Increased ease of access to genetic counseling for low-income women with breast cancer using a point of care screening tool

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Abstract

Increased access to genetic counseling services is of prime importance in minority and underserved populations where genetic testing is currently underutilized. Our study tested a point of care screening tool to identify high-risk low-income patients for genetic counseling in a busy county hospital oncology clinic. Eligible breast patients treated at a “safety-net” hospital, were scored into ‘high-risk’ (≥ 6) or ‘low-risk’ (< 6) groups using a screening tool on personal and family history of cancer. Genetic counseling and testing were provided at the Vanderbilt Hereditary Cancer Program (VHCP) to all ‘high-risk’ and some ‘low-risk’ participants considered to need genetic counseling by their oncologist. Ninety-nine women with a history of breast cancer were enrolled onto the study over a period of 26 months. 53.5% (53/99) had a ‘high-risk’ score and ethnic predominance of African-American (60.4%). Of these, 67.9% (36/53) were counseled, and 91.6% (33/36) tested with a 9% (3/33) mutation positive rate. In the ‘low-risk’ group, 28.2% (13/46) still met current NCCN guidelines and were referred by their oncologist. 69.2% (9/13) were counseled and tested. The ‘low-risk’ group of predominantly Caucasian (41.3%) participants carried a 20% (2/10) mutation positive rate; which was later adjusted to 10% to exclude a mutation not conferring a strong breast cancer risk. The screening tool was well accepted by patients; and increased access to genetic counseling. There was a subset of breast cancer affected women under 45 with no reported family history that failed to be identified. Minor alterations to the tool would enhance concordance with current NCCN guidelines.

Keywords Access to genetic services · Genetic counseling · Evaluation of genetic/genomic tool for public health · Low income · Minority population · Breast cancer

Introduction

It is common practice for cancer clinic staff to include the acquisition of family history in their intake process for patients. In order to identify high-risk patients and provide accurate family history assessment, evidence-based guidelines have been put forth by the National Comprehensive Cancer Network (NCCN) that are consistently used in clinical practice (Daly et al. 2017, Appendix 2). Recent literature indicates that fewer

than 1 in 5 patients that meet NCCN guidelines are referred for genetic counseling (Childers et al. 2017) or get genetic testing (Levy et al. 2011). This is even more evident in the lower socioeconomic (SEC) patient population. Studies have shown that women with a family income of $> \$150,000/\text{year}$ are 1.99 times more likely to undergo testing than women with incomes of $< \$30,000/\text{year}$ (Armstrong et al. 2005). African-American and Hispanic women are significantly less likely to receive *BRCA1/2* testing compared to non-Jewish white women (Armstrong et al. 2005) even when barriers of ascertainment and cost were absent (Susswein et al. 2008).

The United States Preventive Screening Task Force (USPSTF) recommends several screening tools to help capture cancer family histories. Some online assessment tools put forth in the USPSTF guideline are quite lengthy, and each have unique sections geared toward specific populations (Force 2005). The screening tool designed by Joseph et al. (2012) was specifically designed for ease of administration over the phone to identify high-risk women in the lower

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socioeconomic bracket, to increase access to genetic counseling. A validation of the screening tool in a population of mostly healthy, low-income women without a personal history of cancer compared favorably with other family history screening tools in specificity and sensitivity for accurate identification of high-risk women (Stewart et al. 2016).

The primary objective of our study was to evaluate this point-of-care screening tool to quickly and accurately identify breast cancer patients that may benefit from access to genetic counseling and testing services for a population of breast cancer affected, primarily minority, and low socioeconomic (SEC) population in a high-volume county hospital oncology clinic in Nashville, TN. We also report on some unique demographic characteristics of this population.

Methods

Clinical setting The study was conducted by Hereditary Cancer Program at Vanderbilt-Ingram Cancer Center (VHCP) in collaboration with the Robert E. Hardy Cancer Clinic at Nashville General Hospital (NGH), which is a “safety net” hospital that mainly serves un-insured or underinsured residents. Prior to the study, high-risk patients at NGH were referred for genetic counseling at VHCP by the oncologists’ identification and referral based on patient’s age of onset of cancer diagnosis. Patients referred were primarily identified on the personal history indications of NCCN criteria as it would apply during that time (Appendix 1). The study pilot was to evaluate all patients based on their personal history of breast and ovarian cancer and reported family history between August 2015 and October 2017.

Study participants and procedures Inclusion criteria included patients with a history of breast or ovarian cancer. Two participants with ovarian cancer were enrolled into the study, but only the results of the breast cancer cohort are reported here. Study participants were approached by a Research Coordinator to describe the study, and obtain consent. Following informed consent, the research nurse verbally administered a demographic form and the point of care screening tool. The Joseph et al.’s (2012) screening tool (Appendix 3) that was used asked 10 highly weighted questions regarding personal and family history of cancer. Each question was provided a unique score (Joseph et al. 2012). All answered questions were scored such that a score of 6 or higher would be considered ‘high-risk’ and trigger an automatic referral to a genetic counselor. Based on the point of care screening score, participants that met protocol guidelines were referred to the VHCP, a genetics specialty clinic located 2.5 miles from NGH.

Initially, prior to their VHCP genetic counseling appointments, participants were asked to complete a three-generational pedigree on a phone preappointment. On

completing the family history phone preappointment participants were provided a face-to-face appointment to meet with a genetic counselor at the VHCP. The majority of recruitment was completed in the first year with the counseling of enrolled patients in the second year. After the first 18 months of the study, due to significant lack of participant follow-through, the study procedure was altered to where the genetic counselor was housed onsite at NGH. Therefore, in the final 6 months of the study the genetic counselor was able to see participants within 2 weeks of enrolling in the study, without the phone preappointment. She captured the family history as a three-generational pedigree during the counseling appointment itself. Importantly, based on the medical oncologist’s clinical expertise, some ‘low-risk’ participants that scored lower than 6 but yet met NCCN criteria could also be referred for genetic counseling.

On completion of genetic counseling appointment, blood or saliva samples were sent to one of five major commercial genetic laboratories for testing. Genetic testing charges were billed to the participant’s insurance plan whenever possible. However, if the participant did not have eligible insurance coverage, the tests were billed to study funds. The majority of post-test appointments were scheduled in-person to disclose results with participants. A few participants requested phone call-outs for result disclosure.

Study measures and analysis Study data was collected and analyzed using RedCap electronic data capture tools hosted at Vanderbilt University (Harris et al. 2009). RedCap (Research Electronic Data Capture) is a secure, web-based application designed to support data capture for research studies, providing (1) an intuitive interface for validated data entry; (2) audit trails for tracking data manipulation and export procedures; (3) automated export procedures for seamless data downloads to common statistical packages; and (4) procedures for importing data from external sources. This study was approved by both the Vanderbilt University Medical Center and the Meharry Medical College Institutional Review Boards. This study was granted an exemption status from both institutional review boards.

Demographic data (Table 1) included age, ethnicity, education, religious affiliation, employment status, household income, and medical coverage. Screening tool questions centered on age at diagnosis, ER/PR/Her2 markers, and family history of breast and ovarian cancer (number of aunts/sisters/mother) on both sides of the family. Unique study identifiers were assigned to participants.

Results

Breast and ovarian cancer patients were approached for enrollment over a 2-year period, only 2 participants with

Table 1 Demographic data on 99 consented breast patients in ‘high-risk’ (score > 6) and ‘low-risk’ (score < 6) groups

	Overall	Low-risk (<6 score)	High-risk (>6 score)
Ethnicity:	(n = 99)	(n = 46)	(n = 53)
• African American	50 (50.5%)	18 (39.1%)	32 (60.4%)
• Caucasian	30 (30.3%)	19 (41.3%)	11 (20.8%)
• Hispanic	10 (10.1%)	4 (8.7%)	6 (11.3%)
• Asian	5 (5%)	4 (8.7%)	1 (1.9%)
• Other	4 (4%)	1 (2.2%)	3 (5.7%)
Age at Diagnosis	52.05 yrs	52.06 yrs	52.04 yrs
TNBC*	13/99 (13.1 %)	1/46 (2.1 %)	12/53 (22.6 %)
Metastatic Disease	10/99 (10.1 %)	6/46 (13.0 %)	4/53 (7.5 %)
Education:	(n = 77)	(n = 36)	(n = 41)
• < High school	16 (20.8%)	8 (22.2%)	8 (19.5%)
• High School	29 (37.7%)	11 (30.6%)	18 (43.9%)
• Some college	12 (15.6%)	4 (11.1%)	8 (19.5%)
• College	11 (14.3%)	6 (16.7%)	5 (12.2%)
• Graduate	4 (5.2%)	4 (11.1%)	0%
• No response	5 (6.5%)	3 (8.3%)	2 (4.9%)
Household Income:	(n = 78)	(n = 37)	(n = 41)
• < \$15,000	61 (78.2%)	29 (78.4%)	32 (78%)
• \$15,000 – \$25,000	7 (9%)	4 (10.8%)	3 (7.3%)
• \$25,000 - \$50,000	9 (11.5%)	3 (8.1%)	6 (14.6%)
• > \$50,000	1 (1.3%)	1 (2.7%)	0%
Medical Coverage:	(n = 93)	(n = 41)	(n = 52)
• Medicaid (TennCare)	76 (81.7%)	33 (80.5%)	43 (82.7%)
• Medicare	6 (6.5%)	2 (4.9%)	4 (7.7%)
• Private Insurance - Self	5 (5.4%)	2 (4.9%)	3 (5.8%)
• Private Insurance - Emp	5 (5.4%)	3 (7.3%)	2 (3.8%)
• No insurance	3 (3.2%)	1 (2.4%)	2 (3.8%)
Employment Status:	(n = 80)	(n = 37)	(n = 43)
• Unemployed	28 (35%)	13 (35.1%)	15 (34.9%)
• Employed full-time	19 (23.8%)	8 (21.6%)	11 (25.6%)
• Employed part-time	12 (15%)	8 (21.6%)	4 (9.3%)
• Disability	13 (16.3%)	6 (16.2%)	7 (16.3%)
• Retired	6 (7.5%)	2 (5.4%)	4 (9.3%)
• Other (home-maker, student)	3 (3.6%)	1 (2.7%)	2 (4.6%)

Bolded entries are main demographic categories in which data was collected. Unbolded are sub-categories under the main ones

*TNBC = Triple Negative Breast Cancers

ovarian cancer were enrolled. The data reported herein applies only to participants with a history of breast cancer. Data collected on the first 90 potential participants showed a 90% (81/90) enrollment rate. Ultimately, 99 breast cancer participants were eligible and consented to the study. Overall demographics of the group showed 50.5% were African-American and 30.3% Caucasian; 35% were unemployed with an additional 23.75% either retired or on disability. Relating to healthcare coverage, 81.7% of participants had Medicaid (TennCare). 78.2% reported an annual

household income under \$15,000, and 58.5% were either high-school educated or below (Table 1).

On completing the screening tool, 53.5% (53/99) women met a “high-risk” criterion (scored > or = 6) and 46.4% (46/99) were “low-risk” (score < 6). On comparing the two risk groups, 60.4% of the “high-risk” group were African-Americans with 20.8% being Caucasians as compared to 39.1% African-Americans and 41.3% Caucasians in the “low-risk” group (Table 1). The correlation between ethnic group and risk stratification was not statistically significant

using chi-square analysis. In terms of disease characteristics, both the ‘high-risk’ and ‘low-risk’ groups were comparable for age at diagnosis (52.04 vs 52.06 years). The majority of participants with a history of triple negative breast cancer were in the ‘high-risk’ group (22.6%), but a larger percentage with metastatic disease were in the ‘low-risk’ group (13%).

Clinic compliance data indicates that all 53 ‘high-risk’ patients were referred for genetic counseling and contact was attempted for scheduling. Thirty-two percent (17/53) were unable to be reached by phone or failed to keep their appointment (Fig. 1). A total of 36 participants received genetic counseling; 27 at the VHCP location, 1 was seen under study protocol at an outside facility, and 8 were seen after the counselor started at NGH. In the ‘low-risk’ group, 28.2% (13/46) still met NCCN guidelines and were referred for genetic counseling. 69.2% (9/13) participants completed appointments, and 30.7% (4/13) were lost to follow-up (Fig. 1) Of the 9 patients, two were seen after the genetic counselor started at NGH.

A significant proportion of patients undertook testing in both categories (Table 2). In the ‘high-risk’ group,

91.6% (33/36) patients followed through with genetic testing. Of the three that refused—one patient cited concerns over the Genetic Information Non-Discrimination Act (GINA) law, another said that her family was not interested and did not want her to undertake testing, and one patient mentioned religious reasons to decline. In the ‘low-risk’ group, a total of 10 participants were tested—9 that were identified as ‘low-risk’ per the screening tool yet referred for genetic counseling based on current NCCN criteria and one additional participant who was older in age but tested by her medical oncologist due to her aggressive, triple negative disease. Mutation detection was 9% (3/33) in the ‘high-risk’ group versus 20% (2/10) in the ‘low-risk’ group, which was later adjusted to 10%. This adjustment was made to confine the analysis to reflect genes primarily involved in breast cancer risk. Since one of the identified mutations in the low-risk group was SDHD, and SDHD is not a strong breast cancer risk gene, it was excluded from later analysis of the mutation rate in the ‘low-risk’ group.

Fig. 1 Flow-chart of study using the point-of-care screening tool for personal and family history of breast cancer. *One add-on patient did not meet NCCN criteria

Table 2 Genetic testing profile in our patient population. Overall data in the high risk (>6) and low-risk (<6) groups

Score	Referred	Counseled	Tested	Mutation Positive	VUS	Mutation Negative
High-risk (>6 score)	53/53 (100%)	36 (67.9%)	33 (91.6%)	3 (9%) ATM, BRCA1, PALB2	7	21
Low-risk (<6 score)	13/46 (28.3%)	9 (69.2%)	10*	2 (20%) -adjusted to 1 (10%)+ BRCA2, SDHD	1	7

*Number tested is higher than number counseled as one patient who did not meet NCCN guidelines was tested out of office

+ The adjustment to 10% is to reflect the genes primarily involved in hereditary breast cancer risk assessment. Since SDHD is not one such gene, it was excluded from the final analysis of the mutation positive rate

VUSs identified were in the BRCA2, NBN, RAD51D, SMARCA4, ATM, POLD1, APC genes

Discussion

The primary objective of our study was to examine whether a simple screening tool provided in a busy oncology clinic would facilitate appropriate referrals to genetic counseling for breast cancer affected women of low socioeconomic strata. The authors found that using the Joseph et al. (2012), screening tool to gather personal and family history of cancer was extremely feasible in our patient population in this underresourced clinic. This study funded a research nurse to administer the screening tool and demographics form for eligible patients. The authors found that the design of the tool leads to an easy conversational approach with simple, straightforward questions. In keeping with the vision of Joseph et al. (2012), the tool did meet the criteria for ease of use and brevity by personnel with a high school education. The tool was accepted by our patient population and took on-average under 20 min to administer. We propose that the administration of the tool would transfer easily even if it were nonstudy-funded personnel such as front desk administrators or medical assistants.

All enrollees completed the screening tool and those referred for genetic counseling expressed an interest in adding to the cancer family history details before their counseling sessions.

Our study concurs that in patients who reported a family history of breast or ovarian cancer, the screening tool accurately identified women that met current NCCN guidelines. In the ‘high-risk’ group, all 36 participants counseled met NCCN criteria for high-risk identification as well as existing insurance guidelines for genetic testing. Women that consented for genetic counseling were keenly interested in following through with genetic testing to clarify their risk. 91.6% completed genetic testing and very often brought family members to their result disclosure sessions.

A glaring gap in capture of this screening tool was the lack of identification of potential participants that were younger than 45 years at breast cancer diagnosis. This was especially true if they did not report a strong family history of cancer. In our application of this tool, 46.4% of the total participants had a ‘low-risk’ score of which 28.2% still met NCCN criteria for

genetic counseling and testing. This percentage was higher than anticipated and was further investigated. All 13 participants were breast patients diagnosed at or under age 45 with no reported family history of cancer.

In 2012, when this tool was developed by Joseph et al., the NCCN criteria for screening did incorporate an age cutoff for breast cancer affected patients. That cutoff was 45 years when there is no family history reported or 50 years with at least one close blood relative (Appendix 1). The first question in the screening tool does ask if the individual has a breast cancer diagnosis and whether they were diagnosed at age < 50 years. The weightage given for a diagnosis at age < 50 does not independently reach the threshold cutoff of 6 (Appendix 3). The original purpose for the screening tool was to identify unaffected women based on family history and therefore this was not addressed. The weightage favored more heavily on the family history provided. In 2016, when Stewart et al., conducted the validation of this screening tool, they compared it to other family history screening tools as well as independent genetic counselor assessments in a low socioeconomic population of unaffected women presenting at screening mammography or primary care centers. Once again, their target population did not include cancer affected patients especially women with young onset breast cancers. Genetic counselor reviews in their cohort picked up an additional 15% of high-risk patients over what the screening tool picked up (Stewart et al. 2016). Our study of a cancer affected population resulted in a higher proportion (28.2%) that were not captured. The authors acknowledge that NCCN guidelines change over time and generally cast a wider net than what screening tools typically capture. Nevertheless, in 2018, when our study was conducted, the NCCN guidelines continued to maintain test eligibility criteria to include breast cancer diagnosed at age < 45 years with no family history or < 50 when there is family history of cancer in at least one close relative (Appendix 2). Our study brings to light that the screening tool is weighted heavily for family history, does not capture young-onset breast cancer patients who do not have (or do not know) their cancer family history. We recommend adding a subcategory (1d) in the screening tool for “breast cancer < age 45” which should score a 6. The

subcategory of 1c should be further defined to state “breast cancer between the ages of 46–50 years” for a score of 4. Optimizing the identification of at-risk patients is paramount as recent literature reports that even current NCCN guidelines do not capture all potential patients that need counseling and genetic testing (Bietsch et al. 2019).

An analysis of the genetic testing cohort indicates that the screening tool does not specifically capture mutation positive patients. Positive mutations identified in 9% (3/33 patients) in the ‘high-risk’ group were ATM, BRCA1, and PALB2. In the ‘low-risk’ group, the initial finding of 20% (2/10 patients) mutation positive rate was revised to 10% since one of the mutations was an SDHD gene which does not have an immediate bearing on breast cancer risk. Overall, the mutation rate fits with current literature on deleterious germline mutation identification rate in oncology clinics (Tung et al. 2016).

The SDHD gene was included in the expanded panel genetic test run on all study patients that consented to genetic testing. Therefore, although an incidental discovery, the SDHD mutation was not an unexpected finding. Rather, it was included in the panel since a few SDHD mutations have been identified in a Cowden-like presentation of breast cancer. Identifying an SDHD mutation does not have any direct bearing on the medical management for breast cancer and was later excluded from the mutation positive rate statistic. Notwithstanding the exclusion of this unusual finding, the mutation positive rate in the ‘low-risk’ was at least 10% which is equal to the rate found in the ‘high-risk’ group. In the proposed application of the screening tool, the ‘low-risk’ group would not have needed further testing follow-up. Testing was ordered in this group solely based on the clinical discretion of the oncologist’s interpretation of current NCCN criteria. Therefore, having a similar mutation pick-up rate in both groups indicates that the screening tool did not seem to provide a true added benefit to help identify mutation positive patients in our sample population.

In an overall view, there are other confounding factors that may skew the mutation positive rate between the two groups. The study’s relatively small sample size, and importantly, many women initially identified in the ‘high-risk’ group that did not follow through with their initial referrals to the genetic counseling clinic. There was a 31.8% attrition for all referred patients; majority were in the ‘high-risk’ group. Therefore, the mutation rate identified for this ‘high-risk’ group may be artificially low. Of particular note, the two patients that tested mutation positive in the ‘low-risk’ group were 42 and 43 years old at diagnosis with ER+/PR+ disease and no known cancer family history.

Our study was successful in its aim of demonstrating ease of access to genetic counseling. Even though we do not have data on all participants that were approached for the study, a 90% participation rate in the first 90 participants enrolled indicates a higher than expected enrollment rate than comparable

published data. Studies have reported participation rates as low as 45.5% in similar populations (Compadre et al. 2018). Many have studied the African-American patients’ reluctance to participate in clinical research (Hughes et al. 2004; Loftin et al. 2005; Powell-Young and Spruill 2013; Rabinowitz and Gallagher-Thompson 2010; Rivers et al. 2019) and identified factors such as fear of discrimination, lack of culturally appropriate education and outreach materials, and lack of information (Rivers et al. 2019). In other minority populations, additional factors like language and financial compensation have been identified (Davis et al. 2018). Despite such barriers, recent literature indicates that recruitment strategies of utilizing African-American recruiters and community-based programs to reach African-American participants could help double or triple research enrollment rates (Ewing et al. 2015) and retention rates (Yancey et al. 2006). In this study, our research nurse who was primarily responsible for recruitment was of African-American ethnicity. Although no formal attempt was made to measure the association of race impact, it should be noted that 50.5% of our overall study participants were African-American. Study enrollees also exhibited a strong desire to follow through with genetic testing. When offered the test by the counselor, 91.6% of the ‘high-risk’ group and 100% of the referred ‘low-risk’ group completed genetic testing.

The demographic breakdown in this study highlights an interesting contrast between the racial make-up of the ‘high-risk’ group and the ‘low-risk’ group. While Caucasian women (41.3%) made up the bulk of the ‘low-risk’ likely ‘first-in-the-family-diagnosis’ patient, the ‘high-risk’ group was predominantly African-American patients (60.4%) with a prominent family history, with a higher proportion of triple negative breast disease. It is difficult to postulate underlying causes of this finding. It could be that on average more African American women know details of their family history of cancer and Caucasian women in comparison do not. The authors have not come across any literature reporting on this type of ethnic difference in their study population. Average age of cancer onset in both groups was 52 years. This mean age is more than 10 years younger than the national average for breast cancer diagnosis, which demonstrates the uniqueness of this population when compared to a typical oncology clinic population. Although our sample size was too small to draw any definitive conclusions, this contrast sheds light on a group of young patients without a cancer family history that represent women at increased genetic risk. Further independent research is needed to tease out these findings in a larger population in a similar clinical setting.

Another important finding of this study was that a high number of participants did not follow-through with their genetic counseling appointment when they were required to go to another location to complete it. Indeed, 31.8% of referred patients were lost to follow-up after the initial screening process. A recent publication from a CSER consortium for a

heterogeneous group of genetic conditions cited the most common reason for declining participation had to do with study logistics. They highlight the importance of developing genetics and genomic studies that take into consideration the burden on the participant and re-iterate commitment to enhance enrollment (Amendola et al. 2018). After 18 months of our study period, our study was in the counseling phase after a majority of recruitment was complete, we altered our protocol to house the genetic counselor on site for two days every month and resulted in a re-capture of 22% (10/45) of those initially lost to follow-up. Since our sample size was small this did prove to be a significant capture of patients by changing protocol. These participants were subsequently asked open-ended questions to help gather insight into factors for initial non-compliance. As this was not one of the stated aims of the study this feedback was reported only anecdotally. Participants reported issues that differed greatly (side-effects of chemotherapy, work constraints, an infirm spouse) from those initially postulated as barriers during study design (no phone card, transportation barriers or lack of interest). One participant reiterated her interest; *‘I missed the appointment because of being sick after chemo. Maybe if the appointment was before the chemo had started I could’ve made it there.’* The presence of the genetic counselor on-site facilitated education of nursing staff, increased visibility of the study among physicians, research personnel and patients. Our experience was in keeping with other literature that reports on the increased impact of an embedded genetic counselor in a treatment clinic (Pederson et al. 2018; Senter et al. 2017).

Conclusions

Our study demonstrated that a point-of-care screening tool did increase ease of appropriate referrals to genetic counseling services and was well accepted by patients in a busy, safety net hospital clinic setting. It was effective at identifying high-risk women especially when a strong family history of cancer was reported. The weighted advantage questions on the screening tool were not particularly sensitive when family history was lacking. Minor adjustments in the first question of the screening tool to include a sub-category of ‘breast cancer diagnosed under age 45’ to equate to a score of 6 would enhance identification of ‘first-in-the-family’ breast cancer diagnoses that are young women at increased genetic risk. A limitation of our study is that our sample size was small for an in-depth statistical analysis and therefore only descriptive data has been reported. Nevertheless, this data presents some interesting findings in the ‘low-risk’ group. More research is needed to corroborate these findings and tease out this subset of the breast cancer population that may be truly high-risk in order to provide appropriate services.

The study was limited by the lack of data capture on our enrollment rate toward the end of the study. Postulating from the high enrollment rate in the first 90 patients, this low SEC, ethnically diverse population is motivated to participate in research. Study enrollment can be enhanced by timing the approach to patients, using recruitment staff from similar ethnic/racial backgrounds to recruit minority participants, imbedding a genetic counselor in treatment centers and by making the study design easy for participants to navigate. Anecdotal insights indicate that lack of follow-through may be due to issues other than lack of interest, financial or transportation constraints. The multiple faceted barriers to the uptake of genetic counseling services are evolving and may yet be under-studied.

Supplementary Information The online version contains supplementary material available at <https://doi.org/10.1007/s12687-020-00499-9>.

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Author contributions All authors have participated equally in the completion of this project. Conception and design of the project was spearheaded by Drs. Wiesner and Lammers. They were also primary involved in the design of the database for the study. Drs. Singh and Biltibo were instrumental in identifying and enrolling eligible patients for the study in clinic as well as data entry of participant details into the database. Ms. Kim Thomas was the Research Nurse recruiting patients, administering the screening tool, maintaining de-identified patient information, scheduling appointments and completing end of study documentation. Ms. Smita Rao facilitated as the genetic counselor for the study and drafted the manuscript for submission. All authors were involved in review and refine of the draft and finalizing the manuscript.

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Compliance with ethical guidelines

This study design was approved by the Institutional Review Board at Vanderbilt University Medical Center and the Meharry Medical College (IRB protocol number 15–07-409).

All procedures followed were in accordance with the ethical standards put forth by the Helsinki Declaration of 1975, as revised in 2000(5). Informed consent was obtained from all patients included in the study.

Conflict of interest Ms. Smita Rao, Ms. Kimberly Thomas, Dr. Rajbir Singh, Dr. Eden Biltibo, Dr. Philip Lammers and Dr. Georgia Wiesner declare that they have no conflict of interest. The authors declare that they have no competing interests.

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