HHS Public Access

Author manuscript

Clin Genet. Author manuscript; available in PMC 2016 December 01.

Published in final edited form as:

Clin Genet. 2015 December ; 88(6): 523–529. doi:10.1111/cge.12563.

Genetic counselors' practices and confidence regarding variant of uncertain significance results and reclassification from *BRCA* testing

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Abstract

Studies indicate variant of uncertain significance (VUS) results are challenging for genetic counselors and patients, often resulting in negative patient outcomes. Genetic counselors' current practices regarding VUS are unknown. This study utilized a national survey of genetic counselors (n = 932) to examine current practices and confidence related to disclosing *BRCA* VUS results and reclassification information. For participants (n = 398), descriptive statistics were calculated for patient demographic characteristics, practices and confidence, and cross tabulation was used to identify participant's actions when receiving a reclassified VUS. Upon receiving a *BRCA* VUS report, the majority reported providing patients with information about the frequency their VUS was seen and patient ancestry, but a minority discussed DNA banking. Most were confident in their understanding of and ability to explain VUS results to patients, but felt less confident about achieving high levels of patient understanding. Upon reclassification, the majority reported calling the patient and mailing the results, but when the reclassification was deleterious the majority also met with the patient face-to-face. Given the lack of standard professional guidelines regarding informing patients about initial and reclassified VUS results, this overview provides important insight into genetic counselors' current practices and confidence.

Keywords

Genet Counseling; Genet Testing; Hered Cancer; Variant of Uncertain Significance (VUS); VUS Reclassification

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The authors declare they have no conflicts of interest.

Introduction

BRCA1 and *BRCA2* (*BRCA*) mutations account for ∼5-10% of all breast and ovarian cancers (1). Lifetime cancer risks in *BRCA* mutation carriers are approximately 60-70% for breast cancer (2-5) and 40% for ovarian cancer (2, 4). Genetic counseling and testing to identify women with *BRCA* mutations can directly inform screening and prevention measures to reduce cancer incidence and mortality (6-8). The model advocated by leading health and professional organizations (9-11) for providing comprehensive *BRCA* testing consists of a pretest genetic counseling session and posttest counseling for patients who proceed with testing (12).

Although no specific guidelines exist about how to disclose VUS results nor do guidelines exist regarding reclassification of VUS results, basic guidelines from the National Society of Genetic Counselors (NSGC) indicate that genetic counselors (GCs) should discuss variants of uncertain significance (VUS) with a patient during pretest genetic counseling and at posttest genetic counseling, if a VUS is found (13). The pretest genetic counseling session should include a discussion about possible test results, including: (1) positive – a known cancer predisposing gene mutation is identified; (2) negative – no cancer predisposing gene mutation is identified in a family where another member(s) has tested positive; (3) uninformative – no cancer predisposing gene mutation identified in a family where another member(s) has tested negative or not been tested; and (4) VUS – a gene change is identified, but the current cancer risks, if any, are unknown (12). To our knowledge, only one earlier study specifically examined GCs' practices in the United States (US) regarding VUS results at pre-test and post-test counseling (14). Findings from that study indicate most GCs disclosed the possibility of a VUS result prior to testing, but had difficulty achieving patient understanding when presenting VUS results in the post-test session (14), a finding also reported in other studies of genetics professionals (15-17). Furthermore, negative patient outcomes included incomplete understanding of VUS results (18), inaccurate risk perceptions (18), distress (19), and unnecessary prophylactic surgeries (20, 21).

Since the earlier study was conducted, several major clinical and scientific changes likely impacted GCs' approaches to posttest genetic counseling and patient follow-up. Specifically, the majority provider of clinical *BRCA* testing no longer provides a “detailed” report for GCs including information such as: how often the variant was seen, the ethnic groups in which it was observed, co-segregation with disease, and whether it was identified in a person with a known deleterious mutation. Additionally, major efforts by the scientific community to reclassify VUS as deleterious or benign using multifactorial probability-based modeling led to scientific publications and the creation of public databases to help interpret and reclassify VUS (22, 23). Although not indicated by current guidelines, these efforts may require follow-up with patients with reclassified VUS. Therefore, in addition to assessing practices and confidence when disclosing a VUS result, it is important to explore how GCs handle VUS reclassification. The purpose of the current study was to characterize current practices and confidence related to: 1) disclosure of VUS results, and 2) the reclassification of VUS results.

Materials and Methods

Sample

Mailing lists of GCs were obtained from NSGC and the American Board of Genetic Counseling (ABGC). The study population included those who: 1) self-identified as practicing cancer genetic counseling, 2) reported their primary job activity to be clinical patient care, and 3) had a US mailing address. ABGC provided a pre-sorted mailing list by cancer practice setting yielding 621 US-based GCs. NSGC provided a spreadsheet of all members (n = 2,451) including members' area of practice, types of specialization, and work setting. The mailing list was sorted to include members who reported “cancer” or “PGM” [personalized genomic medicine] as their specialization, “cancer” as an area of practice, and/or reported a company name that included “cancer” and/or “oncology,” yielding 751 GCs. After excluding duplicate names appearing on both lists, the compiled mailing list included 546 from NSGC and 429 from ABGC for a total of 975 GCs. Eleven GCs who participated in the piloting of the survey to verify face and content validity and 32 additional duplicate names identified during mailing were omitted for a final list of 932 unique GCs included in this study.

Survey Instrument

Items included those from a previous survey of VUS among GCs (14), a survey about delivery of genetic counseling and testing services for hereditary breast and ovarian cancer (HBOC) (24), and questions developed specifically for the current survey. In addition to demographic and practice characteristics, the final 43-item survey included 2 sections to assess practice patterns and confidence related to *BRCA* VUS upon receiving a VUS result and upon receiving reclassification information. GCs were asked to complete either a scannable paper-based or web-based survey.

Post-test counseling practices

Using a 5-point scale ranging from never to always, participants were asked to report how often they: 1) inform the patient of the number of times their VUS result has been seen before, 2) share the ancestry of families in whom the patients' VUS result has been seen, and 3) discuss the option of DNA banking with the patient. Participants also were asked to report, on a 5-point scale ranging from not confident at all to extremely confident, in their abilities to: 1) personally understand a *BRCA* VUS result, 2) explain a VUS result to a patient, 3) increase a patient's understanding about the meaning of a *BRCA* VUS test result, 4) achieve high levels of patient understanding in the genetic counseling and testing process for patients with a *BRCA* VUS result, and 5) discuss clinical management options with patients who have a *BRCA* VUS result. Additionally, participants were asked to report whether they currently use, would use if available, or would not use even if available the following patient educational materials: a factsheet, booklet, DVD, or template letter for relatives.

Counseling practices when VUS results are reclassified

Regarding VUS reclassification, participants were asked to select all among a list of actions taken upon receiving a VUS reclassified as deleterious and a VUS reclassified as a favor polymorphism (i.e., benign) including: 1) calling the patient and discussing results by phone, 2) mailing an explanation letter to the patient, 3) mailing a copy of results to the patient, 4) mailing an explanation letter to the patient's provider, 5) mailing a copy of the results to the patient's provider, and 6) requesting the patient come in for a face-to-face counseling session. Participants also were asked to report, on a 5-point scale ranging from strongly agree to strongly disagree, whether they believed it was the responsibility of the genetic counselor who counseled the patient for their *BRCA* VUS to inform the patient if the result was reclassified.

Data Collection

Following Institutional Review Board approval from Mayo Clinic and the University of South Florida, a multiphase recruitment approach was used (25). First, prospective participants were mailed a postcard to inform them of the forthcoming survey and allowed the study team to verify mailing addresses. Seven weeks later, GCs were mailed a packet via US Postal Service that included a cover letter, scannable survey, prepaid return envelope, and two articles authored by study team members pertaining to VUS reclassification (22, 26). A prepaid response card was included to allow individuals to indicate completion of the survey, and was mailed separately from the survey to maintain anonymity. The prepaid response cards were used to track who should receive a reminder postcard mailed four weeks later, followed by a final survey approximately three weeks later. In addition to paper surveys, web surveys were emailed to GCs with available email addresses (n = 902) within a week of the first mailed survey packet. Reminder emails were sent three weeks later. Data collection occurred between February and May 2013. GCs were asked to return completed surveys by April 30, 2013 to be entered into a drawing to win one of four $50 bills; the final survey was accepted on May 15, 2013.

Data Analysis

Frequencies and percentages for participant demographic characteristics, practices, and confidence were obtained using SAS 9.3. Cross tabulations were conducted to compare participants' actions taken upon receiving VUS updates about reclassification results (reclassified as deleterious or suspected deleterious vs. no mutation detected or favor polymorphism).

Results

Of the 932 surveys mailed, 29 were undeliverable (3%) and 19 were ineligible based on inclusion criteria (2%). A total of 410 completed surveys were received, for a 46% response rate. Although not included in initial eligibility criteria, the survey was designed for GCs with experience specific to HBOC; therefore 12 participants were excluded because 5 reported no such experience and 7 had missing data for this question, for a final sample of 398. Ten percent of the paper surveys were checked for scanning errors, and none were detected.

The sample was predominately female (94.2%), White/Caucasian (91.7%), and non-Hispanic/Latino (96.7%). Most participants were board certified in genetic counseling, (93.5%), reported direct patient care as their primary role (88.4%), cancer as their primary specialty area (75.1%), and provided counseling for HBOC for an average of 7.4 years. The majority worked in a non-academic setting (63.3%). Over 60% of participants counseled between 1 and 12 patients for *BRCA* genetic testing results in the past month, and approximately 67% reported counseling between 1 and 6 patients with a *BRCA* VUS result in the past year (Table 1).

With regard to VUS results disclosure, the majority reported often or always informing patients about the number of times their VUS was seen (79.1%), patient ancestry of families in whom their VUS result was seen (56.8%), but few often or always discussed the option of DNA banking (17.8%; Table 2). Most reported feeling very or extremely confident in their own understanding of *BRCA* VUS (82.4%), were confident they could explain a *BRCA* VUS result to patients (88.7%), and could increase a patient's understanding about *BRCA* VUS (81.9%). Just over half felt confident in achieving high levels of patient understanding about *BRCA* VUS (62.8%) and were confident in discussing medical management options with patients with *BRCA* VUS results (62.6%; Table 3). Concerning the use of patient education materials about *BRCA* VUS, the highest percentage of respondents reported currently using a template letter for relatives (25.1%); over half reported they would use a template letter for relatives (52.3%); a patient education fact sheet (65.1%), and/or a patient education booklet (56.5%) if these materials were available. The majority indicated they would not use a patient education DVD (62.6%), even if it was available (Table 4).

Regarding VUS reclassification, the majority agreed or strongly agreed that the responsibility for informing a patient about VUS reclassification belongs to the genetic counselor (78.4%). Participants were asked to report all measures taken upon receiving updates about the reclassification of VUS results in the case of a deleterious, or suspected deleterious mutation and in the case of no mutation detected or a favor polymorphism. Among those who reported receiving reclassification information, when reclassified as deleterious, slightly more than half of the sample indicated calling, meeting face-to-face, and mailing the patient results and/or a patient letter (54.2%). However, when reclassified as no mutation detected/favor polymorphism, the majority indicated calling and mailing the patient the results and/or a patient letter (67.3%). With regard to the information provided upon reclassification to the patient's healthcare provider, the majority reported providing both a letter and the reclassification results when the VUS was reclassified as deleterious (53.9%) and when the VUS was reclassified as a benign variant (55.7%; Table 5).

Discussion

Recent changes in clinical laboratory reports and scientific advances in reclassification of VUS results (22, 23) compeled an updated exploration of genetic counseling practice related to *BRCA* VUS. Therefore, this study reports GCs' current practices and confidence related to counseling patients who receive VUS results and reclassification information for VUS results.

When counseling patients with a VUS result, the majority of GCs in this study reported sharing information with their patients about the number of times the variant was seen and the ancestry of families with the VUS, whereas a minority did so in an earlier study (14). At the time of the previous study, the laboratory just began including these items on the report, so the lower rate of information sharing in the earlier study may be an artifact of information availability.

Additionally, Petrucelli and colleagues (14) found just over half of GCs believed patients understood their VUS results, a finding consistent with results of the current study. They speculated that this low rate may “reflect the counselors' own uneasiness with an ambiguous outcome” (14) or difficulty in understanding on the patients' part due to the complexity of VUS results. Results of the current study seem to indicate the latter, as most participants were confident in their own understanding and ability to explain a *BRCA* VUS result to patients, but they were less likely to be confident in their ability to achieve high levels of patient understanding about VUS and discuss medical management options. Consistently, results from previous studies found patients believe VUS results are difficult to understand (15) and perceive genetic counseling and testing to be less informative and reassuring (27). Difficulty achieving high levels of patient understanding may explain participants' desire for patient educational materials. Although few GCs reported using patient education materials, most would use such materials if available, and a preference for paper-based interventions was indicated. Future research efforts should develop empirically tested patient education materials to support GCs' discussion of VUS results.

Low rates of discussion regarding DNA banking were reported in this study as well as in the earlier study (14). DNA banking is the storage of genetic material for future genetic testing or research. As such, DNA banking may primarily be discussed in the context of end of life care, when genetic testing is not an immediate option for the affected family member, providing family members with future access to important health information. As such, it may not be relevant to discuss DNA banking in most instances of cancer genetic counseling.

Reclassification of VUS occurs as data accumulate and new mathematical models are developed (17). Despite ongoing reclassification efforts and recent calls for guidelines, professional organizations including the National Comprehensive Cancer Network and the NSGC do not yet provide guidelines addressing the responsibility of providers when a VUS is reclassified (15). Few studies explored the delivery of VUS reclassification results and those that do focus on patient outcomes (15, 19, 20). Most GCs in this study reported calling the patient and mailing the results and/or a patient letter regardless of whether the VUS was reclassified as deleterious or benign, and took the additional action of meeting the patient in-person when the VUS was reclassified as deleterious. GCs are increasingly using telephone-based results disclosure (28), an effective alternative to face-to-face genetic counseling (29-31). However, it is unclear whether telephone-based reclassification results disclosure is as effective, particularly given a significant amount of time may have passed since the original counseling session, thus reducing rapport established during earlier sessions. Future comparative studies should explore the effectiveness of telephone-based reclassification results disclosure compared with face-to-face reclassification results disclosure. In the meantime, it may be equally important to meet the patient in-person when the VUS is

reclassified as benign to provide any necessary psychosocial support in adjusting to the new information.

When providing information about reclassified VUS to patients' healthcare providers, the majority reported sending a letter and the results to patients' healthcare providers when the VUS was reclassified as deleterious or benign. It is interesting to note, however, that over a quarter of participants reported taking neither of these actions. The patients' primary healthcare provider is likely responsible for the patient's medical management follow-up and was the main factor in patients' adherence to medical management guidelines related to hereditary cancer (32). This communication may be particularly important given documented non-genetics professionals' difficulties related to VUS including providing inappropriate testing recommendations for unaffected relatives of VUS carriers (33), and suggesting inappropriate or unnecessarily invasive medical management in the case of VUS or negative results (33-36). However, it is possible that other channels are being used to communicate genetic test results to other healthcare providers that were not captured in our study. For example, electronic medical records may provide a channel of communication between the genetic counselor and the provider. Additionally, many individuals who undergo *BRCA* testing are cancer patients, and their test results may be discussed by the genetic counselors with other providers in the context of tumor boards. Future research should examine alternative mechanisms and channels for disseminating genetic testing results to other healthcare providers to ensure appropriate communication of results is occurring.

Our study is among the first to explore how GCs deal with reclassification of VUS results, but findings should be considered in light of limitations. GCs self-selected to participate in this study; thus, responses from those who participated may be different from those who chose to not participate in this study, limiting generalizability. This study did not examine the content of GCs' disclosure of reclassified VUS results, but it is important to note updated recommendations provided by the NSGC indicate disclosure of any test results should include: 1) a personalized interpretation of the results 2) a cancer risk re-assessment, and 3) identification of at risk family members (13). Future studies should examine the content of results disclosure counseling sessions upon reclassification of VUS results.

As VUS reclassification will continue to occur over time, understanding how VUS reclassification results are currently delivered in genetic counseling practice and identifying best practices and providing suggestions for guidelines is vital. Exploring how GCs provide information about VUS results to patients and identifying preferred sources of information and education is essential to improve future practice. Results presented here reported on closed-ended questions. A separate manuscript reported results from the three open-ended questions included on this survey (37). However, the open-ended questions did not explore reasons behind certain GC practices and preferences. Further qualitative research would provide an in depth understanding regarding certain GC practices and preferences.

In summary, study results are valuable for identifying needs when disseminating reclassified VUS results to GCs, and ultimately, patients. These findings are particularly important given several recent changes in the genetic testing environment. For example, the entry of other

laboratories into the BRCA testing market since this study has led to the complication of inconsistent classification of variants across laboratories and the increased use of panel testing. In contrast to single gene testing, such as the *BRCA* testing generally done at the time of this survey, panel testing allows for rapid testing of multiple genes at a time (38). Since the number of VUS are directly correlated with the number of genes tested, the increased use of panel testing increases the likelihood and number of VUS results per test (38, 39). The potentially high rate of VUS in the future magnifies the importance of continually assessing practice when providing genetic counseling about VUS results and reclassification.

Acknowledgments

This work was supported by the Breast Cancer Spore CA 116201. Dr. Courtney L. Scherr is supported by the R25 CA 090314 Behavioral Oncology Education and Career Development grant. This work was supported in part by the Survey Methods Core Facility at the H. Lee Moffitt Cancer Center & Research Institute; an NCI designated Comprehensive Cancer Center (P30-CA76292). The authors would also like to thank Nancie Petrucelli for her thoughtful review of the survey instrument used in this study.

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Table 1
Sample demographic and practice characteristics (*N* = 398)

Variable	*n* (%)[a]
Demographic Characteristics	
Age (mean, [range])	36.9 [24-71]
Sex	
Male	23 (5.8)
Female	375 (94.2)
Race	
White/Caucasian	365 (91.7)
Other	33 (8.3)
Ethnicity	
Hispanic or Latino	13 (3.3)
Not Hispanic or Latino	385 (96.7)
Primary Specialty Area (missing = 2)	
Cancer	299 (75.1)
Other	97 (24.4)
Primary Role (missing = 1)	
Direct patient care	352 (88.4)
Other	45 (11.3)
Primary Work Setting	
University medical center (academic)	146 (36.7)
Other (non-academic)	252 (63.3)
Board Certified in GC	
Yes	372 (93.5)
No, board eligible and plan to take/ not board eligible	26 (6.5)
Years provided GC for HBOC (mean, [range])	7.4 [1-33]
Practice Characteristics	
No. Patients Counseled for *BRCA*Results in Past Month	
0	37 (9.3)
1-6	122 (30.7)
7-12	121 (30.4)
13-24	95 (23.9)
≥25	19 (4.8)
No. Patients with *BRCA* VUS in Past Year	
0	45 (11.3)
1-6	268 (67.3)
7-12	56 (14.1)
13-24	17 (4.3)
25-49	4 (1.0)
≥50	1 (0.3)

[a]Percentages may not sum 100 due to rounding or missing data.

Abbreviations: GC, genetic counseling/counselor; HBOC, hereditary breast and ovarian cancer; VUS, variant of uncertain significance

Table 2

Information provided by genetic counselors to patients about VUS (*N* = 398).

Action	Never *n*(%)[a]	Rarely *n*(%)[a]	Sometimes *n*(%)[a]	Often *n*(%)[a]	Always *n*(%)[a]
Inform the patient of the number of times their VUS result has been seen before	6(1.5)	11(2.8)	41(10.3)	118(29.6)	197(49.5)
Share with the patient the ancestry of families in whom their VUS result has been seen	15(3.8)	37(9.3)	94(23.6)	103(25.9)	123(30.9)
Discuss the option of DNA banking with the patient	46(11.6)	127(31.9)	127(31.9)	43(10.8)	28(7)

[a]Percentages may not sum 100 due to rounding or missing data.

Table 3

Genetic counselors' confidence in their ability to do specific actions for patients with *BRCA* VUS results (*N* = 398).

Confidence	Not at all/ Somewhat Confident *n* (%)[a]	Moderately Confident *n* (%)[a]	Very Confident *n* (%)[a]	Extremely Confident *n* (%)[a]
Confident with your understanding of *BRCA* VUS results	9 (2.3)	39 (9.8)	160 (40.2)	168 (42.2)
Explain a *BRCA* VUS result to a patient	3 (0.8)	23 (5.8)	154 (38.7)	199 (50.0)
Increase a patient's understanding about the meaning of a *BRCA* VUS test result	5 (1.3)	47 (11.8)	155 (38.9)	171 (43.0)
Achieve high levels of patient understanding in the genetic counseling and testing process for patients with a *BRCA* VUS result	17 (4.3)	109 (27.4)	166 (41.7)	84 (21.1)
Discuss clinical management options with patients who have a *BRCA* VUS result	28 (7.0)	100 (25.1)	144 (36.2)	105 (26.4)

[a] Percentages may not sum 100 due to missing data.

Note: “Not at all confident” and “Somewhat confident” groups were combined due to low frequencies.

Abbreviations: VUS, variant of uncertain significance

Table 4

Genetic counselors' use of patient education materials regarding *BRCA* VUS results (*N* = 398)

Materials	Currently use *n* (%)[a]	Would use if these materials were available *n* (%)[a]	Would not use if these materials were available *n* (%)[a]
Patient education fact sheet	94 (23.6)	259 (65.1)	17 (4.3)
Patient education booklet	83 (20.9)	225 (56.5)	51 (12.8)
Patient education DVD	5 (1.3)	78 (19.6)	249 (62.6)
Template letter for relatives	100 (25.1)	208 (52.3)	58 (14.6)

[a]Percentages may not sum 100 due to missing data.

Table 5

Frequencies of actions and combined actions taken upon receiving a reclassified VUS result.

VUS reclassified as deleterious (*n* = 308*)	Frequency (percent)	VUS reclassified as benign (*n* = 345*)	Frequency (percent)
Actions taken with the patient			
Call only	10 (3.25%)	Call only	24 (6.96%)
Face-to-Face only	16 (5.19%)	Face-to-Face only	2 (0.58%)
Call + Mail	71 (23.05%)	Call + Mail	232 (67.25%)
Call + Face-to-Face	32 (10.39%)	Call + Face-to-Face	15 (4.35%)
Mail+ Face-to-Face	11 (3.57%)	Mail+ Face-to-Face	3 (0.87%)
Call + Mail + Face-to-Face	167 (54.22%)	Call + Mail + Face-to-Face	34 (9.86%)
None of these	1 (0.32%)	None of these	2 (0.58%)
Actions taken with other healthcare providers			
Letter only	29 (9.42%)	Letter only	29 (8.41%)
Results only	21 (29.87%)	Results Only	29 (8.41%)
Letter + Results	166 (53.90%)	Letter + Results	192 (55.65%)
None of these	92 (29.87%)	None of These	95 (27.54%)

*Note: Only those who reported experience with VUS reclassification were included in this analysis.