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I, Carolyn F Prochniak, hereby submit this original work as part of the requirements for the degree of:

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Barriers to and Motivations for Referral to Cancer Genetics Clinics

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This work and its defense approved by:

Committee Chair: Lisa Martin, PhD

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Barriers to and Motivations for Referral to Cancer Genetics Clinics

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Master of Science

In the Program of Genetic Counseling
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By

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Committee Chair: Lisa J. Martin, Ph.D.
Abstract

Although it is well known that under-referral of colon cancer patients to cancer genetics clinics has been a chronic problem, no study has yet to examine why physicians may be ordering testing independently over referring to cancer genetics clinics. The current study explored variables which may impact a physician’s preference to order testing independently or refer to genetics services. A self-designed online questionnaire was distributed to the membership of the American College of Gastroenterology and the American Society of Colorectal Surgeons. Motivators to refer to cancer genetics clinics rather than order testing in their own office included fear of genetic discrimination issues and the belief that patients benefit from genetic counseling about the risks, benefits and consequences of testing. These results suggest that in order to increase referrals, genetic counselors need to embrace a marketing strategy that delineates the unique psychosocial benefits patients receive from participating in genetic counseling.

Key words: genetic counseling, hereditary colorectal cancer, genetic testing, referral, risk assessment
Acknowledgements

The members of my RAC (Dr. Lisa J. Martin, Sara Knapke and Erin Miller) for their guidance, collaboration and support. Drs. Jim Cranley of Digestive Health Network and Michael Guenther of Cranley Surgical Associates for providing guidance in developing the questionnaire. Dr. Janice Rafferty of Christ Hospital for sponsoring the study with the ASCRS. The staff and members of the ACG and ASCRS for agreeing to participate in this project.
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Introduction

Hereditary colorectal cancer accounts for as much as 5-10% of all colorectal cancer cases (American Cancer Society, 2008). It is essential to identify these individuals in order to provide appropriate medical management, including surveillance and surgical options which have the potential to extend lifespan by up to 15 years (Järvinen, et al., 2000). Despite the relatively high prevalence of hereditary colorectal cancer syndromes and the proven medical management options available, fewer patients are being referred to cancer genetics clinics than expected (Grover, Stoffel, Bussone, Tschoegl, & Syngal, 2004). This pattern of under-referral has persisted despite increasing educational efforts directed towards specialty care providers (Wong, et al., 2008).

Numerous barriers to referral to cancer genetics clinics have been noted in the literature, including lack of knowledge regarding who should be referred to services, doubts regarding the clinical utility of genetic testing and concerns regarding patient confidentiality and genetic discrimination (Freedman, et al., 2003). However, in many of these studies genetic testing is not separated from genetic counseling. This lack of separation makes it difficult to decide the impact of these barriers on provider’s choice not to pursue genetic testing, to coordinate testing independently, or whether they decide to refer to genetics services.

When a patient’s medical and family history are suggestive of a hereditary cancer syndrome, the physician must decide if they wish to pursue genetic testing. The physician then has the option to coordinate testing independently or refer to genetics services. Batra and colleagues (2002) found that only half of gastroenterologists would refer to a genetic counselor prior to
independently ordering genetic testing, while the remainder would provide their own counseling, refer to a counselor only in the case of abnormal results or not provide counseling at all.

The current study aims to characterize how gastroenterologists and colorectal surgeons are using colorectal cancer genetic testing in their practices and their current use of cancer genetics services. Moreover, the study aims to determine the barriers and motivations experienced by physicians when deciding whether to refer patients to cancer genetics services or coordinate genetic testing independently.
Methods

*Study participants:* Participants included gastroenterologists and colorectal surgeons who were recruited through emails directed to the entire membership of both the American College of Gastroenterology in October 2009 and to the American Society of Colorectal Surgeons in February 2010. Recruitment emails directed participants to the online questionnaire: participants had the option to complete the questionnaire anonymously. Participation in this survey was voluntary and all participants provided informed consent. As an incentive for participation, respondents were given the opportunity to request a packet of resources, including literature regarding the process of genetic testing and counseling as well as how to locate a genetic counselor. The study was approved by the IRBs of both Cincinnati Children’s Hospital Medical Center and the University of Cincinnati.

*Study design:* The questionnaire was designed to assess participants’ use of genetic testing for hereditary colorectal cancer and referral to cancer genetics services. It also included a series of possible barriers to or motivations for using genetic testing or genetics services in their practice. Additionally, the questionnaire included three case scenarios regarding the management of various individuals with a family history of colorectal cancer. These case scenarios were designed to fit the criteria for high, moderate and general population risk as delineated by the AGA (American Gastroenterological Association, 2001). The study questionnaire also collected general information such as medical specialty, degree, number of patients seen, and the year of graduation. The study questionnaire was modeled from questionnaires used in the following studies: (Batra, Valdimarsdottir, McGovern, Itzkowitz, & Brown, 2002; Freedman, et al., 2003; Koil, Everett, Hoechstetter, Ricer, & Huelsman, 2003; McCann, et al., 2007).
Derived variables: The risk assessment ability of the participants was derived using information about the three scenarios of risk. For each scenario, a single answer was designated as correct. With respect to risk assessment ability, respondents were classified as correct (if all three risk assessments were correct) or incorrect (if at least one risk assessment was incorrect). Individuals who did not answer all three questions were defined as missing data. Participants who were designated as endorsing a motivator selected that the motivator was either “very influential” or “somewhat influential” while participants who were designated as endorsing a barrier selected either “strongly agree” or “somewhat disagree.” Throughout the analysis, respondents who selected unsure or neutral were designated as missing data.

Statistical analysis: Our primary objectives were to 1) characterize the risk assessment abilities of gastroenterologists and colorectal surgeons, and 2) investigate characteristics of physicians who preferred to refer to genetics services over ordering their own genetic testing. To compare dichotomous outcomes, we used chi-squared analysis or fisher’s exact test.
Results

Participant information: A total of 196 individuals completed the online survey in response to the American College of Gastroenterology mailing. A total of 102 individuals completed the online survey in response to the American Society of Colorectal Surgeons. Based on the memberships for these groups, the response rates for the survey were 4% and 6.5% respectively.

The study sample consisted of 185 gastroenterologists (62.0%) and 105 colorectal surgeons (35.2%), the remaining individuals selected other specialties. The median number of patients seen per year was 2000. The median year of graduation for these physicians was 1988. Only 18.5% of participants reported having formal training in genetics. The vast majority (82.7%) of providers had referred at least one patient to genetics services or had previously ordered either tumor or germline genetic testing. There was no significant difference in rates of referral ($p = 0.26$) or ordering ($p = 0.19$) between gastroenterologists and colorectal surgeons.

Risk assessment

When presented with clinical scenarios, the majority of physicians were able to correctly identify the high risk patient (Figure 1). Physicians were less able to discriminate patients into moderate and general population risk categories, thus the number of physicians that were able to perform an accurate risk assessment for all three patients was low. There was no significant difference between the risk assessment abilities of gastroenterologists and colorectal surgeons ($p = 0.73$).

Physicians who were able to perform an accurate risk assessment for all scenarios were not more likely to have ever ordered tumor or genetic testing ($p = 0.64$) or to have previously referred to genetics services ($p = 0.72$).
**Barriers and motivators to referral preference**

Overall, respondents preferred to refer to genetics services (58.4%) over coordinating testing independently. There was no difference between the referral preferences of gastroenterologists and colorectal surgeons (59.6% vs. 55.1%, respectively; p = 0.51).

We compared physicians who preferred to refer to genetics services to those who prefer to coordinate testing independently (Table 2). Specific barriers to and motivators for referral to genetics services endorsed by all physicians are outlined in Table 1. Physicians who preferred to refer to genetics services over coordinating testing independently were more likely to believe that patients are at risk for genetic discrimination (p = 0.008) and that patients undergoing genetic testing benefit from genetic counseling by genetics specialists regarding the risks, benefits and consequences of the test (p = .048). Physicians who were able to perform an accurate risk assessment were also more likely to prefer to refer to genetics services over coordinating testing independently (p = 0.008).

**Role of physician in genetics consultation**

Physicians who preferred to coordinate testing independently were more likely to feel that their role in the genetics evaluation process included ordering tumor testing, ordering germline genetic testing, interpreting genetic test results and providing emotional support. These physicians were also more likely to feel confident performing these roles, with the exception of providing emotional follow-up support (Figure 2).
Discussion

The current study explored variables which may impact a physician’s preference to order testing independently or refer to genetics services. We found that the ability to perform an accurate risk assessment did not increase overall utilization of genetic testing or cancer genetics services. Moreover, physicians who report that their role includes ordering tumor testing, ordering germline testing and interpreting genetic test results and feel confident doing so are more likely to order their own testing than to refer to genetics services. However, providers who preferred to refer to genetics services over ordering testing independently valued the genetic counseling provided by genetics specialists regarding the risks, benefits and consequences of the test. Thus, future strategies for increasing referral to genetics services may be improved by focusing on the unique benefits that genetic counseling provides patients, over and above the services received when a physician orders their own testing. Future studies are needed to explore whether pre- and post-test genetic counseling is more effective when provided by genetic counselors than physicians.

The main limitation of this study was the low response rates for each group, which has been commented on in other studies as a result of increasing paperwork and decreasing available time for physicians (Lowstuter, et al., 2008). The high rates of physicians that had ever ordered tumor or genetic testing or have referred to genetics services is likely a result of ascertainment bias, with a high percentage of physicians interested in genetics responding. With this in mind, we expect that uncertainty when performing risk assessment and barriers to and motivators for referral would be magnified in a population of physicians not as interested in genetics.

Risk Assessment
Publications in journals directed towards gastroenterologists and colorectal surgeons have focused on providing education regarding hereditary cancer syndrome risk assessment and management (Hendricks, et al., 2006; Terdiman, 2005). Although these educational interventions have been successful in aiding providers in identifying patients at highest risk, educational efforts have had a lesser impact on improving provider’s ability to discriminate between patients at moderate and general population risk. Data from a 1998 survey of gastroenterologists found that 79% were able to correctly identify a scenario patient as having hereditary non-polyposis colorectal cancer syndrome or HNPCC (Batra, et al., 2002). Our study found that over 90% of providers were able to identify a similar high risk patient, but the majority (70%) was unable to stratify patients into high, moderate and low risk categories. This echoes findings from a survey done from 2004-2006 which found that only 30% of physicians were correctly able to identify four or more hypothetical scenarios as appropriate for referral to genetics services (Lowstuter, et al., 2008).

Referral Patterns

It is difficult to compare rates of utilization of genetics services because genetic counseling and genetic testing are rarely separated into two different processes in the literature, leading to a lack of information regarding providers’ confidence in performing the process of genetic counseling independently. In 1999-2000, only 31% of tertiary care providers (including gastroenterologists) felt comfortable providing their own genetic counseling (Freedman, et al., 2003). In the current study, this rate increased to 42% of gastroenterologists and colorectal surgeons preferring to order testing independently rather than referring to genetics services. Although not directly comparable, our data suggest that gastroenterologists and colorectal surgeons have gained confidence in their ability to facilitate genetic testing.
If physicians are performing genetic testing without the aid of cancer genetics services, it is essential that these physicians are able to provide the patient with accurate information. However, of physicians in this study that coordinated their own testing, 44% believe testing is not covered by insurance and 37% believe patients are at risk for genetic discrimination. In order to ensure optimal care for patients whose genetic testing is coordinated by physicians, increased education is needed to correct these misconceptions. This education is especially timely given the recent passage of the Genetic Information Non-Discrimination Act, which many physicians may not realize extends protection for many patients and families. These common misconceptions about risk of genetic discrimination and insurance coverage of clinical genetic testing are likely to result in decreased pursuit of genetic testing and referrals to cancer genetics services.

**Barriers and Motivators to Referral**

Lack of knowledge regarding risk assessment has long been cited as a major barrier to referral, leading to the idea that educational interventions aimed at improving physicians’ ability to perform risk assessment will increase referrals to genetic counseling (Batra, et al., 2002; Freedman, et al., 2003). In this study, physicians who were able to perform an accurate risk assessment were not more likely to have ever ordered genetic testing or referred to genetic counseling. However, these knowledgeable physicians were more likely to prefer to refer patients to cancer genetics services rather than prefer to coordinate testing independently. This may suggest that educational interventions and improved risk assessment skills have not led to a larger patient population having access to genetic testing or cancer genetics services but resulted in referral of patients to genetics services that would have already been selected as candidates for genetic testing. It is also possible that our survey did not have the power to detect a positive
effect of knowledge on referral patterns due to the high number of participants who reported having previously ordered genetic testing and referred to genetic counseling.

In a study conducted in 1999-2000, physicians reported general confusion about who should have genetic testing, what genetic testing meant for patients’ risk of developing cancer and how to manage patients with positive test results (Freedman, et al., 2003). This confusion led over 90% of these physicians to believe that genetic counseling was a mandatory step in the process of genetic testing. However, as physicians have received more education regarding the clinical use of genetic testing, their strong endorsement of genetic counseling as an integral part of genetic testing has decreased. In our study, the majority of physicians reported feeling confident in their ability to order and interpret genetic testing. Thus, only 72% believed that patients benefited from genetic counseling during the testing process.

A significant motivator previously reported in the literature was patient request for referral to genetics services. Brandt and colleagues (2008) found that 73% of primary care physicians and specialists based patient eligibility for referral to genetics services on patient request and that 54% of physicians did not refer eligible patients due to patient disinterest. In another study of general physicians and specialists, variables associated with increased likelihood of ordering genetic testing or referral included receiving advertisements about genetic testing and having patients inquire about genetic testing (Wideroff, et al., 2003). Our data show that having patients request genetics services does not make a physician more likely to refer to genetics services over ordering testing themselves. It may be that referral to genetics services is only increased when patients specifically ask for genetic counseling, and that physicians are providing testing in their office when patients simply inquire about genetic testing. As direct to consumer advertising increases, it will be important for genetic counselors to provide
information directly to consumers about the benefits of genetic counseling in order to ensure that this important route of referral is maintained.

Another route of referral may result from recommendations from professional organizations and guidelines that address whether to refer to genetics services. In our study, 74% of participants in our study identified these recommendations and guidelines as a positive influence on their decision to refer to genetics services. There are several guidelines that refer to the need to offer counseling and genetic testing to individuals at risk for hereditary colorectal cancer (American Cancer Society Colorectal Cancer Advisory Group, U.S. Multi-society Task Force, & American College of Radiology Colon Cancer Committee, 2008; American Society of Clinical Oncology, 2005; Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group, 2009). However, only the guideline produced by the American Society for Gastrointestinal Endoscopy specifics that “expert counseling” should be provided, which may lead to specialist physicians to interpret that the counseling that they provide is in compliance with these guidelines (American Society for Gastrointestinal Endoscopy, 2006). This is in direct contrast to the U.S. Preventive Services Task Force’s (USPSTF) 2005 guideline recommending “that women whose family history is associated with an increased risk for deleterious mutations in \textit{BRCA1 or BRCA2} genes be \textit{referred} for genetic counseling and evaluation for \textit{BRCA testing}” (emphasis added; U.S. Preventive Services Task Force, 2005, p. 355). This lack of emphasis on referral to genetics services in the guidelines may explain our finding that these guidelines did not cause a provider to be more likely to prefer to refer to genetics services than coordinate testing independently. Thus, in order to increase referrals from colorectal specialists, genetic counselors may consider lobbying for adaptation of the above guidelines to include referral to genetics services. Future studies should also examine whether
the specific USPSTF guidelines issued regarding breast cancer referrals have in fact increased referrals from specialist providers.

Benefit of Genetic Counseling

Genetic counseling addresses complex psychosocial issues with participants including determinants of decision making such as risk perception, personal experiences with cancer and personal health beliefs, as well as communication within the family, adjustment to genetic conditions and legal issues such as genetic discrimination (Peters & Biesecker, 1997). Up to 40% of individuals going through genetic testing need professional psychosocial support (Bleiker, Hahn, & Aaronson, 2003). These needs may not even be met in the traditional genetic counseling format as half of participants in a specific study identified needing further support from a genetic counselor (Roshanai, Rosenquist, Lampic, & Nordin, 2009).

Despite educational publications regarding the complexities and importance of psychosocial support provided by genetic counseling, many physicians feel like they are able to provide the same services as trained genetic counselors (Batra, et al., 2002). Providers who participated in the current study felt that if they were going to order genetic testing instead of referring to genetics professionals, it was their role to provide emotional follow-up support. However, these physicians were no more confident in these skills than those physicians who deferred this role to genetics providers. Beyond raising concern that physicians who order testing without consultation with genetics professionals are not able to meet the psychosocial needs of their patients, this data suggests that genetic counselors may be uniquely positioned to address the emotional needs of patients beyond the ability of a busy non-genetics physician. Moreover, as our data shows that physicians are more likely to refer when they understand the benefit of genetic counseling, marketing genetics professionals as uniquely able to address psychosocial
needs of patients, leading to proper informed consent, increased psychological well-being and appropriate health behaviors may be a means of increasing referral to genetics clinics. It is also important that genetic counselors emphasize their willingness and availability to provide genetic counseling at any point in the testing process, including post-testing, in order to increase referrals from physicians who feel confident coordinating the testing process independently but do not feel confident addressing psychosocial concerns after test results are received.

For the role of genetic counselors not to be usurped by physicians ordering their own testing, future studies must prove the worth of psychosocially focused genetic counseling provided by trained genetic counselors over the basic medical genetic risk assessment often performed by non-genetics medical providers (Peters & Biesecker, 1997). An incidental finding of Collins’ and colleagues study of patient satisfaction was that patients who had a genetic counselor or medical geneticist participating in their session with a gastroenterologist were more likely to have a greater increase in knowledge regarding familial colorectal cancer as compared to patients whose sessions did not include a genetics professional, which the authors hypothesized would lead to an increased ability to provide informed consent (Collins, Halliday, Warren, & Williamson, 2000). However, this was not a controlled trial and no psychosocial or health behavior outcomes were measured. Therefore, further controlled studies comparing the genetic counseling provided by non-genetics medical providers and genetic counselors must be conducted. These studies must not only focus on knowledge and risk perception, but psychosocial outcomes such as anxiety and cancer related worry must be included. These studies should also focus on the interpretation of test results within the context of the family history to determine if genetic counselors or physicians are more effective in facilitating communication of
results within the family, thereby allowing for appropriate genetic testing and surveillance of these family members.
Conclusion

In order to assure that the 5-10% of patients who have hereditary colorectal cancer susceptibility syndromes are provided with appropriate care, increased effort is needed to find a way to effectively provide genetic counseling and genetic testing services to these patients. As the frequency with which genetic testing is ordered in the offices of gastroenterologists and colorectal surgeons increases, physicians will become more comfortable performing risk assessment and coordinating testing. Additionally, increased direct to consumer advertising about the value of genetic testing may impact patient requests for genetic testing and the frequency with which genetic testing is ordered by physicians. For genetic counselors to continue to reach patients at risk for hereditary colorectal cancer, they will likely need to embrace a new marketing strategy based on evidence that cancer genetic counselors are better equipped than specialist physicians to meet the psychosocial needs of patients, which in turn leads to better patient well being and medical compliance with surveillance procedures.
Bibliography


Figure 1. Percent of gastroenterologists (GI) and colorectal surgeons (CRS) that was able to correctly identify a scenario patient as high, moderate or general population risk.
<table>
<thead>
<tr>
<th>Barrier or Motivator</th>
<th>% of participants that endorsed item</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clear guidelines or strategies are not available for managing patients with a confirmed inherited colorectal cancer susceptibility syndrome</td>
<td>37.8%</td>
</tr>
<tr>
<td>Genetic tests for inherited cancer susceptibility are not usually covered by insurance</td>
<td>69.3%</td>
</tr>
<tr>
<td>Patients with confirmed inherited colorectal cancer susceptibility syndromes are at risk for genetic discrimination</td>
<td>61.5%</td>
</tr>
<tr>
<td>Genetic testing is useful in making decisions about surgery and medical management</td>
<td>59.1%</td>
</tr>
<tr>
<td>Patients undergoing genetic testing benefit from genetic counseling by genetics specialists about the risks, benefits and consequences of the test</td>
<td>72.3%</td>
</tr>
<tr>
<td>The individual patient’s or family’s request for genetics services influences whether to refer to cancer genetics services</td>
<td>62.2%</td>
</tr>
<tr>
<td>Recommendations and guidelines from professional organizations and government agencies influence whether to refer to cancer genetics services</td>
<td>73.9%</td>
</tr>
</tbody>
</table>

Table 1. Endorsement of specific barriers to and motivators for referral by all participants regardless of referral preference.
<table>
<thead>
<tr>
<th>Item</th>
<th>Percent of those who prefer to order testing independently who endorsed item</th>
<th>Percent of those who prefer to refer to genetics services to coordinate testing who endorsed item</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clear guidelines or strategies are not available for managing patients with a confirmed inherited colorectal cancer susceptibility syndrome</td>
<td>47.7%</td>
<td>52.3%</td>
<td>0.20</td>
</tr>
<tr>
<td>Genetic tests for inherited cancer susceptibility are not usually covered by insurance</td>
<td>44.2%</td>
<td>55.8%</td>
<td>0.24</td>
</tr>
<tr>
<td>Patients with confirmed inherited colorectal cancer susceptibility syndromes are at risk for genetic discrimination</td>
<td>37.0%</td>
<td>63.0%</td>
<td>0.008*</td>
</tr>
<tr>
<td>Genetic testing is useful in making decisions about surgery and medical management</td>
<td>43.9%</td>
<td>56.1%</td>
<td>0.53</td>
</tr>
</tbody>
</table>
Table 2. Influence of barriers and motivators on whether physicians preferred to order genetic testing themselves or refer patients to genetics services. * denotes significant factors (p < 0.05)
Figure 2. Graph on the left: Influence of whether physician feels confident in performing the selected tasks on whether they prefer to refer to genetics services or order testing independently. Graph on the right: Influence of whether physician feels their role in genetics evaluation includes the selected tasks on whether they prefer to refer to genetics services or order testing independently. * Denotes significant results (p <0.05).