This article describes family history assessment for colorectal cancer in three outpatient gastroenterology units and examines gastroenterology unit nurses’ knowledge and attitudes about family history assessments. Eighty-eight colonoscopy records were surveyed, and 16 RNs were interviewed. The medical record documentation was surveyed using a researcher-developed tool to identify type of cancer, age at disease onset, family relationship, and number of family members with cancer. Gastroenterology unit nurses were interviewed to assess knowledge and attitudes about family history assessment regarding colorectal cancer. Findings indicate that limited family history documentation was present in the medical record and that important age-at-disease-onset information was missing in 95% of patients with a family history of colorectal cancer and in 85% of patients with a family history of Lynch syndrome-associated cancers. No documentation was found in any charts about the number of affected relatives within the same family. Inconsistencies in family history documentation within the same medical record were noted, and family history information was found in multiple chart forms. Gastroenterology nurses rated family history as very important but gave a lower rating to personal knowledge about and resources for family history assessment.

Family history evaluation is considered to be one of the most effective tools for predicting disease (Alsopach, 2011; Leach & Eng, 2010) and is used for risk stratification, individualized screening for cancer detection and prevention, and targeted genetic services (Berg et al., 2009). Leach and Eng (2010) compared family history assessment to commercial personal genomic testing, often referred to as direct-to-consumer genetic testing, and family history assessment was found to be superior in stratifying population risk, moderate risk, and high risk for breast, prostate, or colon cancers. Although family history is one of the least expensive and most important genomic tools, it frequently is underused and underdocumented (Freezo, Rubenstein, Dunham, & Ormond, 2003; Murff, Greevy, & Syngal, 2007). Family history information is key for studying single-gene disorders and identifying the shared risk factors, gene alternations (single-nucleotide polymorphisms), lifestyle patterns, and environment of common complex diseases (Berg et al., 2009). Although the presence or absence of family history may be documented in other parts of the medical record, the identification of the number of affected relatives is often not documented.
the medical record, the important family history quality indicators often are not documented (Murff, Byrne, & Syngal, 2004; Sifri, Wender, & Paynter, 2002; Tyler & Snyder, 2006). Quality indicators include type of cancer, degree of relative (e.g., first-degree [father, mother, sister, daughter, son], second-degree [grandparent, aunt, uncle]), number of relatives with cancer, and age at disease onset. The family history details provide the essential information for risk assessment, stratification, and targeted genetic referrals.

Family history is one of the strongest risk factors for colorectal cancer (CRC) (Tyagi & Morris, 2003), the third most common cancer in men and women in the United States (American Cancer Society, 2011a). Family history information is used to identify high-risk individuals, such as those with hereditary colon cancer syndromes such as Lynch syndrome (LS) and familial adenomatosis polyposis (FAP). Patients at high risk for LS and FAP are targeted for in-depth family history assessments and testing by a genetics professional. Patients with an LS or FAP diagnosis may have aggressive screening at more frequent intervals and beginning at a younger age than the general population. These patients also may choose to have risk-reduction surgeries (Burt & Jasperson, 2008; Kohlmann & Gruber, 2006). Identifying patients at moderate risk (i.e., family history is not suggestive of a hereditary colon cancer syndrome but does indicate overall increased risk) is important as well. Patients at moderate risk are counseled to begin screening at an age younger than average-risk individuals and at more frequent intervals (Levin et al., 2008; National Comprehensive Cancer Network [NCCN], 2011; Rex et al., 2009) and follow current evidence-based guidelines for CRC prevention (Ahnen & Macrae, 2010). The American Cancer Society, the American College of Gastroenterology, and NCCN are examples of organizations that have established CRC screening guidelines according to family history information, including age at disease onset and number of relatives with CRC (Levin et al., 2008; NCCN, 2011; Rex et al., 2009). In addition, NCCN (2011) offers an algorithm for CRC screening according to increased risk based on positive family history (see Figure 1).

Family history of CRC can be assessed with a few simple questions and with patient self-report questionnaires; however, assessment and documentation frequently are overlooked in primary care gastroenterology clinics and units (Butterly et al., 2010; Fletcher et al., 2007; Nathanson, Zisman, Julian, McCaffrey,
Methods

Design and Setting

This descriptive study was conducted at three separate hospital-based gastroenterology units in a metropolitan area in the southwestern region of the United States. The hospitals are part of a multi-hospital system. The system’s institutional review board approved the study. The researcher chose the gastroenterology unit setting because of the connection between that unit and a focused family history assessment of CRC. Family history assessment and documentation practices for CRC were studied by surveying outpatient colonoscopy medical records and by interviewing gastroenterology unit nurses.

Sample

A convenience sample of 88 outpatient colonoscopy records were surveyed (about 30 charts per unit) for family history assessment information. Two of the three gastroenterology units were using an electronic health record (EHR), and one unit was in the process of converting to an EHR. The nursing interviews were conducted using a convenience sample of 16 full-time RNs employed at the three hospital-based gastroenterology units.

Procedures

Chart survey: The author developed a chart abstract form to collect family history information that would indicate an increased risk for colon cancer or a hereditary colon cancer syndrome. The abstract form included the family history quality indicators for type of cancer, age at disease onset for affected relatives, degree of relative, and total number of family members with cancer. Ethnicity was not included because the author was not permitted access to this information in the EHR. The author used the widely accepted NCCN Clinical Practice Guidelines™ for CRC screening (NCCN, 2011) as an additional guide in determining family history information. The types of cancer abstracted from the medical record included CRC and LS-associated cancers (i.e., uterine, ovarian, stomach, small intestine, urinary tract, bile ducts, pancreas, and brain cancers) (Eisen & Weinberg, 2005; NCCN, 2011). The survey did not address a family history of polyps as the reliability of a self-reported family history of polyps is unknown. The chart survey met patient privacy guidelines. At two of the facilities, the author surveyed the EHRs. At the third facility, the author abstracted information from a paper medical record.

Nursing interview: The author scheduled meetings with the three gastroenterology unit nurse managers to share project information and solicit input concerning the gastroenterology unit nursing interviews. An e-mail was sent to the nurse managers to be shared with the nursing staff. The e-mail described the project, time commitment (10–15 minutes), and the voluntary and confidential nature of the interview. The author then conducted an in-person interview with all participating gastroenterology unit nurses. The author asked questions using the nurse researcher-developed paper questionnaire as a guide and wrote down the nurses’ verbal responses. The questionnaire assessed the nurses’ perceived importance of a family history of CRC, measured on a 0 (least important) to 5 (most important) scale, and the perceived personal knowledge and resources of family history and CRC, measured on a 0 (least knowledgeable/resources) to 5 (most knowledgeable/resources) scale. Using an evidence-based practice implementation framework from Achterberg, Schoonhoven, and Grol (2008), the author asked the nurses to identify factors that interfere with collecting family history assessment of CRC information: (a) cognition (e.g., lack of evidence for the importance), (b) attitude and motivation (e.g., takes too much time), (c) social (e.g., nobody is interested), (d) organizational (e.g., not feasible in current work setting, no hospital guidelines or protocol), and (e) resource information (e.g., lack of resources to understand and follow-up). Knowledge of family history of CRC risk factors was assessed by asking gastroenterology unit nurses to respond to the following multiple choice questions.

• Which, if any, of the following family history information would change population screening guidelines and/or indicate a referral for genetic counseling (indicate all that apply)?
  – One second-degree relative with CRC
  – Two related second-degree relatives with CRC
  – Personal history of endometrial cancer younger than age 50

• Are the majority of CRCs?
  – Familial?
  – Hereditary cancer syndromes?
  – Not related to hereditary cancer or familial syndromes?

All interview information was deidentified. Participating nurses received a $5 gift card to thank them for being in the study. Descriptive statistics were used to analyze the data.

Results

Sample

Outpatient colonoscopy charts represented more women (n = 57, 65%) than men (n = 31, 35%) with a mean age of 61 and an age range of 26–86 years. The interviewed nurses were diverse in age, ethnicity, and tenure in the gastroenterology unit setting. They were primarily women (n = 15, 94%) and staff nurses (n = 13, 81%), with a mean age of 46 and an age range of 27–69 years.

1 The correct answers are “two related second-degree relatives with CRC” and “personal history of endometrial cancer younger than age 50” (NCCN, 2011).
2 The correct answer is “Not related to hereditary cancer or familial syndromes” (American Cancer Society, 2011b).
Case Vignette: Family History

You are interviewing Ellen, a 45-year-old woman who is scheduled for an outpatient procedure. She tells you that she has a family history of colon cancer; however, no family history is documented in the medical record. Ellen asks you when she should begin colorectal cancer (CRC) screening.

Family history
Ellen shares that her mother had colon cancer at age 48, and her maternal aunt had uterine cancer at age 45.

Ellen is unsure about any other family history. You recognize that Ellen’s family history is suspicious for a CRC hereditary cancer syndrome (i.e., a first-degree relative with colon cancer onset younger than age 50 and a second-degree relative with a Lynch syndrome-associated cancer [uterine cancer] at a young age).

Intervention
As you enter Ellen’s family history information into the electronic health record, a best practice alert comes on the screen and confirms your assessment that Ellen needs a genetics counseling referral and individualized CRC screening recommendations according to family history. You share the information with the healthcare team and make a plan for Ellen’s care according to her family history.

Fifty-six percent of the nurses were Caucasian (non-Hispanic), 25% were African American, 13% were Asian American, and 6% were Hispanic. Eighty-one percent of the nurses were direct-care nurses, 13% were managers, and one (6%) was a charge nurse.

Nursing Interview and Family History

Perceived importance, resources, and barriers: The nurses rated factors affecting family history assessment on a scale of 0 (least important) to 5 (most important). Most nurses (n = 11, 69%) rated the importance of personal CRC and family history assessment as a score of 5, with an average score of 4.6. Nurses rated their personal knowledge of and resources for family history assessment of CRC risk as an average score of 3.8. The most frequent barriers for collecting family history information were no hospital guidelines or protocol (n = 4, 25%) and “other” (n = 6, 38%) including “already done in another area,” “need additional help,” and “language barriers.”

Knowledge of colorectal cancer and family history: Nurses were asked to identify from a list of family history findings which factor, if any, would change population colonoscopy screening guidelines and/or indicate a referral for genetic counseling. Significant family history findings were identified correctly as (a) early onset endometrial cancer, younger than 50 years of age (n = 8, 50%), and (b) two second-degree relatives with colon cancer (n = 12, 75%). Half (n = 8) of the nurses incorrectly identified one second-degree relative with CRC as an indication for enhanced screening guidelines or cause for a genetic counseling referral. Three (19%) said that none of the findings were significant or “have no idea.” Most (n = 14, 87%) of the surveyed nurses overestimated the number of familial and hereditary cases of CRC in the general population.

Discussion

Medical Record Survey

A disappointing but not unexpected overall lack of family history documentation was found in the outpatient gastroenterology unit medical records. Insufficient documentation of...
quality indicator cancer family history information (i.e., type of cancer, age at disease onset, degree of relative, and number of affected family members) has been reported in several primary care studies (Murff, Spiegel, & Syngal, 2004; Sifri et al., 2002; Tyler & Snyder, 2006). When documented, family history of cancer often is recorded as positive or negative, without additional detail (Sweet et al., 2002). The current study’s documentation deficiency findings are congruent with what is reported in the literature for the gastroenterology unit and clinic settings (Butterfly et al., 2010; Dudley-Brown & Freivogel, 2009; Nathanson et al., 2008). Time constraints for the open-access endoscopy procedures (i.e., the gastroenterologist meets the patient only minutes before performing a colonoscopy) and a lack of simple, practical family history assessment tools make family history assessment difficult even in gastroenterology specialty areas (Dudley-Brown & Freivogel, 2009; Kastrinos et al., 2009).

**Family history and age at disease onset:** Of concern in the current study is the lack of documentation relating to family history regarding age at disease onset and identification of the degree of relative. Age at disease onset was missing in almost all of the charts that indicated a family history of CRC or LS-associated cancers. Degree of relative was missing in 42% of the charts identifying a family history of CRC and in about 25% of the charts identifying a family history of LS-associated cancers.

Young age at disease onset (younger than age 50) is considered a red flag for a hereditary CRC syndrome and is an indicator for a more comprehensive genetic evaluation (Eisen & Weinberg, 2005; NCCN, 2011). In primary care settings, Murff, Byrne, et al. (2004) reported that age at disease onset of CRC in first-degree relatives was recorded only 51% of the time. In a gastroenterology specialty area, a greater sensitivity to the importance of age at disease onset would seem likely, although the author of this article did not find this to be true for the current study nor studies reported in the literature (Butterfly et al., 2010; Nathanson et al., 2008). Using the risk assessment screening questions validated in Kastrinos et al. (2009), nurses should be able to conduct an initial assessment for hereditary CRC by asking if the patient (a) has a first-degree relative with CRC or an LS-associated cancer diagnosed before age 50, and (b) has two or three relatives with CRC at any age. Those simple questions would help identify patients who might require additional assessment or a genetics evaluation. Genetics professionals would use the more complex Amsterdam I and II and Bethesda criteria to assess the patient and family for appropriate genetic testing (Kohllmann & Gruber, 2006).

Deficiencies in documenting age at disease onset may explain why CRC genetics referrals are low compared to statistical estimates for hereditary CRC syndromes (Grover, Stoffel, Bussone, Tschoegl, & Syngal, 2004; Kastrinos et al., 2009), and why colonoscopy recommended follow-up intervals are not consistent with guidelines according to family history (Butterfly et al., 2010). Lack of disease onset family history information can lead to missed opportunities for screening and, ultimately, delayed diagnosis for CRC and hereditary CRC syndromes.

As in the Kastrinos et al. (2009) study, prompts for family history of age at disease onset can be as simple as asking if the age at disease onset is younger than age 50. Many individuals or patients may not know their family members’ exact age at CRC or LS-associated cancer onset but may have some idea if the family member was younger than age 40 or 50, for example. If family history information is assessed in general terms, more detailed information can be obtained by a genetic counselor or an advanced practice nurse with specialty training in genetics. A comprehensive family history assessment or referral for genetic counseling is indicated when one first-degree relative with CRC is diagnosed younger than age 50, two first-degree relatives with CRC at any age, an LS-associated cancer diagnosed at younger than age 50, or a pattern of hereditary CRC-related cancers is identified (NCCN, 2011; Tyler & Snyder, 2006).

The NCCN (2011) guidelines on CRC show how age at disease onset, degree of relative, and number of family members with CRC impact screening guidelines. The guidelines are used to improve detection of CRC in a moderate-risk population (i.e., CRC in two or more relatives and/or first-degree relatives younger than age 60). The family history colorectal screening guidelines may have economic benefits based on early diagnosis and curative removal of adenomatous polyps. Using a validated decision model to evaluate the cost-effectiveness of family history assessment, Ramsey, Wilschut, Boer, and van Ballegooinjen (2010) estimated the cost effectiveness of CRC family history-based screening programs to be $18,000–$51,000 per life year gained. Those numbers are based on disease cases averted; productivity loss avoided; and cost savings by the individual, family members, provider or insurer, public health agencies, and society (Tyagi & Morris, 2003).

**Degree of relative and number of relatives:** The deficiencies noted in family history documentation and no notation of the degree of the family member may relate to the overall lack of family history information. The risk for CRC increases with the number of family members diagnosed, early age at disease onset, and the degree of the relative (Tyagi & Morris, 2003). An individual with one first-degree relative with a diagnosis of CRC has twice the risk for CRC as an individual without a family history. This risk increases even more if more than one first-degree relative with CRC is identified (Edwards et al., 2010). The number of family members with CRC is a discriminator for individualizing screening guidelines. A family history of two related first-degree relatives with CRC at any age alters population screening guidelines so that screening should begin at age 40, instead of 50, or 10 years before the earliest diagnosis of CRC (NCCN, 2011).

In this study, the overall number of family members with cancer was not reported for CRC or LS-associated cancers. Reporting a ratio of affected or unaffected family members is an effective way to document family history and estimate significance of family history information (i.e., a patient with two of three [2:3] siblings with CRC is a stronger risk profile than a patient with one of eight [1:8] siblings with CRC). An EHR, which prompts for negative and positive family history information (i.e., unaffected and affected), will help determine an overall number of affected relatives. The ratio of affected and unaffected family members provides additional CRC risk assessment information.

**Patient self-report questionnaires:** No medical record documentation noted the use of patient self-report family history questionnaires or tools. The literature consistently suggests that paper and electronic self-report questionnaires and interviews...
provide more family history information, both quality and quantity, than what is documented in the medical record (Fletcher et al., 2007; Freezo et al., 2003; Murff et al., 2007; Quershi et al., 2009; Sweet et al., 2002), and that the accuracy of self-report family history questionnaires stands up well when compared with a genetics interview (Freezo et al., 2003; Quershi et al., 2009). However, few validated self-report instruments for collecting CRC information exist (Quershi et al., 2009).

For the gastroenterology unit setting, Kastrinos et al. (2009) developed a three-question preprocedural risk assessment tool to identify patients at high risk for hereditary CRC syndromes. The risk assessment tool was validated in a population of more than 5,000 patients scheduled for an outpatient colonoscopy. Patients completed a mailed risk assessment questionnaire prior to their scheduled colonoscopy, and nurses confirmed the risk information with the patient. The tool was studied with a highly educated Caucasian population and did not address patients who were at moderately increased risk for CRC. The Kastrinos et al. (2009) tool is a first step toward developing risk assessment questionnaires and procedures for the gastroenterology unit setting.

Chart documentation: Assessing medical record documentation of CRC family history was challenging. Two of the reviewed hospital-based gastroenterology units used the same EHR system. The EHR system has a family history section, although this section was rarely completed. The EHR family history section prompts included cancer diagnosis (not a specific type), relation, and status (i.e., age at death). The EHR did not prompt for important quality indicators such as age at disease onset or type of cancer; this information could only be captured in the comments section. In this study, the comments section rarely was used to document family history information.

The EHR offers extraordinary opportunities to document family history of CRC, quantify risk, and connect with decision support tools such as the NCCN (2011) guidelines. The ultimate goal is to optimize screening and identify high-risk patients (Vogt, 2008). However, many EHR systems have underdeveloped family history sections that do not allow for interpretive information (Feero, Bigley, & Brinner, 2008). Finding inconsistencies in family history information in the same health record could lead to missed opportunities for screening and genetic referrals.

Nurses’ Interviews

The gastroenterology unit nurses’ interviews provided valuable information concerning family history assessment practice issues and further insight concerning the need for EHR family history documentation. Although nurses rated family history assessment as very important, they rated their knowledge of and resources for assessment at a lower level. The number of incorrect responses to the two knowledge-based family history assessment questions reinforces the need for additional CRC and family history education.

According to the Consensus Panel on Genetic/Genomic Nursing Competencies (2009), all nurses should be able to “demonstrate an understanding of the relationship of genomics to health, prevention, and screening” (p. 11). That includes a nurse’s responsibility to assess family history information, facilitate genetic referrals, and incorporate risk information into a plan of care. Those competencies are consistent with the Oncology Nursing Society (ONS) position statement on the “Role of the Oncology Nurse in Genetic Counseling” (ONS, 2009) and the National Coalition for Health Professional Education in Genetics (2007) Core Competencies for All Health Care Professionals. The advanced practice nurse with specialty training in cancer genetics provides more comprehensive care in clinical genetics including genetic testing (ONS, 2009).

A draft report by the Secretary’s Advisory Committee on Genetics, Health, and Society (2011) described family history assessment barriers and emphasized the critical importance of genetics and genomics education for all healthcare professionals. An initial approach for nursing education and family history assessment is to start with the clinical relevance of family history assessment of CRC using case studies and stories (Kelly, 2009).

A frequent nursing interview response concerning family history practice issues was that assessments are “done in another department.” No sense of ownership or accountability was present for the family history assessment process. Without EHR prompts, gastroenterology unit nurses and other staff may not recognize that key information is not being assessed and documented. EHR decision support tools that incorporate family history quality indicators and link to best practices (e.g., NCCN guidelines) are needed to improve risk stratification of CRC screening practices and increase genetics referrals.

In this study, nurses identified “no hospital guidelines or protocols” as an interfering factor in family history assessment. In the primary care setting, Quershi et al. (2007) reported greater accuracy (86%–90%) identifying a positive family history of CRC with a telephone interview and a self-report questionnaire versus a personal interview alone (57%–65%). For the gastroenterology unit, Kastrinos et al. (2009) used a mailed preprocedural self-report questionnaire for initial family history assessment. When patients arrived for their scheduled colonoscopy, a unit nurse did a verbal confirmation of the questionnaire family history information. Neither Quershi et al. (2007) nor Kastrinos et al. (2009) studied the paired approach of self-report questionnaire and confirmation of personal interviews or telephone calls; however, this combined approach does warrant additional research. In the meantime, nurses should consider adding family history confirmation to gastroenterology unit protocols. Verbal confirmation may increase accuracy and, in the process, nurses can teach about risk stratification, screening according to family history, and clarify family history myths and misconceptions. Nurses provide a supportive presence for patients who are sharing what may be sensitive cancer family history information.

Limitations

This descriptive study had several limitations. The chart documentation survey and nursing interviews used convenience sampling with small sample sizes. The study was done at gastroenterology units from three separate hospitals that were within the same healthcare system. The nursing interview questionnaire was developed and administered by one nurse researcher (the author). Questionnaire validity and reliability is unknown. Because of the variability in medical record forms and processes at the three separate hospitals, the author was unable to determine when “no documentation” was charting by exception.
Conclusion

Research is needed on the optimal approaches to improve family history assessment in a variety of settings, not just in the gastroenterology unit. The research should evaluate patient family history reporting strategies or questionnaires, identify barriers to family history assessment, explore processes for confirming family history information, and target EHR improvements. Nurses have tremendous opportunities to be innovative leaders in family history assessment. Family history is the first genetic screening. Identifying family history assessment documentation and knowledge gaps are the first steps toward improving practice.

This study found worrisome deficiencies in CRC family history quality indicators and documentation as well as in nurses’ knowledge. Family history was not assessed, was inconsistently assessed, or not documented in the medical record. Family history assessment is a powerful tool for the prevention and early detection of CRC and identification of hereditary cancer syndromes, but only if used. Patients expect and deserve genomically competent nursing care and effective EHR systems that support family history assessment and documentation.

The authors gratefully acknowledge Elizabeth Winslow, PhD, RN, FAAN, for manuscript review and Kathy Baker, PhD, RN, ACNS-BC, CGRN, FAAN, participating gastroenterology unit nurses, and the medical librarians at Texas Health Presbyterian in Dallas for their support.

Author Contact: Patricia Paul Kelly, DNP, APRN, CNS, AOCN®, can be reached at pakelly1027@sbcglobal.net, with copy to editor at CJONEditor@ons.org.

References


*Receive free continuing nursing education credit* for reading this article and taking a brief quiz online. To access the test for this and other articles, visit http://evaluationcenter.ons.org/Login.aspx. After entering your Oncology Nursing Society profile username and password, select CNE Tests and Evals from the left-hand menu. Scroll down to *Clinical Journal of Oncology Nursing* and choose the test(s) you would like to take.

*The Oncology Nursing Society is accredited as a provider of continuing nursing education by the American Nurses Credentialing Center’s COA.*