



Short communication

Room for improvement in capturing cancer family history in a gynecologic oncology outpatient setting

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ABSTRACT

The literature demonstrates that the quality of cancer family history (CFH) as currently collected in the outpatient setting is inadequate to assess disease risk. Prior to implementation of a web-based application for cancer family history collection, we aimed to review the quality of collected CFH in a gynecologic oncology outpatient clinic and determine contributing patient factors. Medical records were reviewed for 200 new patients presenting between 4/2019–7/2019. CFH was collected during the patient interview and evaluated for inclusion of eight elements based on standards set by the genetics community. Univariate and multivariable linear regression analyses were utilized to evaluate the effect of patient characteristics on the number of relatives included in the CFH. Among our cohort of 200 patients, CFH was documented for 185 patients (92.5%). On univariate analysis, patients with a family history of cancer and prior genetic testing had significantly greater median number of relatives included in the CFH. On multivariable analysis, patients with family members with cancer had significantly more relatives included. Our data are consistent with the literature, suggesting that the current collection methods may not adequately capture all measures of a high quality CFH. Patients reporting no family history of cancer and those without prior genetic testing were least likely to have CFH that included key quality elements and these patients might benefit from health information technology CFH collection tools.

1. Introduction

An accurate cancer family history (CFH) can identify individuals at increased risk for inherited diseases and allow such patients to be triaged to genetic counseling and testing. For gynecologic cancer syndromes, including hereditary breast and ovarian cancer and Lynch syndrome, diagnosing familial mutations is critical, with implications for the health of the patient and at-risk relatives. Individuals with a cancer predisposition syndrome may be candidates for targeted therapies, increased cancer surveillance (e.g., breast and colon screening) and cancer prevention (e.g., salpingo-oophorectomy, mastectomy, colectomy), which can decrease cancer morbidity and mortality (Wood et al., 2014; Moore et al., 2018). Multiple organizations including the American College of Obstetricians and Gynecologists (ACOG) and Society of Gynecologic Oncology (SGO) have guidelines that rely on family history to prompt

referral for genetic assessment (Hereditary Cancer Syndromes and Risk Assessment, 2019; Committee on Practice Bulletins–Gynecology CoG, Society of Gynecologic Oncology, 2017).

The collected CFH must be accurate and sufficiently comprehensive for providers to rely on CFH for genetic risk assessment. Unfortunately, prior studies suggest that collection of CFH in a primary care setting is variable and the data collected are often inadequate to accurately assess disease risk (Acton et al., 2000; Murff et al., 2004). To the best of our knowledge, there exists no prior literature on the quality and comprehensiveness of cancer family history in a gynecologic oncology setting. Prior to implementing a prospective trial utilizing a web-based CFH collection tool to address these issues, we aimed to determine the existing quality of collected CFH in a gynecologic oncology clinic and to identify modifying patient factors.

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2. Methods

This study was approved by the Weill Cornell Medicine Institutional Review Board (IRB) and requirement for written informed consent was waived by the IRB. A review was conducted of all new patients presenting to the gynecologic oncology outpatient clinic between April 2019 and July 2019 to reflect the most recent CFH prior to the implementation of a web-based CFH collection tool. A three-month time period was chosen to allow an appropriate number of new patients to matriculate. Standard of care was collection of CFH verbally during patient face-to-face interviews. For non-English speaking patients, a telephone interpreter was used for the collection of all health information. The CFH was transcribed into the electronic medical record (EMR) (EpicTogether) by attending physicians and physician assistants at the time of the visit. Patient demographics and clinical information were obtained from the EMR. Patients who were adopted were excluded from this study.

CFH was evaluated for quality based on standards set by the genetics community including the following elements: 1) Patient's ethnicity, 2) Relatives' gender, 3) Relatives' lineage (maternal vs. paternal), 4) Inclusion of at least three generations, 5) Pertinent negatives, 6) Relatives' cause of death, 7) Relatives' age at death, and 8) Relatives' age at cancer diagnosis (for patients with a family history of cancer) ([Committee on Practice Bulletins–Gynecology CoG, Society of Gynecologic Oncology, 2017](#); [Bennett, 2004](#); [ACOG Practice Bulletin No, 2014](#)). The presence of pertinent negatives was determined by explicit denotation of “no family history” of cancer or specific types of cancer in the CFH. The EMR was evaluated to determine if the patient's ethnicity was documented.

2.1. Statistical methods

The distribution of continuous variables was tested for normality via the Shapiro-Wilk normality test. To evaluate if certain CFH elements were associated with sociodemographic or clinical factors, univariate tests were applied based on whether the variable of interest was distributed normally (i.e., *t*-test, analysis of variance) or not normally (i.e., Mann–Whitney *U* test, Kruskal–Wallace test). Associations between categorical variables were evaluated using the chi-square test or Fisher's exact test, as appropriate for category size. Multivariable linear regression analysis was performed to evaluate the independent effect of age, race, personal cancer history, family cancer history and prior genetic testing on the number of relatives included. Statistical significance was evaluated at the 0.05 alpha level, and 95% confidence intervals were calculated for all obtained estimates. Data were analyzed using Stata Version 16.0 (StataCorp, College Station, TX) and R version 3.6.1 (R Foundation for Statistical Computing, Vienna, Austria).

3. Results

3.1. Patient characteristics

The analysis included 200 patients presenting between 4/2019–7/2019. The median age was 52 years (range 23–93). The documented race of participants included White (90, 45%), Other (28, 14%), Asian (22, 11%), Black (20, 10%), and data not available (40, 20%). One hundred and twenty-seven patients (64%) were non-Hispanic/Latino, 14 (7%) Hispanic/Latino, and 59 (30%) data not available. Reasons for new patient appointment included pelvic mass (54, 27%), gynecologic cancer diagnosis (44, 22%), cervical or vulvar dysplasia (27, 14%), thickened endometrium (25, 13%), genetic mutation (20, 10%), postmenopausal bleeding (10, 5%), abnormal uterine bleeding (10, 5%), family history of cancer (4, 2%), and other (10, 5%). Five patients had multiple reasons for new patient appointment.

Sixty-three patients (32%) had prior cancer diagnoses, including 21 patients with breast cancer, 16 with uterine cancer, 8 with ovarian cancer, 7 with colorectal cancer, 2 with melanoma, and 18 with other

cancer diagnoses. Nine patients had two cancer diagnoses. One hundred and forty-seven patients (74%) reported at least one relative with cancer.

One hundred and eighty-five patients (93%) had CFH documented in the EMR, defined as patient history included in the family history portion of the chart. One hundred and fifty-six patients (78%) reported no previous genetic testing, 43 (22%) underwent prior testing, and one patient was unsure. Among those with prior testing, 24 (12% of the study population) were found to have a pathogenic variant, 16 (8%) had negative testing, and 3 (2%) reported inconclusive results. Thirty patients (15%) were referred for genetic testing following their gynecologic oncology appointment based on physician discretion, of which 19 (63%) completed genetic counseling and 16 (53%) completed testing ([Table 1](#)).

3.2. Characteristics of collected cancer family history

The collected CFH included a median of two generations (range 0–4). Pedigrees included a median of three relatives (range 0–15), including a median of two first-degree relatives (range 0–8), a median of one second-degree relative (range 0–7) and a median of zero third-degree relatives (range 0–4). The quality of CFH for the 185 patients with documented CFH in the new patient appointment was evaluated based on quality elements set by the genetics community. The number of patients with certain quality elements is depicted in [Fig. 1](#).

3.3. Number of relatives included

On univariate analysis, patients with a family history of cancer had significantly greater median number of relatives included in the CFH (4 [range 1–15]) than patients without a family history of cancer (2 [range 0–6]) ($P < 0.001$). Patients who had previously undergone genetic testing had significantly greater median number of relatives included in the CFH (4 [range 0–13]) than patients who had not undergone previous genetic testing (3 [range 0–15]) ($P = 0.012$) ([Table 2](#)). On univariate analysis, there were significant differences in the median number of relatives included in the CFH between races: Asian (3 [range 0–12]), Black (5 [range 1–15]), Other (3 [range 0–11]), and White (4 [range 0–13]) ($P = 0.04$). On multivariable linear regression analysis, controlling for age, personal cancer history, family cancer history, race, and prior genetic testing, family history of cancer was associated with a greater number of relatives included in the CFH ([Supplementary Table 1](#)).

3.4. Predictors of referral to genetic counseling

With the overarching goal of identifying high-risk patients for genetic assessment, we evaluated for predictors of referral to genetic counseling. Patients with a family history of cancer were more likely to be referred to genetic counseling (28 [23.5%] vs. 2 [4.4%], $P = 0.003$). Patients with a greater median number of included relatives were also more likely to be referred to genetic counseling (4.5 [range 1–15] vs. 3 [range 0–11], $P = 0.003$). Among the established quality measures for CFH, inclusion of the following were associated with increased likelihood of referral to genetic counseling: at least three generations (20 [29.4%] vs. 10 [11.8%], $P = 0.008$), relatives' lineage (24 [28.2%] vs. 6 [8.8%], $P = 0.004$) and age of relatives' death (5 [50.0%] vs. 25 [17.5%], $P = 0.026$).

4. Discussion

Our study highlights areas for improvement in CFH collection for patients presenting to a gynecologic oncology outpatient clinic. There are approximately four million individuals in the U.S. with a deleterious mutation in a cancer-associated gene but fewer than 20% are aware of their underlying genetic condition and cannot take advantage of

Table 1
Patient Demographics (N = 200).

Age (median, range)	52 (23–93)	
	N	%
Race		
Asian	22	11.0
Black or African American	20	10.0
Other	28	14.0
White	90	45.0
Data not available	40	20.0
Ethnicity		
Hispanic or Latino	14	7.0
Not Hispanic or Latino	127	63.5
Data not available	59	29.5
Marital Status		
Married	26	13.0
Single	16	8.0
Divorced/Separated	4	2.0
Widowed	3	1.5
Data not available	151	75.5
Children		
None	91	45.5
≥ 1	90	45.0
Data not available	19	9.5
Personal history of cancer diagnosis		
Yes	63	31.5
Breast	21	10.5
Ovarian/fallopian tube	8	4.0
Uterine	16	8.0
Colorectal	7	3.5
Melanoma	2	1.0
Other	18	9.0
No	137	68.5
Reason for new patient appointment		
Pelvic mass	54	27.0
Cervical or vulvar dysplasia	27	13.5
Thickened endometrium	25	12.5
Genetic mutation	20	10.0
Postmenopausal bleeding	10	5.0
Abnormal uterine bleeding	10	5.0
Endometrial cancer	26	13.0
Ovarian cancer	8	4.0
Cervical cancer	6	3.0
Vulvar cancer	3	1.5
Vaginal cancer	1	0.5
Family history of cancer	4	2.0
Other	10	5.0
Prior genetic testing		
Tested	43	21.5
Not tested	156	78.0
Uncertain if prior testing	1	0.5
Results of prior genetic testing		
Pathogenic variant detected	24	12.0
No pathogenic variant detected	16	8.0
Inconclusive results	3	1.5
Referral to genetic counseling/testing after visit		
Yes	30	15.0
No	136	68.0
No due to prior testing	34	17.0
Completion of genetic counseling after referral		
Yes	19	63.3
No	11	36.7
Completion of genetic testing after referral		
Yes	16	53.3
No	14	46.7

genetically targeted preventative cancer care (Childers et al., 2018). Collection of accurate and comprehensive CFH is invaluable for identifying at-risk patients for triage to genetic counseling and testing. However, prior literature suggests that the current system of CFH collection results in data inadequate to accurately assess disease risk (Acton et al., 2000; Murff et al., 2007; Summerton and Garrod, 1997). We found, in a population of patients presenting for a gynecologic oncology appointment, that CFH often lacks important quality elements.

In our cohort, fewer than 50% of patients had documentation of at

least three generations of relatives, pertinent negatives, relatives' cause of death, relatives' age at death and relatives' age at cancer diagnosis, all elements of a high quality CFH previously established by the genetics community (Committee on Practice Bulletins–Gynecology CoG, Society of Gynecologic Oncology, 2017; Bennett, 2004; ACOG Practice Bulletin No, 2014). Patient age, ethnicity, and personal cancer history were not significantly associated with the quality of collected CFH. Not surprisingly, patients who were aware of a prior diagnosis of cancer in their family provided information on a greater number of relatives and were more likely to provide information on relatives' gender, relatives' lineage, pertinent negatives, and relatives' cause of death. Cancer diagnoses in families may prompt greater awareness and discussion among patients and their relatives. However, patients who are unaware of cancer diagnoses in their families could also benefit from a dedicated discussion of this topic with family members.

Patients with prior genetic testing provided information on a greater median number of relatives and were more likely to include information on relatives' lineage, three generations, pertinent negatives, and age of relatives' cancer diagnosis. It seems plausible that patients with prior genetic testing previously underwent genetic assessment with a genetics provider, prompting in-depth discussion of family cancer history.

The goal of collecting an accurate and sufficiently comprehensive CFH is to identify patients who will benefit from genetic counseling and/or testing and make appropriate referrals. In our population, patients were more likely to be referred to genetic counseling when their CFH included a greater number of relatives, indicating that the documentation of essential elements of CFH is instrumental to identification of these high-risk individuals for genetics assessment.

This study has important limitations. As a retrospective analysis, it cannot identify which barriers to CFH collection were most critical. There are patient-related barriers including lack of awareness of relatives' health, inaccuracies in recall, poor family communication, and language barriers in the medical setting. Conversely, clinician-related factors include inadequate time to collect CFH, lack of standardization, and lack of training needed for providers to feel confident collecting CFH and transforming the data into an appropriate management plan (Sussner et al., 2011). There is no way to ascertain whether patient or clinician-related factors contributed more to deficiencies in documented CFH. Due to the small patient population, it also is difficult to fully assess the impact of race and ethnicity on CFH, but this is a topic that should be evaluated in future larger studies. Additionally, these data were collected prior to the COVID-19 pandemic. It remains to be seen how the emphasis on limiting a patient's in-person time with medical providers and on telemedicine will affect CFH collection.

Our data are consistent with the literature, suggesting that standard collection of family history may not adequately capture CFH (Wood et al., 2014; Murff et al., 2007; Sussner et al., 2011; Lanceley et al., 2012). We have planned a prospective randomized trial utilizing health information technology to collect CFH versus standard of care (face-to-face collection by a medical provider), as our data demonstrate that there is ample room for improvement. Health information technology has been shown to successfully improve clinical documentation, workflows, quality of care, patient safety, communication, and clinical decision support (Ritchie et al., 2020; Committee opinion no, 2015). With the growing recognition that it is critically important to identify individuals with cancer-associated pathogenic variants, medical providers must review the quality of the CFH they collect, as we have done in this study, and use these results to drive innovation in patient care and preventative medicine.

CRedit authorship contribution statement

Jenny Lin: Conceptualization, Data curation, Formal analysis, Investigation, Methodology, Project administration, Resources, Software, Supervision, Validation, Visualization, Writing – original draft, Writing – review & editing. **Isabel Wolfe:** Writing – review & editing.

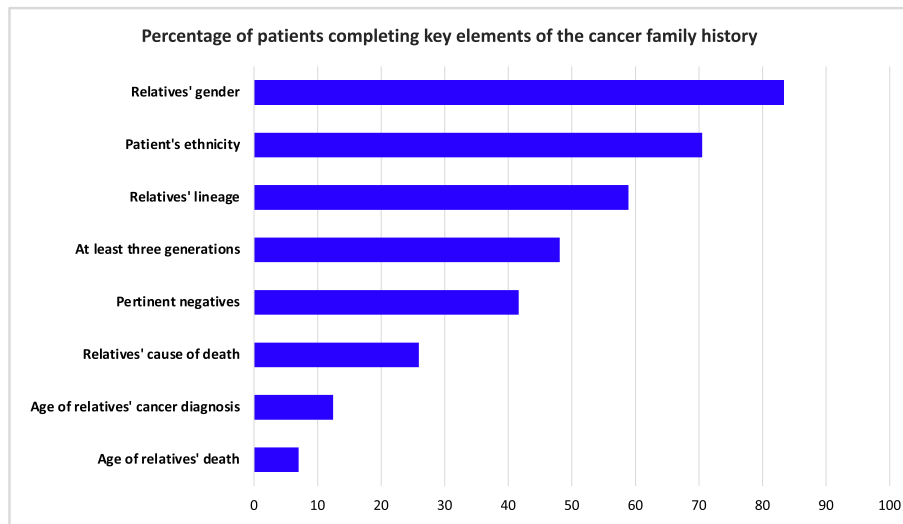


Fig. 1. Percentage of patients completing key elements of the cancer family history. The percentage of patients including ethnicity was calculated out of 200 patients. The percentage including age of relatives' cancer diagnosis was calculated out of 147 patients who included a relative with cancer in their cancer family history. All other percentages were calculated out of 185 patients with documented cancer family history.

Table 2
Univariate analysis for number of relatives included in the pedigree.

	Relatives included in the cancer family history		
	Median	Range	P Value
Age			0.302
< 65 years	4	0–15	
≥ 65 years	3	0–13	
Race			0.04
Asian	3	0–12	
Black	5	1–15	
Other	3	0–11	
White	4	0–13	
Ethnicity			0.465
Hispanic/Latino	4	0–11	
Non-Hispanic/Latino	3.5	0–15	
Personal history of cancer			0.813
No	3	0–15	
Yes	3	0–13	
Family history of cancer			<0.001
No	2	0–6	
Yes	4	1–15	
Prior genetic testing			0.012
No	3	0–15	
Yes	4	0–13	

Muhammad Danyal Ahsan: Data curation, Writing – review & editing. **Hannah Krinsky:** Conceptualization, Data curation, Investigation, Methodology, Writing – review & editing. **Andreas I. Lackner:** Data curation, Investigation, Methodology, Writing – review & editing. **Joe Pelt:** Data curation, Writing – review & editing. **Karen Bolouvi:** Data curation, Investigation, Methodology, Validation, Writing – review & editing. **Charlotte Gamble:** Investigation, Methodology, Writing – review & editing. **Charlene Thomas:** Formal analysis, Investigation, Methodology, Validation, Writing – review & editing. **Paul J. Christos:** Formal analysis, Investigation, Methodology, Validation, Writing – review & editing. **Evelyn Cantillo:** Investigation, Methodology, Writing – review & editing. **Kevin Holcomb:** Investigation, Methodology, Writing – review & editing. **Eloise Chapman-Davis:** Investigation, Methodology, Writing – review & editing. **Ravi Sharaf:** Conceptualization, Data curation, Investigation, Methodology, Writing – review & editing. **Steven M. Lipkin:** Investigation, Methodology, Writing – review & editing. **Stephanie V. Blank:** Investigation, Methodology, Supervision, Writing – original draft, Writing – review & editing. **Melissa K. Frey:** Conceptualization, Data curation, Formal analysis, Investigation, Methodology,

Project administration, Resources, Software, Supervision, Validation, Visualization, Writing – original draft, Writing – review & editing.

Declaration of Competing Interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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Appendix A. Supplementary data

Supplementary data to this article can be found online at <https://doi.org/10.1016/j.gore.2022.100941>.

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