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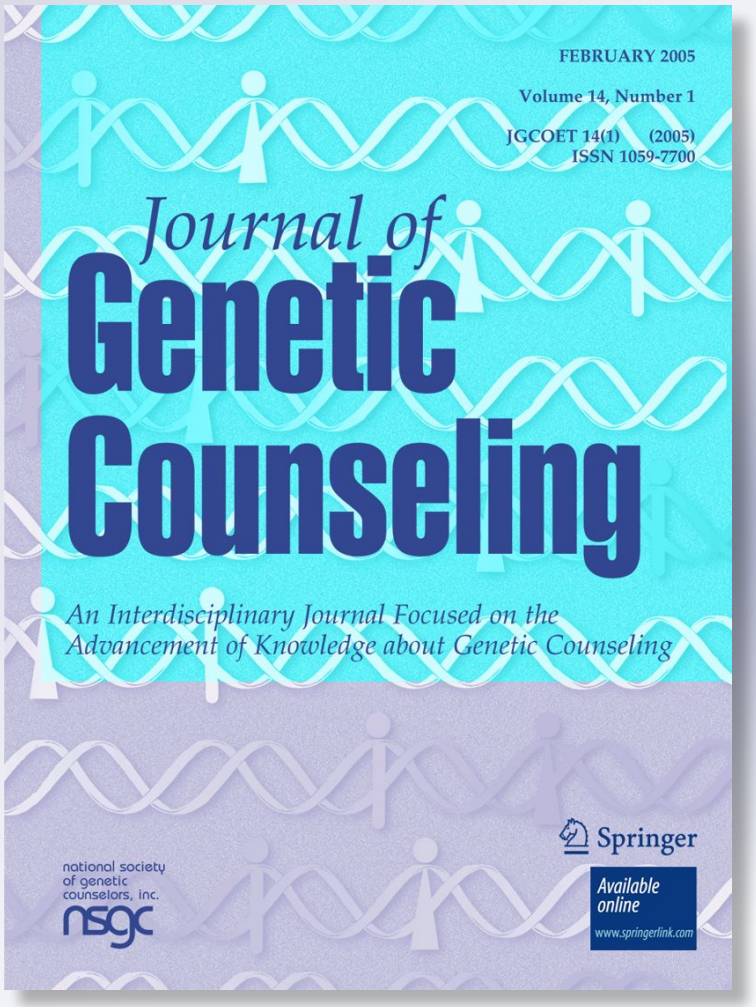

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ORIGINAL RESEARCH

Bias in the Reporting of Family History: Implications for Clinical Care

Elissa M. Ozanne · Adrienne O'Connell · Colleen Bouzan · Phil Bosinoff · Taryn Rourke · Dana Dowd · Brian Drohan · Fred Millham · Pat Griffin · Elkan F. Halpern · Alan Semine · Kevin S. Hughes

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Abstract Family history of cancer is critical for identifying and managing patients at risk for cancer. However, the quality of family history data is dependent on the accuracy of patient self reporting. Therefore, the validity of family history reporting is crucial to the quality of clinical care. A retrospective review of family history data collected at a community hospital between 2005 and 2009 was performed in 43,257 women presenting for screening mammography. Reported numbers of breast, colon, prostate, lung, and ovarian cancer were compared in maternal relatives vs. paternal relatives and in first vs. second degree relatives. Significant reporting differences were found between maternal and paternal family history of cancer, in addition to degree of relative. The number of paternal family histories of cancer was significantly lower than that of maternal family histories of cancer. Similarly, the percentage of grandparents' family histories of cancer was significantly lower than the percentage of parents' family histories of cancer. This trend was found in all cancers except prostate cancer. Self-reported family history in the community setting is often influenced by both bloodline of the cancer history and the degree of relative affected. This is evident by the under-reporting of paternal family histories of cancer, and also, though to a lesser extent, by degree. These discrepancies in reporting family history of cancer imply we need to take more care in collecting accurate family histories and also in the clinical management of individuals in relation to hereditary risk.

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Introduction

The identification of patients with hereditary cancer susceptibility is critical for both the prevention and early detection of many cancers. The first step in recognizing patients at high risk is the collection, recording, and evaluation of family history data. While a family history of cancer is highly predictive of an individual's susceptibility, this information collected in typical clinical settings is known to be imprecise (Wilson et al. 2009). The most common sources of inaccuracy include patient reporting errors, clinician querying and recording errors, and systemic factors.

Patient reporting errors typically stem from limited knowledge of their family history, or errors in interpreting or reporting family history information. These errors are known to vary based on patient factors such as age, education, type of cancer and degree of urbanization (Abraham et al. 2009). The accuracy of patient-reported family history data has been shown to vary by bloodline for gender specific cancers such as breast and ovarian cancers (Quillin et al. 2006) and also by degree of relative (Ziogas and Anton-Culver 2003). Quillin et al. found that patient-reported family history of breast cancer in an urban Women's Health Clinic is likely to underrepresent paternal cancer histories. Similarly, Ziogas and Anton-Culver conclude that the degree of relationship predicted underreporting of cancer incidence in a large family registry of cancer at an academic medical center, with the higher degree of relative predicting greater underreporting.

Clinician querying and recording errors are typically related to the failure to collect family history data, incomplete understanding of hereditary diseases and related syndromes, and biases in collecting data for only a subset of specific family members or towards one bloodline (clinicians have been shown to probe more deeply into maternal than paternal family history in standardized patients with breast cancer risk (Burke et al. 2009)).

Systemic factors that can lead to incomplete or inaccurate family history data include the lack of time for data collection (Ozanne et al. 2009b), the absence of effective family history functionality within Electronic Health Records (EHRs) (Drohan et al. 2009; Ozanne et al. 2009a), and the lack of standard approaches to data collection and recording (Acheson et al. 2006).

Together, these factors constitute significant barriers to the accurate collection of family history data and result in missed opportunities for the prevention and early detection of cancer., The vast majority of studies examining the accuracy of family history collection has been focused on single cancers (i.e. familial breast cancer), conducted using demonstration patients in an educational setting, or relied on cancer registry data (Wilson et al. 2009). Because these studies cannot be easily generalized to broader, real-world clinical settings, we sought to better understand the accuracy of reporting family history of the most common cancers in a large community hospital setting. Rather than use the approach of comparing recorded family history against a more detailed review of actual records of family members, we opted to compare reported rates of cancer in maternal relatives vs. paternal relatives, and in first vs. second degree relatives, assuming the numbers should be similar.

Methods

At the Newton Wellesley Hospital, every woman presenting for a mammogram is interviewed by the mammography technologist to collect family history. The data is entered real-time into HughesRiskApps Software (www.HughesRiskApps.com, Boston Massachusetts) (2010) as part of routine patient care. This software allows the mammogram technologist to enter a patient's family history of cancer and risk analyses using the established models (BRCAPRO, Myriad, Claus and Gail) are run immediately. HughesRiskApps was designed to collect family history and other risk factor information, to store this data in an easily updated format, to provide clinicians with the necessary information to identify and manage risk, and to streamline the counseling process. This system can be used in many settings including primary care, breast imaging centers, or similar settings to identify patients who may benefit from a referral to a specialized risk assessment and counseling appointment. HughesRiskApps is equipped with two tiers of patient surveys: the "Standard Survey," designed to efficiently identify potentially high risk patients in a mammography imaging center or the primary care setting; and the "Risk Clinic Survey," designed to fine-tune the risk assessment and develop a management plan for patients seen at cancer risk clinics. The Standard Survey was used in the current study. This software is also described in more detail elsewhere (Drohan et al. 2009; Ozanne et al. 2009b).

With Institutional Review Board (IRB) approval, a retrospective review was performed of cancer family histories collected between October 12, 2005 and March 10, 2009. Within this large community sample, we examined whether the presence of a family history of common cancers was similar across bloodlines (paternal vs. maternal) and degree of relative (first vs. second degree). Data were retrospectively reviewed to compare rates of lung, colon, breast, ovarian, and prostate cancer across maternal and paternal bloodlines, and between generations. To minimize reporting bias, the sample was limited to women who had no personal history of breast cancer. Because the community medical center serves an ethnically homogeneous population, the sample

was limited to Caucasian women. The study sample was further split into two groups: women who self reported to be Caucasian non-Jewish women and those self reported to be Caucasian Jewish women to explore possible differences in cancer awareness.

We hypothesized that reported rates of these cancers should be the same between the bloodlines in the absence of reporting biases. Because the incidence of cancer increases with age, even in a population of mixed ages, we also hypothesized that the reported rates of cancer in older generations should be greater than in younger generations. We further hypothesized that this trend would be less pronounced in the Jewish population (Mogilner et al. 1998). Within the study population, we analyzed the data to determine if the reported rate of cancer in maternal relatives was different than that in paternal relatives. Similarly, we analyzed the rate of cancer in grandparents as compared to the rate of cancer in parents based on the expected number of cancers in each group given the size of each group (2 parents vs., 4 grandparents), and for aunts and uncles as compared to parents. As cancer history in aunts and uncles was recorded, but the number of unaffected aunts and uncles was not recorded, we made very conservative assumptions using national census data regarding the average number of aunts and uncles (1 aunt and one 1 uncle per side of the family) per family and tested this assumption in our statistical analyses (2006–2008 U.S. Census Bureau 2010). These data were analyzed using Chi-Square statistical tests to compare for reporting accuracy.

Results

During the study period, family histories were collected on 43,257 women. Ethnicity was reported for 82.3% of these women (Table 1). The age distribution is shown in Fig. 1. Of these women, 31,115 (72%) were self-identified as Caucasian women with no prior history of breast cancer. Twenty-six thousand four hundred and two (61%) of these women self-identified as non-Jewish women and 4,713 (11%) self-identified as Jewish.

Table 1 Reported demographics for sample population

Race	Count	%
Total	43,257	
Caucasian	33,703	77.9%
Unknown	7,674	17.7%
Asian or Pacific Islander	948	2.2%
African American	472	1.1%
Other	300	0.7%
Hispanic	117	0.3%
American Indian/Aleutian/Eskimo	41	0.1%
Caribbean/West Indian	2	0.0%

Given the size of the sample in this study, there should be no discernable difference seen in the rates of cancer by bloodline. However, when examining the Caucasian non-Jewish population, the reported rates of paternal cancer were significantly lower than the rates of maternal cancer. Similarly, the rates of cancer in older generations should be significantly higher than the rates of cancer in younger generations. However, our study found the opposite result. In this study, the reported rates of cancer among grandparents were significantly lower than the reported rates of cancer among parents. This trend was also seen in comparing the rate of cancer in parents and within the aunts and uncles, a result which held for all assumptions regarding the average number of aunts and uncles per family. When examining the Jewish population, the general trend of underreporting was also seen, though to a lesser degree. Together, these results indicate a systematic bias in the reporting of cancer family history that was present across all cancers examined in this study other than prostate cancer. The full results are presented in Tables 2 and 3 and corresponding Figs. 2, 3, 4, 5 and 6 and are described by main result below.

Result 1: Maternal vs. Paternal

In our study sample, the number of cancers within the study population are consistently reported to be higher in maternal relatives as compared to the paternal relatives, representing a reporting bias when comparing maternal with paternal bloodlines. This result was statistically significant in all cancers except prostate cancer for both the Jewish and non-Jewish sub-populations.

As presented in Table 2, maternal second degree relatives within the Caucasian non-Jewish population had higher reported number of cancers than paternal second degree relatives for lung cancer (921 vs. 676), colon cancer (1,125 vs. 715), breast cancer (4,698 vs. 2,914), and ovarian cancer (624 vs. 371). However, when looking at the data collected for prostate cancer, we see similar numbers in the maternal and paternal second degree relatives at 239 and 240 respectively.

When looking at the Jewish population, a consistently higher number of cancers were reported in the maternal second degree relatives than the paternal second degree relatives (Table 3). However, when looking at specific comparisons of second degree relatives, fewer discrepancies are found than in the non-Jewish population. For example, in lung cancer, there were no significant differences between the number of reported cancers in the maternal grandfathers and paternal grandfathers (48 vs. 42) and similarly for the grandmothers (42 vs, 26). In colon cancer, similar findings were found in the grandfathers (70 maternal vs. 57 paternal), the aunts (36 maternal vs. 27 paternal), and the uncles (35

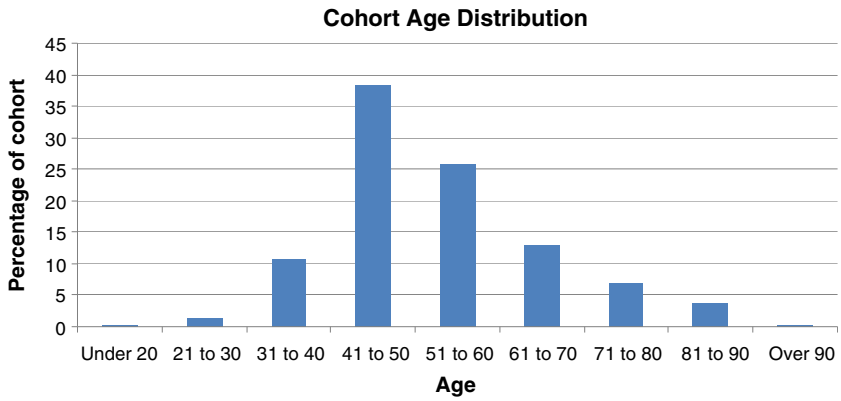

Fig. 1 Age distribution of cohort

maternal vs. 24 paternal). In ovarian cancer, this was also seen with the aunts (47 maternal vs. 39 paternal).

Result 2: Generational Effects (Parents vs. Grandparents)

Because the rate of cancer increases with age, one would expect to see a significantly higher rate of cancer in grandparents as compared to the rate of cancer in parents. Therefore, the rate of parental cancers is expected to be much less than that of the grandparents. However, the reported rates of cancers in the older generations (grandparents) are consistently lower than younger generations (parents) in all cancer types.

Within the Caucasian non-Jewish population, the reported number of cancers in the parents is higher than that reported in the grandparents in all cancers: lung cancer (1,605 vs. 780), colon cancer (1,447 vs. 1,036), breast cancer (2,904 vs. 2,719), ovarian cancer (443 vs. 440), and prostate cancer (1,201 vs. 269). This trend is found in the Jewish population also for lung, colon, and prostate cancers. Interestingly, this trend was not found for breast or ovarian cancers. The number of reported cancers was greater in the grandparents than in the parents (738 vs. 625 breast, 86 vs. 77 ovarian), though these rates are still much less than what you would expect given there are twice as many individuals at risk.

Result 3: First Degree vs. Second Degree Relatives (Parents vs. Aunts and Uncles)

Based on US Census data, individuals on average have an equal or greater number of aunts and uncles than parents

Table 2 Reported number of cancers by relative for non-Jewish Caucasian women

Total patients	26,402									
	Lung		Colon		Breast		Ovarian		Prostate	
Father	985	$p<0.0001$	799	$p<0.0001$	31		0		1,201	
Mother	620		648		2,873		443		0	
Paternal Uncle	204	NS	154	$P=0.0011$	12		0		118	NS
Maternal Uncle	239		229		12		0		92	
Paternal Aunt	149	$p<0.0001$	158	$p<0.0001$	1,822	$p<0.0001$	208	$p<0.0001$	0	
Maternal Aunt	225		263		3,047		347		0	
Paternal Grandfather	211	$P=0.002$	176	$p<0.0001$	18	$P=0.047$	0		122	NS
Maternal Grandfather	278		287		32		0		147	
Paternal Grandmother	112	$p<0.0001$	227	$p<0.0001$	1,062	$p<0.0001$	163	$p<0.0001$	0	
Maternal Grandmother	179		346		1,607		277		0	
All Paternal Second Degree Relatives	676	$p<0.0001$	715	$p<0.0001$	2,914	$p<0.0001$	371	$p<0.0001$	240	NS
All Maternal Second Degree Relatives	921		1,125		4,698		624		239	
All Parents	1,605		1,447		2,904		443		1,201	
All Grandparents [#]	780	$p<0.0001$	1,036	$p<0.0001$	2,719	$p<0.0001$	440	$p<0.0001$	269	$p<0.0001$
All Aunts/Uncles [#]	817	$p<0.0001$	804	$p<0.0001$	4,893	$p<0.0001$	555	$P=0.0004$	210	$p<0.0001$

[#] Comparison to cancers reported in all parents

NS not significant

Table 3 Reported number of cancers by relative for Jewish Caucasian women

Total patients	4,713									
	Lung		Colon		Breast		Ovarian		Prostate	
Father	137	NS	166	$p=0.024$	8		0		227	
Mother	140		128		617		77		0	
Paternal Uncle	11	0.002	24	NS	0		0		11	$p=0.039$
Maternal Uncle	31		35		1		0		23	
Paternal Aunt	23	$P=0.031$	27	NS	281	$p<0.0001$	39	NS	0	
Maternal Aunt	40		36		456		47		0	
Paternal Grandfather	42	NS	57	NS	5	NS	0		30	$p=0.039$
Maternal Grandfather	48		70		8		0		29	
Paternal Grandmother	26	NS	54	$P=0.008$	222	$p<0.0001$	28	P0.036	0	
Maternal Grandmother	42		85		322		46		0	
All Paternal Second Degree Relatives	102	$P=0.0002$	162	$P=0.001$	508	$p<0.0001$	67	$P=0.039$	41	NS
All Maternal Second Degree Relatives	161		226		787		93		52	
All Parents	277		294		625		77		227	
All Grandparents [#]	105	$p<0.0001$	122	$p<0.0001$	738	$p<0.0001$	86	$p<0.0001$	34	$p<0.0001$
All Aunts/Uncles [#]	158	$p<0.0001$	266	$p<0.0001$	557	$P=0.0015$	74	NS	59	$p<0.0001$

[#] comparison to cancers reported in all parents

NS not significant

(2006–2008 U.S. Census Bureau 2010). Accordingly, it is expected that among an individual's aunts and uncles, the incidence of cancer would be similar or higher than that of an individual's parents. However, when looking at our data, we see the reported numbers of cancer in aunts/uncles are consistently lower than that of parents, except for breast and ovarian cancers: for lung cancer (817 vs. 1,605), colon cancer (804 vs. 1,447), breast cancer (4,893 vs. 2,904), ovarian cancer (555 vs. 443), and prostate cancer (210 vs. 1,201). In the Jewish population, all reported rates for parents are greater than the reported rates in the aunts and uncles.

Discussion

On a population basis, the rates of cancer should not differ between maternal and paternal relatives (Quillin et al. 2006). However, studies have indicated that paternal relatives are often underreported in various cancers when compared to maternal relatives (Green et al. 1997; Tinley and Lynch 1999). The results of our study confirm this finding for a large community population, across numerous common cancers. However, in the Jewish population, there were fewer discrepancies in the reported rates for some second

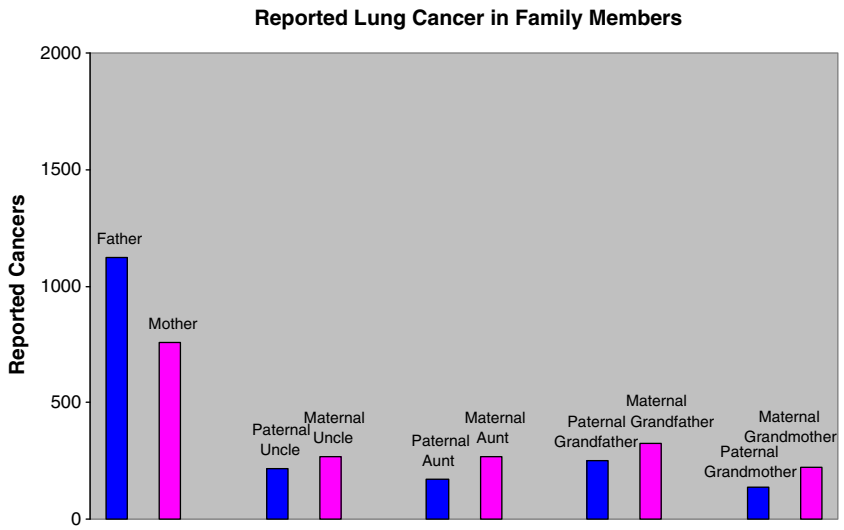

Fig. 2 Reported lung cancer by relative for total study population (Caucasian Jewish and non-Jewish)

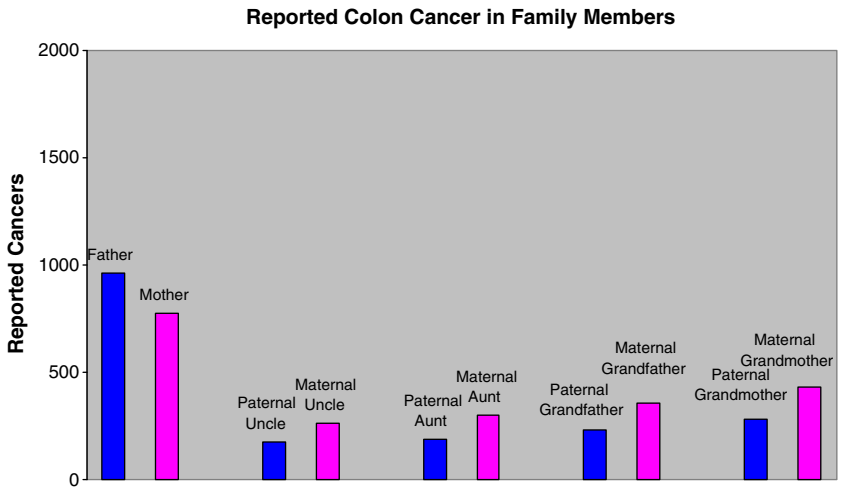

Fig. 3 Reported colon cancer by relative for total study population (Caucasian Jewish and non-Jewish)

degree relatives, and for breast and ovarian cancers in the closest generation (aunts/uncles vs. parents). Both findings carry significant practice implications. The family history of cancer in maternal lines is more highly reported in comparison to paternal lines for lung, colon, breast, and ovarian cancer. However, this result did not hold true for prostate cancer, which we cannot explain.

When comparing the data collected between first and second degree relatives, we see that there is a higher degree of cancer family history among first degree relatives. Again we see a bias in the reporting as it would be assumed that the second degree relatives would have an increased cancer history solely based on their age; grandparents are significantly older than parents and therefore are assumed to have increased incidence rates of cancer within the general population. Similarly, as the number of aunts and uncles is expected to be greater than the number of parents an individual has, it would stand to reason that the number of these relatives reported to have cancer should be higher in these second degree relatives. We found the opposite to be true. This bias is most likely due to the fact that the quality of reporting worsens with increasingly distant relatives (Couto and Hemminki 2007). These analyses demonstrate that family history of cancer is significantly underreported in paternal lines and significantly underreported in second degree relatives.

It is possible that these results suggest an over reporting of maternal cancer histories rather than underreporting of paternal cancer histories. However, this had been shown to occur very rarely in the literature and is not a likely explanation of the result of this study (Orom et al. 2008; Wilson et al. 2009). For populations such as the Jewish populations, where particular cancers are more common (breast and ovarian) (King et al. 2003), there appears to be fewer discrepancies in reporting, though still a significant underreporting despite the outreach that occurs within this population (Antman et al. 2002). There are a number of possible explanations for this finding. One is that cancers that are common are more readily reported, another is that that community education and awareness can improve the self report of cancer history.

These biased results in the reporting of family history of cancer can possibly be explained in several ways. First is the possibility of patient reporting errors. The reporting of

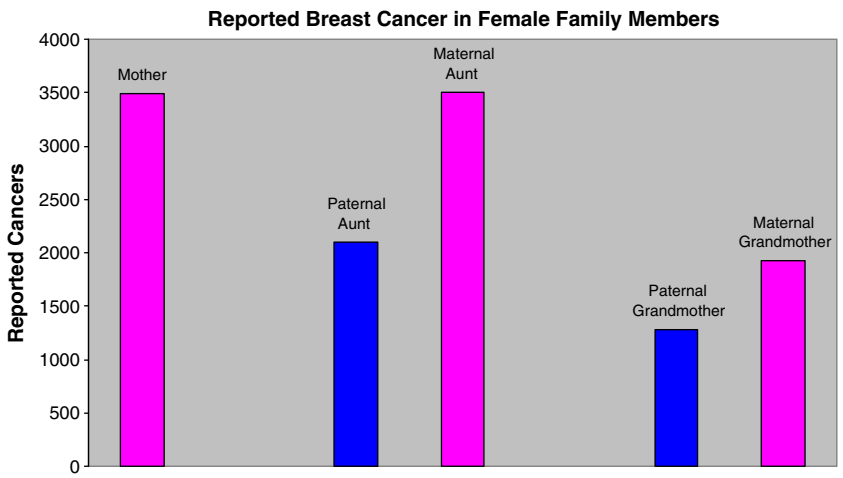

Fig. 4 Reported breast cancer by relative for total study population (Caucasian Jewish and non-Jewish)

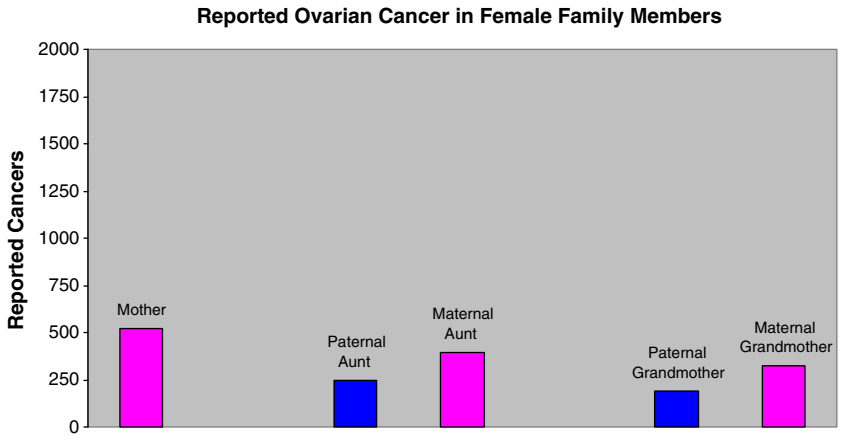

Fig. 5 Reported ovarian cancer by relative for total study population (Caucasian Jewish and non-Jewish)

family history of cancer can be subject to a number of patient reporting inaccuracies and biases. Patients with heightened awareness of a particular cancer, often due to personal experience of cancer diagnosis in themselves or a close relative, tend to report a more significant family history for that cancer than control subjects (Hughes et al. 2003; Mitchell et al. 2004; Dominguez et al. 2005; Jones et al. 2005; Chang et al. 2006). Conversely, social stigma may play a role in decreased patient reporting, causing a patient to be less aware of cancers in the family and thus unable to completely report a positive cancer family of certain types of cancers. For example, it has been shown that often people are embarrassed or saddened to discuss a diagnosis of cancer with other members of their family (Mitchell et al. 2004).

Studies suggest that underreporting is far more common than over reporting, with sensitivities ranging from 50 to 98% depending on cancer site (Orom et al. 2008). Quillin et al. report that the odds of reporting a maternal family history of breast cancer was close to two times greater than the odds of reporting a paternal family history of breast cancer (Quillin et al. 2006), a result that is similar to those of our study. The explanations of this finding include excessive reporting of breast cancer from the maternal side, failure to adequately communicate breast cancer risk by male relatives, underreporting of paternal family history, and a general inadequate reporting of family histories (d'Agincourt-Canning 2001; Tercyak et al. 2001; Forrest et al. 2003; Quillin et al. 2006). Biases in family reporting of disease may also be more prevalent among older generations (Mai et al. 2011) and certain ethnic groups (Orom et al. 2010). This unawareness may present a barrier to prevention behaviors, thus, contributing to unnecessary cancer morbidity and mortality (Ramsey et al. 2006; Orom et al. 2010).

A second possible explanation for the observed biases in family history reporting is clinical recording errors. When determining the accuracy of cancer family history collection, clinician errors and bias can play a significant role. Upon questioning of family physicians, it was found that most do not obtain a thorough family history, often due to lack of education regarding cancer syndromes or lack of time in a typical visit. It has been documented that family history was discussed during 51% of new patient visits and 22% of visits with established patients (Acheson et al. 2000). Physicians' rates of family history-taking varied from 0% to 81%, and it has been shown that patients are asked

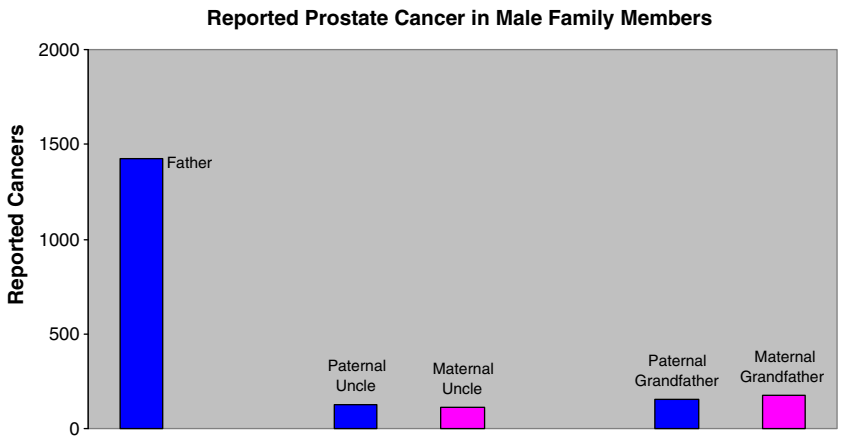

Fig. 6 Reported prostate cancer by relative for total study population (Caucasian Jewish and non-Jewish)

systematically less about paternal history than maternal history (Acheson et al. 2000; Burke et al. 2009). In our study, it is possible that there were biases introduced in the collection of the family history data. Radiology technologists assessed the family histories from women presenting at the mammography center. This setting could have easily biased the questions asked in terms of the type of cancers within the family, and it is possible that the technologists had similar biases to physicians and emphasized the maternal side.

Finally, systemic issues, such as time constraints and validity of screening tools can also introduce biases in the collection of cancer family history. Most physicians have limited time with each patient; in fact it was noted that the average duration of family history discussions were less than 2.5 min (Acheson et al. 2000). Collecting the information to draw a pedigree takes between 15 and 30 min, which is longer than the average face-to-face time for the doctor has for an entire primary care visit (Acheson et al. 2006). Ideally, once recorded, family history should be portable, private, yet accessible in various electronic medical record systems (Rich et al. 2004). Appropriate software and secure Internet technology can facilitate this goal, especially once there is widespread adoption of standard data formats for digitally communicating family history and cancer risk information (Shabo and Hughes 2005). Lacking systematic ascertainment, cancer risk counseling and management based on family history collection will require redundant, time consuming data collection and will therefore remain underutilized. More evidence is needed about the effects of using informatics to facilitate familial cancer risk assessment.

Limitations

While the results of our study are compelling, there are a few limitations that must be considered in the interpretation of these findings. First, our analyses did not verify the accuracy of the reporting by the patients beyond their initial assessment with the radiology technologists. Instead, we relied on comparisons of these reported histories to the expected averages within a large population and found discrepancies that could not have been accounted for by chance. Further, it is possible that the technologist had a particular bias in the way they elicited the family histories, and this could be explored in future studies. Another possible limitation is the setting of a breast screening setting. It is feasible our results would be different had this study taken place in a colorectal screening center, or a general clinic setting. However, given the robustness of our results across all cancers, we do not feel this bias was significant, if present at all.

Clinical Implications

Family history is one of the most important clinical tools in identifying patients at high risk for disease due to genetic susceptibilities and one of the most important tools for genetic counselors. Hence, having an adequate understanding of family history confers an opportunity to personalize and target disease, preferably through prevention (Yoon et al. 2002, 2004). Several clinical practice guidelines suggest that persons meeting family history criteria for specific cancers may benefit from particular screening programs and initiating screening at earlier ages compared with the general population (Ramsey et al. 2006). It is clear that family history of cancer is a valuable tool for reducing the societal burden of cancer if it is used appropriately in combination with risk assessment and genetic testing, in addition to the allocation of cancer prevention and screening services (Orom et al. 2008). However, given the results of this study that show a significant underreporting of paternal family histories of cancer and underreporting by increasing degree of relative, the full value of these services are not realized in clinical practice. To improve clinical outcomes, these possible biases must be accounted for when determining the validity of patients' family histories in clinical care.

In the field of genetic counseling, ensuring the accuracy of patient reported family history is critically important and understanding the common biases patients typically exhibit can guide clinicians in assessing more accurate family histories. Future research efforts need to be focused on identifying methods for which accurate and complete family histories can be collected efficiently. Further study of how to optimize this process of data collection is needed. Methods for accurate patient self-report need to be determined, as are methods that can increase the completeness of family histories that are elicited by clinicians. In addition, as data collection tools become more advanced, it needs to be studied how they can be designed to prompt elicitation of family histories in a more comprehensive manner. As the field of whole genome sequencing advances, it is possible that the importance of collecting family will decrease. However, we are far from this reality and it is unlikely that family history data will not be useful to understand family penetrance.

In all cancer types, with the exception of prostate cancer, the reported cancers in the maternal lineage is higher than the reported cancers in the paternal lineage. Although this is not consistent with basic genetics, this is a consistent theme throughout various studies and is thought to be due to several factors. While it is possible that genes inherited through males confer a low risk for cancer, research to date does not support this (Quillin et al. 2006). Another possible factor is over reporting from the maternal line. However, earlier studies that maintain high specificity of reported family history of cancer suggest that this explanation is

unlikely (Tinley and Lynch 1999). More likely, it is thought that men may be less likely to be told about cancer risk or to communicate that information with their family and relatives (d'Agincourt-Canning 2001; Tercyak et al. 2001; Forrest et al. 2003). This could also be due partly to unknown or unexpected paternity. And still age, education and socio-economic background can contribute to the under reporting of cancer family history in the paternal line. However, when examining the prostate cancer data, we see that maternal and paternal reported cancer family histories are in fact the same. This result is not understood, but suggests that there may be different social factors involved in this cancer, or could have been due to some unexplained bias within our sample.

Clinical tools, education and support for patients and clinicians designed to facilitate the accurate collection of family history are critical to advances in prevention, earlier diagnoses and treatment. Identifying the common systematic biases in reported family histories can focus these efforts in order to obtain the largest improvement possible. Efforts need to be made to educate men about the importance of family history and also to educate women to be more forthcoming with their male relatives regarding their family history. It is important for clinicians to be aware of these possible biases in the reporting of family history when caring for and treating patients.

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