* found that 79% of physicians (family practitioners, general internists, obstetrician-gynecologists) felt as though the information obtained from genetic testing could be used by employers and insurance companies to discriminate against people who had an increased risk for a hereditary cancer (9). We hypothesized that a similar attitude may be present in HTCs since many patients, particularly potential carriers, are concerned that having genetic testing will result in insurance

discrimination, in the form of increased premiums or gap in coverage due to a pre-existing condition.

Comparison of knowledge and attitude questions addressing the same theme revealed discordance between provider knowledge and perception of ability to provide a specific genetic counseling service. While not statistically significant these results illustrate the dilemma that arises when providers inaccurately educate/counsel patients when they believe that they are providing the correct information. Specifically, question 13 in the knowledge section asked providers to identify an obligate carrier in a pedigree. This question was compared to the provider's response to the statement: I feel comfortable educating my patient about X-linked inheritance. Twelve out of the 16 (75.0%) providers who incorrectly identified an obligate female carrier, somewhat to strongly agreed that they were comfortable discussing inheritance with a patient. This highlights an inconsistency in the accuracy of the information being delivered by providers and their recognition of their own abilities. Two other hereditary questions produced similar results (questions 7 and 9 in the knowledge section) when compared to the provider's attitude toward discussing heredity with a patient.

Limitations

Despite our best efforts, this study had a few limitations which must be considered. One significant limitation of this study is that it was only distributed to physicians and nurses whose email addresses were listed in the CDC's online directory. This directory does not include the contact information for every provider in every center nationwide, and therefore this sample is biased by the fact that it was selectively distributed to providers listed in the directory. Also, the number of complete responses is not sufficiently large to produce results with a high statistical power. In addition this study lacked a validated instrument. Even though our tool was modeled after the survey developed and validated by Hofman *et al.* in the 1990s, it was not validated prior to use in the hemophilia community. As a result, some of the incorrect responses may not indicate a deficit of knowledge, but rather a misinterpretation of the question or answer choices.

Future Directions

While this study provides a snapshot of the knowledge and attitudes of hematologists and nurses in U.S. HTCs it does not provide insight into the knowledge and attitudes of genetic counselors or other providers who are affiliated with HTCs. Based on the results of our questionnaire, the majority of providers (91.6%) report having access to genetic counseling

services via a genetic counselor; however, not every HTC has a genetic counselor working on site. A follow-up study assessing the knowledge and attitudes of genetic counselors providing genetic counseling services to patients affected by hemophilia would be beneficial to further refine the understanding of the level of service available to patients at U.S. HTCs.

Conclusion

Hematologists and nurses practicing in U.S. HTCs demonstrate sufficient knowledge of the genetics of hemophilia, and they generally feel confident in their ability to provide genetic counseling services to their patients. While their knowledge is sufficient, the average knowledge score was lower than the 75% that we had anticipated. In addition, approximately 23% of physicians and nurses did not demonstrate sufficient knowledge of hemophilia genetics in our study. Based on these findings, there is room for improvement in the genetics education of providers working in HTCs. Interestingly, there is no significant difference in the knowledge or attitudes between hematologists and nurses in HTCs; however, there is evidence to suggest that there are some topics that physicians feel more comfortable discussing with their patients than nurses do. In addition, level of knowledge has no effect on a provider's level of confidence in providing genetic counseling services. Finally, many clinics report having "access" to a counselor, but the reality is that in clinic, it is oftentimes the nurse or physician who is providing genetic counseling services to patients. Given the inconsistency in provider knowledge coupled with the high confidence in one's ability to counsel patients, it leaves room to question whether information about the genetics of hemophilia is being communicated to patients in the most appropriate and accurate manner.

Appendix:

Table 1: Summary of demographic characteristics of study population

Characteristic	Physicians		Nurses	
	n	%	n	%
Age				
20-29 years	0	0.00%	5	8.33%
30-39 years	7	13.73%	8	13.33%
40-49 years	13	25.49%	11	18.33%
50-59 years	19	37.25%	29	48.33%
60+ years	12	23.53%	7	11.67%
Gender				
Male	24	47.06%	3	5.00%
Female	25	49.02%	54	90.00%
Not disclosed	2	3.92%	3	5.00%
Patient Population				
Adult	12	23.53%	6	10.00%
Pediatric	20	39.22%	16	26.67%
Both	16	31.37%	38	63.33%
Not disclosed	3	5.88%	0	0.00%
Certified				
Yes	47	92.16%	16	26.67%
No	3	5.88%	31	51.67%
I don't know or N/A	1	1.96%	13	21.67%
Years of HTC experience				
Less than 1 year	0	0.00%	4	6.67%
1-5 years	7	13.73%	15	25.00%
5-10 years	11	21.57%	6	10.00%
10-20 years	18	35.29%	21	35.00%
More than 20 years	14	27.45%	14	23.33%
Not disclosed	1	1.96%	0	0.00%
Average size of patient population				
Less than 50 patients	4	7.84%	14	23.33%
50-100 patients	20	39.22%	21	35.00%
100-200 patients	17	33.33%	15	25.00%
More than 200 patients	9	17.65%	9	15.00%
Not disclosed	1	1.96%	1	1.67%

Table 2: Pass Rates by provider type

Score	Physician		Nurse	
	n	%	n	%
Pass	38	74.51%	43	71.67%
Fail	13	25.49%	17	28.33%

Table 3: Summary of knowledge section, by provider type

Question (correct answer)	Physician				Nurse				P-value
	Correct		Incorrect		Correct		Incorrect		
	n	%	n	%	n	%	n	%	
1. Genetic testing of the F8 gene detects mutations in what percentage of men with severe Hemophilia A? (80-100%)	38	74.51%	13	25.49%	32	53.33%	28	46.67%	0.021
2. Which genetic test is most likely to be informative in a male with severe hemophilia A? (Factor VIII intron 22 inversion analysis)	39	76.47%	12	23.53%	43	71.67%	17	28.33%	0.566
3. Prenatal diagnosis is most informative when a familial gene mutation is known. (True)	48	94.12%	3	5.88%	58	96.67%	2	3.33%	0.660
4. Preimplantation genetic diagnosis can be useful even if the familial gene mutation is not known (False)	20	39.22%	31	60.78%	25	41.67%	35	58.33%	0.793
5. A female with normal factor VIII level (70-140%) cannot be a carrier (False)	49	96.08%	2	3.92%	60	100.00%	0	0.00%	0.209
6. The chance of a couple having a boy with hemophilia if the mother is a carrier is: (1 in 4 live births)	26	50.98%	25	49.02%	23	38.33%	37	61.67%	0.181
7. The chance of a couple having a boy with hemophilia if the father has hemophilia and the mother is not a carrier is: (None of the above)	49	96.08%	2	3.92%	59	98.33%	1	1.67%	0.593
8. An 8 year old boy with hemophilia A comes into clinic with his mother and 4 year old sister. The sister had a blood test which revealed a factor level of 60%. What do you tell the mother about her daughter? (There is no way to know . . .)	45	88.24%	6	11.76%	58	96.67%	2	3.33%	0.140
9. A boy is diagnosed with severe hemophilia after developing bleeding symptoms. Genetic testing for this boy revealed an intron 22 inversion mutation in the F8 gene. The chance that his mother is a carrier for hemophilia is: (90-100%)	37	72.55%	14	27.45%	40	66.67%	20	33.33%	0.503
10. If one boy in a family has hemophilia but he has no other family members with bleeding symptoms, the chances that the next son of the same parents will have hemophilia is: (50%)	35	68.63%	16	31.37%	52	86.67%	8	13.33%	0.021
11. In order to determine a female's carrier status one should order: (Both factor level and genetic testing)	42	82.35%	9	17.65%	39	65.00%	21	35.00%	0.040
12. In an X-linked condition: (50% of the daughters of female carriers will be carriers)	49	96.08%	2	3.92%	58	96.67%	2	3.33%	1.000
13. Which female family members are obligate carriers? (Individual II-2 only)	45	88.24%	6	11.76%	50	83.33%	10	16.67%	0.464
14. Which person in this family would be the best candidate to offer genetic testing? (Individual III-1)	20	39.22%	31	60.78%	23	38.33%	37	61.67%	0.924

Table 4: Summary of knowledge section

Question (correct answer)	Correct		Incorrect	
	n	%	n	%
1. Genetic testing of the F8 gene detects mutations in what percentage of men with severe Hemophilia A? (80-100%)	70	63.06%	41	36.94%
2. Which genetic test is most likely to be informative in a male with severe hemophilia A? (Factor VIII intron 22 inversion analysis)	82	73.87%	29	26.13%
3. Prenatal diagnosis is most informative when a familial gene mutation is known. (True)	106	95.50%	5	4.50%
4. Preimplantation genetic diagnosis can be useful even if the familial gene mutation is not known (False)	45	40.54%	66	59.46%
5. A female with normal factor VIII level (70-140%) cannot be a carrier (False)	109	98.20%	2	1.80%
6. The chance of a couple having a boy with hemophilia if the mother is a carrier is: (1 in 4 live births)	49	44.14%	62	55.86%
7. The chance of a couple having a boy with hemophilia if the father has hemophilia and the mother is not a carrier is: (None of the above)	108	97.30%	3	2.70%
8. An 8 year old boy with hemophilia A comes into clinic with his mother and 4 year old sister. The sister had a blood test which revealed a factor level of 60%. What do you tell the mother about her daughter? (There is no way to know . . .)	103	92.79%	8	7.21%
9. A boy is diagnosed with severe hemophilia after developing bleeding symptoms. Genetic testing for this boy revealed an intron 22 inversion mutation in the F8 gene. The chance that his mother is a carrier for hemophilia is: (90-100%)	77	69.37%	34	30.63%
10. If one boy in a family has hemophilia but he has no other family members with bleeding symptoms, the chances that the next son of the same parents will have hemophilia is: (50%)	87	78.38%	24	21.62%
11. In order to determine a female's carrier status one should order: (Both factor level and genetic testing)	81	72.97%	30	27.03%
12. In an X-linked condition: (50% of the daughters of female carriers will be carriers)	107	96.40%	4	3.60%
13. Which female family members are obligate carriers? (Individual II-2 only)	95	85.59%	16	14.41%
14. Which person in this family would be the best candidate to offer genetic testing? (Individual III-1)	43	38.74%	68	61.26%

Table 5: Attitudes toward genetic counseling services, by provider type

Statement	Physicians			Nurses			P-value
	Disagree	Agree	Strongly Agree	Disagree	Agree	Strongly Agree	
1. All mothers of sons with hemophilia should be offered genetic counseling.	3	2	45	4	9	47	0.164
2. All first degree female relatives of a patient with hemophilia should be offered genetic counseling.	3	2	45	5	8	47	0.217
3. The majority of my patients would decline genetic counseling if it were offered to them.	44	3	4	43	11	6	0.119
4. I commonly refer patients for genetic counseling.	11	9	31	16	22	22	0.027
5. In special circumstances I refer patients for genetic counseling.	10	8	33	12	14	31	0.534
6. I only refer patients for genetic counseling when they request it.	39	4	6	49	5	5	0.875
7. I would feel comfortable explaining the benefits and limitations of genetic testing for hemophilia.	2	14	34	10	18	31	0.067
8. I feel comfortable educating a patient about X-linked inheritance.	3	3	45	4	8	48	0.464
9. My patients would benefit from meeting with a genetic counselor.	5	8	38	1	13	45	0.163
10. My HTC would benefit from having a genetic counselor on staff.	5	8	37	4	17	36	0.230

Table 6: Confidence in ability to provide genetic counseling services, by provider type

Statement	Physicians			Nurses			P-value
	Not confident	Neither	Confident	Not confident	Neither	Confident	
11. Discuss the risks and benefits of being tested for hemophilia	3	1	47	5	5	48	0.291
12. Help a patient understand the possible implications/use of a positive test result	3	2	46	6	4	48	0.586
13. Discuss possible insurance implications of having genetic testing for hemophilia	8	11	32	22	13	24	0.026
14. Help a patient decide whether to be tested	3	5	43	5	15	38	0.063
15. Help the patient cope with a positive test result	2	4	44	3	5	51	1.000
16. Discuss the patient's fears and concerns about having a child with hemophilia	2	2	46	1	4	53	0.653
17. Discuss the risks and benefits of being tested for hemophilia	2	3	45	3	4	52	1.000
18. Help a patient understand the possible implications/use of a positive test result	2	2	45	1	8	50	0.221
19. Help a patient decide whether to be tested	2	4	41	4	10	45	0.349
20. Discuss the meaning and implications for patients of a negative test result	2	3	45	3	6	50	0.752

Table 7: Access to Genetic Counseling Services

Access	Physicians		Nurses		Combined	
Access to Genetic Counseling Services	**n**	**%**	**n**	**%**	**n**	**%**
Yes	45	91.84%	53	91.38%	98	91.59%
No	4	8.16%	5	8.62%	9	8.41%
Type of Access						
Genetic counselor attends every clinic	15	33.33%	9	16.98%	24	24.49%
Genetic counselor available by phone as needed	2	4.44%	2	3.77%	4	4.08%
Genetic counselor available by referral to an outside institution	2	4.44%	6	11.32%	8	8.16%
Genetic counselor available to meet with patient at your clinic as needed	14	31.11%	20	37.74%	34	34.69%
Other	12	26.67%	16	30.19%	28	28.57%

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VITA

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