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Molecular Genetic Cancer Screening: Role of Prediction of Colorectal Disease in the Clinic Setting

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Dedication

This paper is dedicated to my husband, Aaron Spinks, who pushed me to pursue my passion and taught me that the price of success is hard work. Your support allowed my dreams to become a reality. To my children Blake, Ashley, Abigail, and Annabeth who were my strength, my passion, and reason for success. You have taught me that it is not about being perfect, it is about loving the simple things, doing what is right, at the right time, and never giving up. My children, you are every reason, every hope, and every dream I have ever had. To my sisters, Anne, and Michele, who have been my beacon of light and have lifted me up after every fall. You have believed in me and have carried me on your wings when I was weak. You taught me that small setbacks are just a blessing in disguise. To Peggy Spinks and Barbara Kitterman who have patiently cared for and nurtured my children so I could complete this journey. To my brave, beautiful sister-in-law, Ashleyanne Hensley, who was my inspiration for starting this project. Thank you for allowing me to share your story of courage and survival. You have changed lives. To all the single mothers, this is your chance, anything can happen. Your future is not yet determined. Put on your crown, you are a child of God. With God all things are possible. Embrace your calling and become the woman you are meant to be.

"And so it was with me, brothers and sisters. When I came to you, I did not come with eloquence or human wisdom as I proclaimed to you the testimony about God. For I resolved to know nothing while I was with you except Jesus Christ and him crucified."

-1 Corinthians 2:1:1,2

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Abstract

Colorectal Cancer is the third most common cancer in men and women and the second leading cause of cancer related deaths (ACS, 2019). Colonoscopy screening can prevent colon cancer by early detection and removal of adenomatous colon polyps. The ACS has been lowered from age 50 to 45 due to an increase in the prevalence of colon cancer in people below the age of 50 years of age. Molecular genetic screening is a tool that providers can use to identify patients who are at risk for premature adenomas. People who have a genetic variant are more likely to develop adenomas at a young age and have a faster adenoma to adenocarcinoma conversions time. The purpose of this study is to identify if hereditary genetic screening has a positive or negative predictive value on patients who present below the screening age and have the presence of adenomas on a colonoscopy. This is a prospective study evaluated if a hereditary cancer screening assessment was a viable tool to identify patients who were at risk for early adenomas. A total of 150 charts were reviewed. The mean age of the participants was 40 years old, gender distribution (39.3%) female and (60.7%) male, average BMI was 32.85, with 76.7% non-smokers and 23% smokers. There was no statistically significant relationship found between patients who answered positive for a personal family history of colon cancer, first degree family history, and extended family history. There was a statistically significant correlation with patients who answered positive to a second-degree family history. Limitations of the study include small sample size, Participant recall or knowledge of family history and accuracy of the genetic screening process. Doctorally Prepared Nurses serve as leader working at the top of their education doing research to improve patient care outcomes. Nurse practitioners are a vital member of the healthcare team.

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CHAPTER 1

INTRODUCTION

Empirical Evidence of Prevalence and Significance of Colon Cancer

Colorectal cancer is the third most common cancer in men and women in the United States (American Cancer Society, 2017). It is the second leading cause of cancer death in the United States (JAMA, 2016). In 2015, 140,788 new cases of colorectal cancer (CRC) were reported, and 52,396 people died from colorectal cancer (CDC, 2017). The age adjusted rate of new colorectal cancer was 39.7 per 100,000 people (95% Confidence Interval: 38.3- 42.2); from a total of 3,206 reported cases of colorectal cancer. The survival rate of all types of cancer is 65.6%, with colon cancer having a 5-year survival rate of 65.9% making the prediction and screening of high-risk patients a vital issue to address in medicine. Incidence and death rates that have been recorded by the United States Cancer Statistics identify a need for increased awareness and screening protocols. One of the most disturbing developments in trends with colon cancer is the marked increase in the incidence and mortality in the younger population who are not of age to undergo colonoscopy screening. Data show that there was a sharp rise in the 1990's that increased the incidence of colon cancer in Americans under the age of 50 years of age with no epidemiological evidence explaining a cause.

The purpose of this study was to evaluate if hereditary cancer screening is appropriate to initiate in the clinic setting as a valid screening tool to identify patients at greater risk for early development of colorectal cancer or colon adenomas. Empirical evidence and research will be taken from peer reviewed articles and professional medical associations.

Recommendations for Colonoscopy Screening

The American Cancer Society (ACS) recently changed their recommendations to begin colonoscopy screening on average risk patients to 45 years of age, continuing until the age of 75 years old. Between the ages of 76 and 85 years the decision for colonoscopy should be based on the individual's state of health, life expectancy, and prior colonoscopy screenings. After the age of 85, patients should not need colonoscopy screening.

Colonoscopy remains the gold standard for the detection and removal of colorectal polyps. Other test modalities that may be used are annual high-sensitivity fecal occult blood test, annual Cologuard test, flexible sigmoidoscopy every 5 years, or CT colonography every 5 years. If any of these tests are positive, colonoscopy would be indicated and would no longer be considered a screening exam which is covered 100% by insurance but would default to a diagnostic colonoscopy which would be subject to the individual's deductible. The decision for what type of screening should be individualized and discussed with the patient.

There are currently two different organizations that regulate when insurance companies will pay for a colonoscopy screening. The U.S. Preventive Task Force (USPTF) recommends screening for colorectal cancer beginning at 50 years of age and continuing until age 75 years of age. The decision to screen for colorectal cancer should be individualized and based on the patient's current state of health and previous colonoscopies (USPTF Colon Cancer Screening, 2016). Colonoscopy screening reduces colorectal cancer mortality (JAMA, 2016). Potential adverse events associated with colonoscopy screening are rare however, increase with age. The USPSTF states that with the with high certainty of colonoscopy the benefit outweighs the harm (USPTF, 2016)

State laws determine whether insurance companies will honor colonoscopy screening to begin at age 45 per ACS guidelines or age 50 per USPTF guidelines. Tennessee follows ACS guidelines (CRC Screening Law: State of Tennessee, 2019). Populations who are excluded from screening colonoscopy include: a strong family history of colorectal cancer, a personal history of colorectal cancer or colon adenomas, a personal history of inflammatory bowel disease, a known family or personal history of genetically associated cancer syndrome such as familial adenomatous polyposis syndrome also known as Lynch syndrome or hereditary non-polyposis colon cancer (HNPCC); a personal history of radiation to the abdomen or pelvis to treat a prior cancer (ACS, 2019). Reasons for these exclusions are that this population has already been targeted as being high risk and American Gastroenterology Association and American Cancer Society have published specific guidelines for this group of patients.

Risk Factors

Many risk factors have been associated with the development of colon cancer. Patients who smoke, have a greater than normal BMI, consume high amounts of alcohol, decreased dietary fiber, sedentary lifestyle, diet high in red meat and low in fruits and vegetables are at increased risk for colon cancer (CDC, 2019). Patient who have certain medical conditions such as inflammatory bowel disease (Crohns Disease, Ulcerative Colitis), or a history of abdominal or pelvic radiation are at increased risk. Individuals who have a primary family history of colon cancer or adenomatous colon polyps also have increased risk. All these factors are taken into consideration when providers discuss colonoscopy screening with patients.

Changes in Screening Age by the American Cancer Society

The state of Tennessee and many other states set their screening protocols by the recommendations of the American Cancer Society. The ACS bases its guidelines on trends noted

over time periods. The ACS has recently lowered the screening age of average risk individuals to begin at age 45 years old due to the increase in incidence rates of colon adenomas in the middle age population. According to an observational study performed by Sigel et al. (2017), taken from data recorded by the Surveillance, Epidemiology, and End Results (SEER) program, 1975-2014 there has been a 51% increase in CRC in those younger than 50 years of age. Also noted was an increase in the incidence of rectal cancer. According to Howlander et al. (2019) adults born around 1990 have twice the risk of colon cancer and four times the risk of rectal cancer compared to adults who were born in the 1950's. Also noted was an increase in incidence in the white population and an increase in incidence and mortality with successively younger birth cohorts. Sigel's study (2017) has been labeled an epidemiological phenomenon. Colon cancer rates have nearly doubled from the rates since the 1980's. The researchers felt that the study was not subject to bias arising from colonoscopy in the older cohorts because negligible screening and case finding occur in the youngest cohorts. CRC mortality rates among whites ages 30-39 years have increased since 1995, and have been decreasing in African Americans, however, African Americans still have a 50% higher chance of developing colon cancer than whites.

Purpose of Molecular Screening and Hereditary Cancer Questionnaire

The Human Genome Project commenced in 1990 and was completed in 2003. As a result of this project scientists can identify gene variants that increase the chance of genetic disease. In 1994, the first BRCA1 breast cancer gene was discovered. This was monumental as it allowed for increased surveillance in women who carry the gene. It also allowed women the option to undergo elective mastectomy based on their risk of developing breast cancer.

The field of gastroenterology can also benefit from genetic testing to help strategize on when to start colonoscopy screenings and when to repeat colonoscopy screening. Approximately three percent (1 in 30) of patients who are diagnosed with CRC have an inherited genetic condition that can be identified by genetic testing. People with hereditary cancer syndrome are more likely to develop CRC before the screening age of 50 years of age (Jenkins et al., 2015). Mortality rates attributed to CRC have decreased 53% from 1970-2016 because of increased screening and advances in technology and treatments (ACS, 2018). The increase in incidence of CRC and mortality in patients younger than age 55 indicates the need for improved screening and prediction models (Ma & Melson, 2018).

The application of molecular screening in clinal practice has a role in identifying patients who are at risk for the development of CRC. Patients with known Lynch Syndrome are more likely to get CRC, uterine, stomach, liver, kidney, brain, and skin cancer. In addition to identifying patients with Lynch Syndrome it can aid in the identification of at-risk family members. Lynch syndrome is caused by inherited genetic mutations which affect the DNA sequence of replication. Lynch Syndrome is the most common autosomal dominantly inherited cancer syndrome that predisposes a person to CRC. Mutations that have been identified in Hereditary nonpolyposis colorectal cancer (HNPCC) include: *MLH1* (chromosome 3p21); *MSH2* (chromosome 2p16); *MSH6* (chromosome 2p15); *PMS2* (chromosome 7p22, *MLH3* (chromosome 14q24.3; and possibly *PMS1*. Most HNPCC associated cancers, (90%) arise from mutations in MLH1 or MLH2 (Lynch et al..,2003). Once a patient has been identified as having HNPCC/Lynch syndrome the screening colonoscopy changes from age 45-50 years of age to age 20-25 years with repeat colonoscopy every 1-2 years. The death rate of patients who have HNPCC colon cancer compared to the general population of unknown or sporadic causes of

colon cancer accounts for 66% of CRC related mortality. Early identification of HNPCC can reduce mortality of colon cancer through colonoscopy and frequent screenings. A recent prospective study (Jarvinen et al., 2000) included 22 families with Lynch syndrome comparing cancer incidence and mortality between 133 at risk members who had undergone colonoscopy screening over 15 years, and 119 members who declined screening. Individuals who underwent colonoscopy screening every three years were found to have had a lower CRC incidence of six percent as compared to 16% in the unscreened group (Jarvinen, et al.,2000) and a lower mortality rate of eight percent as compared with the unscreened group of 22%.

HNPCC associated adenomas are believed to arise in the proximal or ascending colon (60-80%). Adenomas in the proximal colon show a higher progression to high-grade dysplasia and the development of colon cancer. Patients with Lynch Syndrome typically have polypoid villous histology with a more rapid progression from polyps to carcinoid of 35 months compared to 10-25 years in patients without Lynch Syndrome (Giardiello et al., 2014) Patients who have a genetic mutation have an average mean diagnosis age of 27-46 years of age in comparison of the mean age of sporadic cancer of 69 years of age. The diagnosis of HNPCC or Lynch syndrome can be made by either immunohistochemical analysis of the MMR gene protein in tumors or by DNA testing showing a pathogenic germline mutation in the DNA mismatch, MMR or EPCAM genes. Germline testing on all patients suspected to have Lynch Syndrome can aid with early diagnosis.

"It is becoming the standard of care at many centers that all individuals with newly diagnosed CRC are evaluated for Lynch syndrome through molecular diagnostic tumor testing" (Tomiak et al., 2014). Genetic testing involves either giving a blood or saliva specimen. Many insurances will cover genetic testing if a patient meets criteria. Hereditary Cancer Screening

Questionnaire is a prediction model that helps guide the healthcare team on recommended age of screening procedures. Prediction models include family history of hereditary cancers (pedigree), age, sex, and previous history of colon cancer to help identify and provide quantitative estimates of the likelihood of a DNA mismatch. Although there are several models available, they all include family history of hereditary cancers (pedigree), age, sex, and previous history of colon cancer

Genetic labs use a multi-step approach to ensure the accuracy of genetic testing. The early part of the process starts in the clinic where patients who have a significant family or personal history of Lynch associated cancers submit a hereditary cancer screening questionnaire. Informed consent must obtained prior to any genetic testing. The specimen is sent to a lab with the patient's demographics, insurance information, and hereditary cancer screening assessment. Genetic counselors evaluate the hereditary cancer screening form and provider documentation. Financial counselors work with payer sources to obtain coverage. If the patient does not meet criteria per insurance guidelines cash payment and payment plans are accepted. The geneticist evaluates the specimen for germline mutation. If the patient has a germline mutation the gene that is affected is identified with a lifetime risk of developing cancer and recommending screening protocols.

Clinical Question

Gastroenterology clinics are seeing an alarming number of young patients presenting for colonoscopy for diagnostic issues that are incidentally diagnosed with adenomas. These patients would not have undergone an initial screening colonoscopy until age 45-50 years old (AGA, 2018). A hereditary cancer screening questionnaire could aid in identifying patients who are higher risk for the development of colon cancer (ACS, 2017). As advanced practice nurses, it is

our responsibility to identify better ways to care for our patients. According to the Code of Ethics developed by the American Nurses Association (ANA, 2015) “The nurse, in all roles and settings, advances the profession through research and scholarly inquiry, professional standards development, and the generation of both nursing and health policy.” With genetic testing being available and highly reliable we can decrease mortality and incidence of colon cancer by colonoscopy surveillance. This would allow providers to personalize a plan of care based on the patient, considering society guidelines per the American Cancer Society. The purpose of this study and integrative review was to identify if initiating a hereditary cancer screening assessment in the clinic setting aids in early identification of patients who are at higher risk for colon cancer and adenomas below the screening age of 50 years of age.

Thus, the purpose of this study and integrative review was to identify if initiating a hereditary cancer screening assessment in the clinic setting aids in early identification of patients who are at higher risk for colon cancer and adenomas below the screening age of 50 years of age.

PICO Question

In the outpatient gastroenterology clinic, for patients who present with issues that warrant colonoscopy below the screening age, does implementation of a Hereditary Screening Questionnaire have a positive or negative predictive value for risk of colon cancer and adenoma detection rate.

Definition of Terms

The following definitions are included as they are directly related to this research and to provide better clarity.

- **AACN Synergy Model –** nursing theoretical framework designed by the American Critical Care Nursing Association that has a core concept that synergy occurs when the

needs or characteristics of the patients and families influence and drive the characteristics or competencies of the nurse (AACN, 2018).

- **Adenoma** – precancerous polyp (Merrian Webster,2020)
- **American Cancer Society (ACS) –** one of the largest private, non-profit cancer research funds ("About us | What is the American Cancer Society, 2018.)
- **American Gastroenterology Association (AGA) –** Professional Medical Association for Gastroenterologists (*American Gastroenterological Association*, 2019)
- **Colonoscopy –** medical exam to evaluate the inside of the large intestines (Colonoscopy, 2018)
- **Computed Tomography Colonoscopy –** three-dimensional model of the colon that a radiologist uses to evaluate the bowel, also known as a virtual colonoscopy ("CT Colonography," 2018)
- **Cologuard –** stool test to identify DNA mutations and other abnormalities in the stool to determine is a test is positive or negative ("At-home colon cancer screening test | Cologuard® | Risk info," n.d.)
- **Colorectal Cancer (CRC)** Cancer that arises from the colon/rectum ("About colorectal cancer," n.d.)
- **FIT Test – fecal immunochemical test** – screening test for blood in the stool ("Meet Cologuard® | Learn about effectiveness | Risk info," n.d.)
- **Hereditary Cancer** – Cancers that are more likely to present in patients wo have genetically identified gene mutations. ("Lynch syndrome," 2020)
- **Lynch Syndrome** – autosomal dominate condition, most common cause of inherited CRC. The eponym “Lynch syndrome” is named after Dr. Henry T. Lynch who is credited

for discovery of this condition. The designation applies to families and patients with a germline mutation in an MMR gene of loss of expression of the MSH2 gene due to deletion in the EPCAM gene ("Lynch syndrome," 2020)

- **HNPCC** – Hereditary nonpolyposis colorectal cancer syndrome. This term may be used interchangeably with Lynch syndrome. It is an autosomal dominant genetic condition that is associated with a high risk of colon cancer. HNPCC is associated with germline mutations of DNA mismatch (American Gastroenterology Association, 2017)
- **Mortality –** death associated with a condition, for the purpose of this study mortality relates to deaths associated with colon cancer
- **Occult Blood –** blood in stool
- **Colon polyp –** abnormal tissues growth in the colon, polyps can be characterized by their pathology as malignant, adenomatous, hyperplastic, or inflammatory
- **Polyposis syndrome** – Identified in patients who have greater than 20 lifetime adenomas, patients with a personal history of desmoid tumor or other extracolonic manifestations of familial adenomatous polyposis (FAP), (American Society of Colon and Rectal Surgeons, 2016)
- **Sigmoidoscopy –** evaluation of the sigmoid colon with either a colonoscope of sigmoidoscope ("Definition of sigmoidoscope," n.d.)
- **The U.S. Preventive Task Force (USPTF) –** independent, volunteer panel of national experts in disease prevention and evidence-based medicine

Theoretical Framework

The theoretical framework (see Figure 1) chosen to support this research was the AACN Synergy Model of care. The AACN synergy model was created by a group of nurses

representing the American Association of Critical Care Nurses (Swickard, Swickard, Reimer, Lindell, and Winkelman, 2014). The Synergy Model was intended to put the focus on the needs of the patient and the competencies of the nurse. According to the AACN, synergy results when the needs and characteristics of a patient, clinical unit, or system are matched with a nurse's competencies (AACN, 2016). The model described seven characteristics that a patient will manifest during various phases of their illness: stability, complexity, predictability, resiliency, vulnerability, participation in decision, making and care, and resource availability. This theoretical framework applies to this study as it puts the focus on the patient and uses the skills and knowledge of the advanced practice nurse to improve patient outcomes. The role of molecular biology and genetic screening is complex but has a purpose in predictability of a disease and illness. Decision making for colonoscopy before the screening age on vulnerable patients at risk due to their history can be enhanced using hereditary screening tools as a valid resource.

The Adventist Framework Model has three key constructions including caring, connecting, and empowering. Nursing as a profession, is known for their culture of caring. The primary concept of caring means that nursing practice and patient care should evolve with evidenced based practice guidelines to alleviate suffering and disease processes. Genetic testing falls into this category as it is evidence based and can prevent colon cancer. The second concept is connection. Healthy relationships are derived from connecting with people. Ellen G. White wrote that “Christ came to the earth and stood before the children of men and through our connection with Him we are to receive, to reveal, and to impart.” ("Counsels on Stewardship," 1886). Her words tell us that for us to connect with our patients we must connect on a physical, psychological, and spiritual levels. Our goal is to create positive outcomes and trust with the

people we are have been entrusted to care. The last concept in this model is empowering. Empowering means that we provide quality care to improve patient care. As evidence has shown, the gold standard for prevention of colon cancer is colonoscopy (ACS, 2017). The screening age as previously mentioned is 50 years old. With genetic testing we can identify patients who have a hereditary predisposition to developing adenomatous colon polyps at an age younger that 50 of age. Genetic anomalies can be detected now because of the Human Genome Project. Individuals with an HNPCC gene mutation have an estimated 80 percent lifetime risk of developing colon or rectal cancer (NIH, 2012).

Conceptual Model- AACN Synergy Model Adventist Framework

Biomolecular Screening for Adenomatous Colon Polyps

Advanced Practice Nurse

Caring → Connecting → Empowering

Nurse Practioner

Clinical Inquiry

Patient Advocacy

Systems Thinking

Moral Agency

Patient Needs

Quality of Life

Trust and Comfort

Complexity and Vulnerability

Resource Availability

Figure 1: American Association of Critical Care Nurses (1996) From *American Association of Critical Care Nursing Synergy Model*. Retrieved from (https://www.aacn.org/nursing-excellence/aacn-standards/synergy-model).

Adventist Framework for Nursing (2017) Patricia S. Jones et al., "A Distinctive Framework for Adventist Nursing," *The Journal of Adventist Education* 79:5 (October-December 2017): 4-13. Available at https://jae.adventist.org/en/2017.5.2.

CHAPTER 2

Review of Literature

The topic of the review this literature is colon cancer and hereditary genetic conditions such as Lynch syndrome and HNPCC and how genetic testing would be beneficial to implement as a screening tool for patients under the age of 50 years who are at risk for developing adenomatous colon polyps. A search of the following databases was conducted: CINAHL complete (from 2009-present), the AACN website, American Gastroenterology Association, American College of Gastroenterology, National Institute of Health, National Comprehensive Cancer Network, UpToDate, Centers for Disease Control, American Cancer Society and Google Scholar. The key words that were searched included the following: *colon cancer, genetic testing, hereditary nonpolyposis syndrome, Lynch syndrome, adenomatous colon polyps, colonoscopy screening guidelines, and prediction tools for colon adenomas.*

Society Guidelines for Colon Cancer Screening

The American Gastroenterology Association along with the U.S. Multi-Society Task Force of Colorectal Cancer (MSTF) which represents the American College of Gastroenterology, the American Society for Gastrointestinal Endoscopy updates the screening recommendations for screening colonoscopy. Three tiers of colon cancer screening have been identified with tier one considered the gold standard and cornerstone of surveillance methods. Colonoscopy screening should begin at age 50 and age 45 for African Americans of average risk. Tier one screening involves colonoscopy every 10 years and an annual fecal immunochemical test (FIT). Second tier testing includes CT colonography every five years and FIT-fecal DNA test every 3 years, and a flexible sigmoidoscopy every five to ten years. Screening colonoscopy should be offered to patients up to the age of 85 if they have not had previous colonoscopy. Cessation of screening

colonoscopy should be considered at age 75 or with less than 10 years of life expectancy (Rex, et al., 2017)

Average risk patients start colorectal cancer screening at age 45 regardless of ethnicity (ACS, 2018). Patients over the age of 85 should no longer have colonoscopy screening. In May 2018, the ACS, lowered the age to started screening at age 45 instead of 50 due to the increasing number of new cases of colon cancer in younger adults (National Cancer Institute, Surveillance, Epidemiology, and End Results Program 2018).

Society Recommendations for Evaluation of Patients with Lynch Syndrome

Lynch Syndrome is the most common heritable cause of colorectal cancer and accounts for up to three percent of colorectal cancers. Patients who are diagnosed with Lynch syndrome have a cumulative incidence of colorectal cancer up to 80% in a lifetime. Patients with a family history suggestive of Lynch syndrome with no personal history of colon cancer should be offered a predictive model to evaluate their probability of having Lynch syndrome (American Gastroenterology Association, 2015). Patients who have greater than five percent on a prediction model should have genetic germline testing (Rubenstein, et al., 2015)

Lynch syndrome patients have a higher risk of developing colorectal cancer. Most colorectal cancers in patients with hereditary cancer develop before the age of 50 (American Cancer Society, 2017). Patients who have Lynch syndrome should start their colonoscopy in their early 20’s and have colonoscopy every 1-2 years for surveillance (National Comprehensive Cancer Network, 2015). Genetic testing is advised for family members of patients with Lynch syndrome.

Colon Cancer Incidence Rates and Adenoma to Carcinoid Progression Rates

The best way to prevent colon cancer that is associated with Lynch syndrome CRC is through screening colonoscopy (ACS, 2018). Through regular screening colonoscopy in patients with Lynch syndrome, mortality is decreased by 67% (Niv et al., 2014). Studies to estimate age and sex specific colorectal cancer risks have been unsuccessful due to sample sizes being too small to produce reliable data. A meta-analysis of 1,114 Lynch syndrome families with MLH1 and MSH2 mutations were studied to better estimate age and sex specific short-term risks of CRC in patients with Lynch syndrome (Jenkins et.al, 2015). The estimated cumulative risk of CRC by age 70 ranged from a 27%-47% for males and 22%-37% for females. The hazard ratio for CRC decreased with age being the highest when they were ages 30-39, and lowest after age 70. This study estimates by applying hazard ratios to the US general population that 1.4 % of male carriers and 1.0 % of female carries between the age of 20-29 will be diagnosed within five years. Short-term risk of CRC in patients with Lynch syndrome is dependent on age, increasing rapidly until middle age. While yearly colonoscopy in their 20's may not be indicated, annual colonoscopy for those older than 30 should be required to help in the early identification of symptoms, and thus to prevent the development of colon cancer (Jenkins et al., 2015)

The increasing incidence of young adults who have been diagnosed with colorectal cancer was assessed by Sinal et al, (2017). CRC incidence trends were taken from 1974-2013 (n=490,305) and analyzed by five-year age group and birth cohort. From the mid 1980's through 2013 the diagnosis of CRC in adults ages 20-29 increased 2.4%; for ages 30-39 there was a one percent increase; and ages 55 and older the incidence rate declined. In the mid 1990's rates increased 1.3% in younger adults for ages 40-49. Also noted was that people under the age of 50 were more likely to have distal colon tumors whereas people over the age of 50 were more likely

to have proximal colon tumors. CRC screening at age 45 is not yet supported by all insurance but it is worth noting that in 2013 there were 10,400 new CRC diagnosis in adults age 40-49.

Colonoscopy findings from 54 people with pathogenic mutations in MSH2 and MLH1 were evaluated for the development of CRC in patients with Lynch syndrome (Edelstein et al., 2011). Differences in colorectal phenotype were analyzed with genotype and dwell time was calculated for advanced neoplasms. Among mutation carriers the cumulative risk of CRC by age 40 was 43% and 72% by age 80. Polyp dwell time from advanced adenomas to colon cancer was 33 months plus or minus 16.2 months for people under the age of 40 and 35.3 months plus or minus by age 80 (p=.198). Annual colonoscopy screenings and surveillance for patients with Lynch syndrome based on short adenomatous polyp to carcinoma dwell time were recommended (Edelstein et al., 2011). Limitations include a small number of patients and failure to find statistical analysis of colorectal neoplasia distribution by mutation type or the chance occurrence of a high risk of CRC in women. Selection bias was also a concern.

Frequency and Prevalence of Colon Adenomas in Patients with Genetic Variants

The frequency and spectrum of cancer susceptibility genetic mutations among colorectal cancer diagnosed under age 50 were studied. Patients with colorectal cancer younger than age 50 were accrued. Germline DNA was tested for mutations in 25 cancer susceptibility genes using gene sequencing. Of the 450 patients with early-onset CRC, 75 pathogenic genetic mutations were found. Thirty-six patients (8%) had Lynch syndrome. Patients with pathogenic mutations were more likely to report a family history of CRC (45.8% vs 14%; P<.001).

The frequency of the factors associated with genetic testing of first-degree relative of Lynch syndrome probands were assessed in study by Sharaf, Myer, Stave, Diamond & Lababaum, 2011. They found that 52% or less of first-degree relatives of Lynch syndrome

probands received genetic testing. Genetic testing is underutilized by first degree relatives of patients with Lynch syndrome.

Colonoscopy and pathology records of patients were studied to document the number of adenomas in patient with Lynch syndrome (Kalady, Kravochuck, Heald, Burke & Church, 2015). A total of 263 patients with a germline mutation in one of the four DNA mismatch repair genes were included. A total of 107 of the 263 patients had one or more adenomas, 61 patients had one adenoma, 29 patients had two to five adenomas, and 11 patients had 20 or more adenomas. The maximum number of synchronous adenomas was 22 and the maximum cumulative was 24. Of the 220 patients (54%) had hyperplastic polyps, 313 adenomas were found on colonoscopy, and 123 of the adenomas were advanced.

Barriers to Biomolecular Genetic Testing

Factors involved in the implementation for universal Lynch syndrome screening were evaluated via interview with health-plan and clinical stakeholders at large HMO companies. Prior to the implementing the study (Schneider, et al., 2015) estimated only five percent of CRC were screened for Lynch syndrome relying on providers or self-referrals. Open-ended interviews phase one consisted of responsibilities of Lynch Syndrome screening, perceived impact on department workflows and staff, concerns about implementing universal screening, and factors that facilitate screening. Phase 2 focused on asking about the potential to establish universal Lynch syndrome screening and their perceived value of change in the organization. Participants included representatives from pathology, oncology, medical genetics, gynecology, surgery, laboratory service, and healthcare administration.

Most people felt Lynch syndrome screening should be standard practice of the organization metric to improve CRC screening and prevention. Half or more advocated for

Lynch screening stating it is the “right thing to do” for patients. The participants in the study felt that the United States was behind on screening options for CRC and many leaders expressed that they did not know who was implementing national recommendations for CRC prevention. Barriers that were addressed were fear of health insurance discrimination, enough time with already heavy workloads on staff, and uncertainty on who would be responsible for testing.

Colorectal Cancer Screening for Average-risk adults: 2018 Guideline Update from the American Cancer Society

A recent observational study by Siegel et al. (2017) was performed to trend incidence rates of colon cancer. The study found a 51% increase in the amount of colon cancer being diagnosed in individuals under the age of 50 years old since 1994. The analysis showed that adults born around 1990 had twice the risk of colon cancer and four times the risk of rectal cancer compared to those born in 1950. Colon cancer has varied by sex, race, and ethnicity. The incidence of colon cancer is similar in men and women until the age of 35 and then increases in men. Incidence rates in African Americans are declining although, they are still historically higher than whites. Mortality rates among the white population have increased since 1995 in ages 30-39 and have decreased in African Americans since 1970. Due to the increase in mortality and incidence noticed in trends the American Cancer Society changed its recommendation to start colonoscopy screenings at age 45 instead of 50 due to the potential early detection and prevention benefit for adults ages 45-49 years of age. The USPSTF continues to recommend screening to begin at age 50 years of age and recommends flexible sigmoidoscopy every 10 years combined with an annual FIT test.

American Gastroenterology Lynch Syndrome Management

Patients who have been identified with a genetic variant are diagnosed with Lynch syndrome. These individuals develop adenomas at a younger age, the adenomas have a predominance to the ascending colon with a shorter conversion time to an adenocarcinoma. People who took up colonoscopy surveillance that were identified with having Lynch syndrome had 65% (*p=0.03)* fewer deaths in comparison to those who refused colonoscopy (Jarvinen, 1995). For the purpose of this study the American Gastroenterology Association (AGA) recommends persons who have been diagnosed with Lynch syndrome or have a first degree relative with Lynch syndrome begin colonoscopy at age 20-25 and repeat every 1-2 years. This is considered a strong recommendation, with evidence level 3 and GRADE moderate-quality (AGA, 2014)

Barriers to Research

This review of literature revealed an upward trend in the younger population particularly patients diagnosed around the 1980's. Little is known as to why this generation seems to be targeted. Genetic testing is now available and is a reliable tool to identify patients who are at risk for developing aggressive cancers at a younger age. Patients who have Lynch syndrome have a 25% risk of developing colon cancer by age 50 and a 90% chance of developing other Lynch syndrome associated cancers in their lifetime. Patients who answer positive to a medical questionnaire will benefit from genetic testing specifically if they have colorectal or endometrial cancer before the age of 50 or two or more Lynch syndrome cancers at any age (Myriad Lab, 2018). Many studies have been done assessing patients who have a known history of genetically associated cancers. However, the review of literature does not demonstrate

hereditary cancer screening as a primary prevention tool to screen and identity at risk individuals.

Chapter 3

Methodology

Purpose Statement

The purpose of this project was to determine if there is an association between hereditary cancer, gender and the incidence of colon adenomas and colon cancer in patients under the screening age of 50 years. The concept of implementing a hereditary cancer screening tool and lifestyle assessment as a predictive tool for people between the ages of 18-49 years old was explored due to data showing a rise in incidence of colon adenomas and colorectal cancer (ACS, 2018). In patients without a personal history of colorectal or another cancer but with a family history suggestive of Lynch syndrome, the American Gastroenterology Associations suggests that risk prediction models be offered rather than doing nothing (Rubenstein, Enns, Heidelbaugh & Barkun, 2016).

Objective

A quality improvement project was implemented to evaluate the feasibility and effectiveness of implementing the hereditary cancer screening questionnaire in patients between the ages of 18-49 years of age in the gastroenterology clinic. The objective was to evaluate if there is an association between a family history of hereditary cancer, gender, and premature diagnosis of adenomatous colon polyps. If association is statistically significant, this tool could be of value for primary care providers to identify people who are at higher risk for development of premature adenomas warranting referral to gastroenterology providers for further evaluation. The overall objective was to identify and reduce the incidence of colon cancer in people between the ages of 28-49 years of age. Patients deserve a personalized plan of care and need to be armed with the best tools available for the early detection of colon polyps to potentially prevent

colorectal cancer. Early detection and resection of precancerous polyps are critical to interrupt the adenoma-carcinoma sequence and prevent the development and spread of CRC (Simon, 2016). This information could be used to help improve health and quality of life and reduce healthcare costs.

Design

The project design was a retrospective correlational study to evaluate if patients who test positive to a hereditary cancer screening have a statistically significant correlation with the early development of adenomas below the screening age. The patient BMI, age, gender, number of positive factors identified on the questionnaire were recorded. The dependent variable was colorectal adenoma by colonoscopy evaluation. The independent variables tested were gender and a personal history of genetically associated cancers: colorectal, endometrial, ovarian, and gastric cancers.

Procedure

Several steps were taken in the construction of this project. A problem was identified in the gastroenterology clinic. Several patients were incidentally diagnosed with adenomatous colon polyps who underwent colonoscopy for diagnostic purposes under the age of 50 years. A review of literature was done to assess ways to identify patients who are at risk for the early development of colorectal cancer. Hereditary cancer screenings were recommended by the American Cancer Society and the National Institute of Health. The Amsterdam Criteria was chosen as a tool to identify whether a patient had relatives that were diagnosed with cancers that can be genetically associated (UpToDate, 2019). All patients were asked to fill out a hereditary cancer screening questionnaire that assessed a personal history, first degree family history,

second degree family history, and extended family history of genetically associated cancers (Appendix G).

The procedure for this project was based on the American Association of Critical Care Nurses Synergy Model. The fundamental part of the study was focused on the patient and disease prevention. Patients who underwent diagnostic colonoscopy between the ages of 18 and 49 were included. Anyone who presented for a screening colonoscopy were excluded from the study. Data were collected from the EMR GMED after colonoscopy and the results of their findings including: location of polyp, type of polyp per pathology report done by Path Group, demographics, as well as answers to their hereditary cancer screening questionnaire were imported into a password protected excel file until the project was completed. Consent was implied as patients sign a general consent when they present to the office. Patient names were replaced with case study numbers and manually entered into SPSS for statistical analysis.

Description of Measures

For the purpose of this study the incidence of colorectal adenomas was measured with a personal history of genetically associated cancers and age. Colorectal adenomas were detected by colonoscopy. Once the polyp was removed it is sent to pathology for evaluation on the type of adenoma. Adenoma progression rates vary from the site, size, and type of adenoma. A polyp starts out as a small growth of excess tissue in the lining of the colon. Not all polyps turn into cancer. There are two shapes of polyps (sessile and pedunculated); and five types of polyps (tubular adenoma, hyperplastic, serrated, inflammatory, and villous adenomas). The development of more than 70-95% of colorectal cancer can be prevented by early detection and removal of colon polyps (AGA, 2017). Sessile serrated polyps have a 20-30% chance of developing into a neoplasm (Makkar, Pai, & Burke, 2012), 91% being in the proximal colon. A

quality improvement initiative for gastroenterology has been implemented to increase the adenomas detection rate by colonoscopy to prevent colon cancer (Gurude, et al., 2018). Patients who have Lynch syndrome have an adenoma-carcinoma progression rate of 35 months versus 10-15 years (ACS, 2018). There are often no symptoms of colon polyps, making surveillance and identification of patients more important. Genes that have been identified in the development of colon cancer associated with Lynch syndrome include: MLH1, MSH2, MSH6, PMS2, and EPCAM gene (Win & Lindor, 2019).

The Amsterdam 1 criteria Hereditary Cancer Screening Questionnaire (Appendix G) was developed to identify patients who are likely to be mutation carries for Lynch syndrome. This prediction model provides quantitative estimates of the likelihood of mismatch repair mutation (American Gastroenterology Association, 2001). Patients between the ages of 18-49 who presented to the gastroenterology clinic were provided with a hereditary cancer screening questionnaire that was developed from the Amsterdam1 criteria. A detailed family history of Lynch syndrome associated cancers was documented. Patients who qualified for colonoscopy underwent diagnostic colonoscopy. The number of adenomatous colon polyps were compared to the hereditary screening survey and patients' gender. Patients who met criteria for germline testing either by questionnaire or colonoscopy were offered genetic testing. Relationships between screening factors and diagnosis of adenoma were determined by statistical analysis per SPSS software.

Protection of Human Subjects

Ethical considerations ensured minimal to no risk to study participants. Data retrieval was kept in a password protected excel document and later imported into SPSS with a case number being assigned to each patient for privacy protection. Patients were evaluated and treated in

accordance to the American Gastroenterology standards. There were no unnecessary procedures ordered on patients for the purpose of this study. Patients who scored high on the hereditary cancer screening questionnaire were educated on their risk factors and offered genetic testing if warranted. HIPPA compliance was maintained. All patients received post-procedural counseling and education on their findings. No personal patient identifiers were published. Data collection was done by the primary researcher and with the assistance of the attending physician, and medical assistants. Children were excluded from the study as the research site was credentialed to take care of adult patients only. The project was sent to the IRB committee and was approved on July 26, 2019 as an expedited review (Appendix F).

Evaluation Plan

Descriptive statistics were generated to display associations between participants, gender, and family history of hereditary cancers. Chi square goodness of fit contingency table were used to analyze the data. G-Power was set at a goal of 95% which would include 145 chart reviews to a minimum of 88 chart reviews to achieve an 80% power. Frequency tables were used to evaluate age, gender, BMI, and smoking status in correlation with the presence of colon

Chapter 4

Analysis of Results Description of Sample/Population

The sample was taken from a gastroenterology clinic in East Tennessee using G-Med and Pathogroup Electronic Medical Records (EMR). Patients who had colonoscopy exams between January 1, 2019 and December 31,2019 were considered. Exclusions for this study included anyone over the age of 49 years old, who would have been normally eligible for colonoscopy. There were 150 patients total that met criteria for the study. Of the 150 total subjects 59 (39.3%) were males and 91 (60.7%) were females. The mean age of the sample was 40.14; Median 44; Mode 48; with an age range from 18-49 years old. The average patient was obese with a mean BMI of 32.85. There were 115 (76.7%) patients who said they were non-smokers and 35 (23%) who did smoke.

Hereditary Genetic Screening Survey

Three out of 150 patients (2%) reported a personal history of cancer with breast and endometrial being the most common. Patients who reported having first degree relatives with a history of cancer included 49 (32.7%) of the sample with the most common cancer being colorectal 17 (11.3%); breast 6 (4%); followed by pancreatic 5 (3.3%); and ovarian 5 (3.5%). Second degree relatives with a history of cancer were reported by 42 (28%) of the sample with colorectal 16 (10.7%), other 10 (6.7%), and breast 8 (5.3%) being the most common. Patients who reported extended family members with cancer were 41 (27.3%) people in the study with the leading reported cancers being colorectal 11 (7.3%), breast 14 (9%); and pancreatic 6 (4%).

Pathology

Patients diagnosed with colon adenomas included 58 (38.7%) participants. The percent of patients with hyperplastic polys which have zero chance of developing into an adenocarcinoma

included 29 (19.3%) of the sample. The most common location of the adenoma was the ascending colon 21 (14%) followed by the transverse colon 17 (11.3%), and the rectum 15 (10%). The most common type of adenomas was tubular adenomas 37 (24.7%), and sessile serrated adenomas 19 (13%).

Research Problem

This study was performed to see if there was a correlation between patients answering positive to a hereditary cancer screening and the detection of early adenomas. The dependent variable was colon adenomas with the independent variables answering positive to a hereditary cancer screening. Co-variables included sex, age, smoking status, BMI, first, second, and third-degree relatives with hereditary cancer. Other co-variables included the location of the polyps, the type of pathology, and the type of cancer reported on the hereditary cancer screening tool.

Analysis of Project Question

The purpose of this study was integrative review to identify if initiating a hereditary cancer screening assessment in the clinic setting aids in early identification of patients who are at higher risk for colon cancer and adenomas below the screening age of 50 years of age. A Chi-Square Goodness of Fit test was used to calculate whether there is a statistically significant relation between having a history of heredity cancer and early onset adenomas. Analysis of personal history, first degree and higher history of hereditary cancers, second degree family history, and extended family history were completed.

Personal History of Hereditary Cancer and Adenoma Detection

Of the 150 charts reviewed, three patients (2%) reported a personal history of hereditary cancer, one with breast cancer, one with endometrial cancer, and one with “other” cancer. A chi-square goodness -of-fit was conducted to determine whether the patients that reported a personal

history of hereditary cancer had early onset adenomas. The chi-square goodness-of-fit test indicated that there is not a significant difference in the occurrence of adenoma on colonoscopy for individuals who give a positive versus negative personal history $X^2(1) = 1.012$, $p = 0.314$).

First Degree Family History of Hereditary Cancer and Adenoma Detection

Of the 150 patients reviewed, 49 (32.67%) reported a first-degree family history of hereditary cancer with pancreatic, breast, and colorectal cancer being the most common. The chi-square goodness-of-fit test indicated that there is not a significant difference in the occurrence of adenoma on colonoscopy for individuals who give a positive versus negative first degree family history,($X^2(1)$ 3.264=, p=.071)

Second Degree Family History of Hereditary Cancer and Adenoma Detection

Of the 150 patients reviewed, 42 (28%) reported a second-degree family history of hereditary cancer with colorectal, “other”, and breast cancer being the most common. The chi-square goodness-of-fit test indicated that there is a statistically significant association in the occurrence of adenoma on colonoscopy for individuals who give a positive versus negative second degree family history ($X^2(1) = 5.347$, p =0.021).

Extended Family History of Hereditary Cancer and Adenoma Detection

Of the 150 patients reviewed, 41(27.2%) reported extended family history of hereditary cancer with pancreatic, breast, and colorectal cancer being the most common. The chi-square goodness-of-fit test indicated that there is not a significant difference in the occurrence of adenoma on colonoscopy for individuals who give a positive versus negative extended family history ($X^2(1)$ .486=, p=0.486).

Additional Statistical Analysis

Gender

Of the 150 patients reviewed, 59 (39.3%) were male and 91(60.7%) were female. A chi-square goodness-of-fit indicated that gender had no effect on the occurrence of adenomas ($X^2(1) = .563$, $p = .453$)

Smoking

Of the 150 patients reviewed, 73 (48.6%) were non-smokers and 42 (28%) were smokers. A chi-square goodness-of-fit indicated that smoking status had no effect on the occurrence of adenomas ($X^2(1) = .956$, $p = 0.328$)

Prevalence Rates

Prevalence rates were calculated on total of all patients who answered positive on a heredity cancer screening questionnaire as well as the presence of colon adenomas. In addition, positive and negative predictive values were calculated. Prevalence rates of patients who reported a positive history or family history of hereditary cancer was 44.67%. The prevalence of patients who were found to have colon adenomas on a colonoscopy examination was 38.67%. The sensitivity of the questionnaire for predicting adenoma as 40.96%, and the specificity was 64.18%. Positive predictive value (PPV) was 58.62% with a negative predictive value (NPV) of 46.74%.

Table 1

	Hx +	**Hx -**
Adenoma +	34	24
Adenoma -	49	43

Unintended Consequences

During this review, Blue Cross Blue Shield of Tennessee changed the age for colonoscopy screening to start at age 45 instead of 50 years of age. This was done in response to the American Cancer Society position statement that was issued to the public on May 31, 2018. Blue Cross Blue Shield made this change effective October 1, 2019. Several other insurance companies followed the same change. The population studied were people under the age of 49 years old. The National Conference of State Legislators Colorectal Cancer Screening Laws by State (2010) state that "All individual and group plans mandate offering individuals defined by the American Cancer Society (ACS) as average risk follow in accordance with the ACS screening options," As a result, the data extracted from patient files between October 1, 2019 – December 31, 2019 reflected this change.

Summary

The sample included a total of 150 participants. Most of the sample were women. The mean BMI of the sample size was 32.3 indicating that most were obese. Most of the population were non-smokers. Adenomas (pre-cancerous colon polyps) were found in 38.7% of the sample with the most common location of the polyps being in the ascending colon. Hereditary Cancer Screening Questionnaire was done and 2% reported a personal history of hereditary cancer; 32.6% reported a first degree family history of hereditary cancer with colorectal cancer being the most common; 28% reported a second degree history with colorectal cancer being the most common; and 27.2% reported extended family history of hereditary cancer with pancreatic; breast and colorectal being the most common.

Statistical analysis indicates that there was not a significant relationship to answering positive on a hereditary cancer screening questionnaire and adenomas found on colonoscopy

with a personal history; 1st degree relatives; and extended relatives. Patients who reported yes to a second-degree family history of colorectal cancer showed a significant association with adenomas found on a colonoscopy.

Chapter 5

Discussion of Findings

Relationship of Outcomes to Research

The purpose of the study was to evaluate if answering positive to a hereditary cancer screening questionnaire was predictive of people who were at risk for the development of premature adenomas. While patients who answered positive on a hereditary screening questionnaire for personal history, first degree history, and extended history did not show an association with adenomas on colonoscopy; second degree family history did show a significant correlation. Smoking and gender also did not have a significant effect on the findings of colon adenomas.

The hypothesis was that people who fall below the screening age and answer positive to a hereditary cancer screening would be more likely to have adenomas found during colonoscopy. A statistically significant association in the occurrence of adenoma on colonoscopy for individuals who give a positive versus negative second-degree family history ($X^2(1) = 5.347$, $p = 0.021$) was found. Patients with a positive personal history, first degree relative history, and extended history did not show an increase in incidence of early adenoma. The hypothesis was partially correct in that there was a significant relationship with people who presented for early adenomas that had a second decree history of hereditary cancer syndrome.

Observations

During the study it was observed that many patients did not know their family history. Some of the patients guessed on what they thought was their family history. Many patients did not realize the relevance of genetics and chronic disease. Patients who presented with rectal bleeding were much more concerned about getting their colonoscopy done as soon as possible

than patients who presented with diarrhea, constipation, or a change in bowel habits. Many patients exhibited fear of doing a colon prep and admitted this is what deterred them from having a gastroenterology visit and colonoscopy.

Limitations

Several limitations were noted on the study. First and foremost, the sample size was relatively small to answer the research question. The knowledge of the participants on their family history was not equally distributed. Some patients were very confident of their family pedigree and others were speculating what they thought was their family history. Patients who were adopted were only able to fill out a partial family history based on their knowledge and probably should have been excluded.

Other factors that may have influenced the study include they type and quality of the colon prep that was used. The quality of the colon prep can impact the endoscopist's ability to identify colon polyps, specifically in the right colon. Patients who had co-morbid conditions were not separated from patients who were relatively healthy and presented with subtle symptoms. Additionally, the data were entered manually from one database to another. Human error in data transfer may have occurred.

Implication for Future Research

Many studies have been done to identify the cause of colon cancer and to predict people who are at risk for premature adenomas. Genetics are often used after a patient has been diagnosed with colon cancer to determine if there is a genetic variant to strategize future care and as a risk assessment for family members. Hereditary genetic testing is relatively new to gastroenterology as far as a pre-assessment for patients for colon cancer. Future research to support this would include a larger sample size and a wider demographic area.

Colonoscopy remains the gold standard for colon cancer screening. It is an invasive procedure that deters some people as they seek to avoid due to an unpleasant colon preparation and the invasiveness of the test. Virtual Colonoscopy is a CT scan that is non-invasive however and is an alternative to patients who do not want to have a colonoscopy. Future studies may include a combination of virtual colonoscopy with genetic screening to identify patients that are at risk. Another option may be to offer genetic screening to everyone at an early age to identify the high-risk patients and proceed with screening exams.

Implications for Practice/Health Policy/Education

Colon cancer can be prevented by early detection of adenomas and removing them before they convert to an adenocarcinoma. Use of a combination of known factors is needed to identify patients who are at increased risk. Gastroenterology clinics need to educate primary care to be aware of new screening tools as well as the reliability of these tools. Cologuard is often used as the preferred method of screening colonoscopy by primary care however, it is not appropriate to use in people who have a personal history of colon polyps or a strong family history of colon cancer. Providers must be credulous in their committed to continued education and keeping up with evidence-based practice.

One of the findings that was not recorded in the study that was noticed was hesitancy of the patient to come to the GI clinic based on fear of rumors of how bad the colon preparation would be and fear of colonic perforation. In response to these fears, the practice plans to launch on their website the preparation that we use and how it works. Previously patients had to drink a gallon of a hyperosmotic colon prep that made them nauseated and had a sour taste. New preps are available that are only 6 ounces of medication with most of the prep being water. The providers have also decided to create a colon preparation “survival kit” which includes flushable

wet wipes, barrier ointment, anti-emetics, and 24-hour access to a provider in the event the patient has a question or issue with the prep. Video example of the colon prep on the office smart boards provide the link so patients can watch it at home. Each patient can look at a sample of the colon prep, watch the video, speak with a nurse about the prep and go home with written instructions to resolve their fears.

During this project health care policy was used to challenge insurance companies to cover screening colonoscopy at age 45 instead of age 50 due to the new recommendation set by the American Cancer Society. The American Cancer Society changed its protocol in 2018, however, insurance companies were still not covering a screening until age 50. The local Senator, Mike Bell, was contacted via email (Appendix C) about this concern. He is appointed over the Health Board in Tennessee. Senator Bell responded to the email and ensured he would address at the Capital. Approximately one month later our office received a letter from Blue Cross Blue Shield of Tennessee that they will begin covering screening colonoscopy at age 45 according to ACS guidelines. This is an example of how nurse practitioners and legislators work together as a team to promote health for the community. This new ruling will allow colonoscopy and polyp diagnosis in the age range of 45-50 years of age and remove them before they progress to an adenocarcinoma.

Conclusion

Doctorally prepared nurses have an obligation to protect, promote, and restore health to the community. It is our duty to continue to find ways to improve healthcare through evidence-based practice and personally involvement in research. APN's must always continue to strive to find newer and better ways to educate and treat people. For the purpose of this project, there was not a strong correlation with hereditary cancer screening and the diagnosis of early adenomas.

However, there was a significance with patients who had second degree family history of Lynch associated cancer and colon cancer.

Hereditary cancer screening is a simple form that is non-invasive and may enlighten healthcare providers on patterns found in a person's pedigree. This tool would be helpful with assessment of other risk factors that are known to be linked to colorectal cancer including: diet high in red meat, alcoholism, smoking, and obesity (American Cancer Society, 2019). Having individualized care plans for each patient allows providers to strategize care based on that specific person. APNs must continue to look for better ways to practice, while being open to accepting new technology and medical advancements that may seem overwhelming and challenging.

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Appendix A

Hereditary Cancer Questionnaire

Risk Assessment for Hereditary Cancer Syndromes

Patient Name: _____ Physician: _____

Date of Birth: _____ Date Completed: _____

Instructions: Please circle Y for those that apply to YOU and/or YOUR FAMILY (on both your mother's/maternal or father's/paternal side). Next to each statement, please list the relationship to you and age of diagnosis. You and the following family members should be considered:

Mother Father Brother Sister Children Paternal Uncle/Aunt Maternal Uncle/Aunt First Cousins Niece/Nephew Maternal Grandmother/Grandfather Paternal Grandmother/Grandfather

Each statement should be answered individually, so you may list the same cancer diagnosis more than once as you answer these questions. This is a screening tool for the common features of hereditary cancer syndromes. Share this information with your healthcare professional to help determine your hereditary cancer risk.

		BREAST AND OVARIAN CANCER	SELF	FAMILY MEMBER	AGE AT DIAGNOSIS
Y	N	Breast cancer before age 50			
Y	N	Ovarian cancer			
Y	N	Two primary (unrelated) breast cancers in the same person or on the same side of the family			
Y	N	Male breast cancer			
Y	N	Triple negative breast cancer* (ER-, PR-, HER2-pathology)			
Y	N	Pancreatic cancer with breast or ovarian cancer in the same person or on the same side of the family			
Y	N	Ashkenazi Jewish ancestry with breast, ovarian or pancreatic cancer in the same person or on the same side of the family			
		COLON AND UTERINE CANCER	**SELF**	**FAMILY MEMBER**	**AGE AT DIAGNOSIS**
Y	N	Uterine (endometrial) cancer before age 50			
Y	N	Colorectal cancer before age 50			
Y	N	Two or more Lynch syndrome cancers* in the same person or on the same side of the family			

(*Lynch syndrome cancers include: colorectal, uterine/endometrial, ovarian, stomach, ureter/renal pelvis, biliary tract, small bowel, pancreas, brain or sebaceous adenomas)

		POLYPOSIS SYNDROMES	SELF	FAMILY MEMBER	AGE AT DIAGNOSIS
Y	N	10 or more cumulative (lifetime) colorectal adenomas (colon polyps)			
		MELANOMA	**SELF**	**FAMILY MEMBER**	**AGE AT DIAGNOSIS**
Y	N	Two or more melanomas in an individual or family			
Y	N	Melanoma and pancreatic cancer in an individual or family			
Y	N	Have you or any member of your family ever been tested for hereditary risk of cancer? If yes, please explain:			

Patient's Signature Date

FOR OFFICE USE ONLY

☐ Candidate for further risk assessment and/or genetic testing:
☐ HBOC ☐ Lynch ☐ Polyposis ☐ Melanoma

☐ Information given to patient to review

☐ Follow-up appointment scheduled Date: _____

☐ Patient offered genetic testing: ☐ Accepted ☐ Declined

Healthcare Professional's Signature Date

*For a better understanding of triple negative breast cancer, please ask your healthcare provider.
Assessment criteria based on medical society guidelines. For these individuals society guidelines go to www.myriadtests.com/patient_guidelines

MORA/12-11

MYRIAD

Appendix B

IRB Approval

SOUTHERN ADVENTIST UNIVERSITY

Power for Mind & Soul

July 26, 2019

Principal Investigator: Samantha L. Spinks
Research Project: Molecular Genetic Cancer Screening in the role of Prediction of Colorectal Disease in the Clinic Setting
IRB Tracking Number: 2019-2020-005

Dear Logan,

It is a delight to inform you that the Institutional Review Board examined your research study proposal and supporting documents and the IRB committee has approved your research request as **expedited.** We wish you the very best as you move forward with this study and look forward to reading your findings when they are ready.

If there are minor changes to this research, before making those changes please notify us by completing and submitting FORM B (Certification of Modification, Annual Review, Research Termination, or Research Completion). Please submit applications to irb@southern.edu. If substantial changes are planned you, as the principal investigator, should submit a new IRB FORM A application.

Many blessing to you as you move forward. Please let us know if there is anything else we can do to assist you with this research study.

Always in His service,

Cynthia

Cynthia Gettys, Ph.D.
IRB Chair
Southern Adventist University
423-236-2285
cgettys@southern.edu

Appendix C

Dear Senator Bell,

My name is Samantha L. Spinks. I am a board-certified acute care nurse practitioner in Cleveland, TN. I work at East Tennessee Gastroenterology. I am also a board member of Foundation House Ministries in Cleveland, and if I recall, you have attended one of our banquets in the past. The purpose of my letter is that I am asking for legislative help and guidance on an issue that is very important to me and my patient population. Colorectal cancer is the third most common cancer in men and women in the United States (American Cancer Society, 2017) and the second leading cause of cancer related deaths in the United States (JAMA, 2016). In 2018, based on observational study from data recorded by the Surveillance, Epidemiology, and End Results (SEER) program (1975-2014) by Siegel et al. the American Cancer Society changed its recommendations to start colonoscopy screenings at age 45 years instead of age 50 due to an increase in incidence rates and mortality in Americans under the age of 50 years of age with no epidemiological evidence explaining the cause. There has been a 51% increase in CRC in those younger than 50 years of age. As a nurse practitioner in GI, this concerns me greatly. We have recently implemented a genetics program to help identify patients who are at elevated risk due to genetic variants however, this is also insurance dependent.

Depending on state laws insurance companies cover screening colonoscopy at 45 years of age if their state follows ACS guidelines and age 50 if the state follows United States Preventative Service Task Force (USPSTF) guidelines. I have researched this information and I am finding that Tennessee follows ACS guidelines (CRC Screening Law: State of Tennessee, 2019) which states that we should order colonoscopy on average risk patients at age 45 years old. What we are finding is that some insurance companies are covering this at 100% as a screening exams and others are not adhering to this guideline and are applying the charge to the patient's deductible. This is very confusing to myself as a provider as I am trying to do what is recommended by my state and give the best care to my patient without causing them to have financial burdens. I recently had the opportunity to speak with a physician in Kentucky, Dr. Whitney F. Jones, who states that they had similar issues and that he had to reach out to a legislator for help. Now the state of KY follows ACS guidelines as is stated in the state screening laws. I was born in the city of Cleveland and I want to give the best care of the residents of this community. I realize that I am a small voice and that I have very little knowledge on health policy and legislation. However, I do feel that I have a duty to advocate for the patient population that entrust me to their care. I have always supported you as your constituent. If you have any knowledge or can help in this area, I would greatly appreciate it.

Respectfully,

Samantha L. Spinks, MSN, AGACNP-BC, CCRN
East Tennessee Gastroenterology
423-339-2000 (work)
423-716-0839 (cell)

Appendix D

Student Learning Outcomes

Cultural Competence

Cultural sensitivity was observed by understanding that each patient may have diverse beliefs, values, and strong feelings toward colonoscopy screening and genetic testing. The United States has a diverse population. Individuals have unique linguistic abilities and ethnic backgrounds. To provide effective communication literature will was provided verbally, pictorially, and written at an educational level was appropriate for reading level. Everyone had the opportunity to express their concerns and answers was provided at a level that they could understand. The purpose of the procedure was explained to the patient including risk versus benefits. Certain racial and ethnic diversities were individually addressed to ensure that all patients received patient centered care to improve the quality of care and reduce the disparity of colorectal cancer based on evidenced practice.

- Explanation of procedures was provided verbally and with literature in the preferred language of the individual.
- UpToDate database has patient information on colonoscopy in two educational formats (The Basics is at a 5th grade reading level and Beyond the Basics is above a 5th grade reading level for patients who have the aptitude skills to comprehend the data)
- Interactive wallboards in the exam rooms are available for visual demonstration of colonoscopy screening and colorectal cancer.
- Literature was provided in the primary language of the patient

- Patient-based approach assessing cross-cultural issues was addressed prior to proceeding with colonoscopy or genetic screening. Core issues warranting the need for colonoscopy were explained.
- Sensitivity to patient's preference of learning was assessed and respected.
- Clear communication, focused reassurance, and respect for the patient's perspective and concern was done to enhance the therapeutic relationship between that patient and the nurse practitioner.

Evidence Based Practice:

The purpose of this project was to determine if there is an association between patients who answer positive to a hereditary cancer screening questionnaire and the premature diagnosis of colon adenomas. Statistical analysis was performed to determine if the results of the study were significant. The American Cancer Society, in 2019, reduced the age of colonoscopy screening from age 50 to 45 on all patients regardless of gender or age based on an increase in the incidence of colon polyps in patients under the age of 50 years of age (ACS, 2019). Many risk factors for colorectal cancer have been identified including obesity, history of inflammatory bowel disease, certain ethic groups, family history of colon cancer, and genetic variants. Genetic variants that have been associated with the development of colorectal cancer (MLH1; MSH2; MSH6; PMS2) show a 11% to 47% increase in lifetime risk of developing colon cancer before the age of seventy years (UpToDate, 2019). Patients who have any of these genetic variants have a higher risk for developing genetic cancer, some of which have no screening guidelines.

- Societal Guidelines were observed in formulating a plan of care for patients.
- Hereditary Cancer Screening questionnaire is a tool to evaluate a family history of genetically associated cancers.

- If there is an association between patients that have colon adenomas and test positive on a hereditary cancer screening questionnaire, the questionnaire could be implemented in the primary care setting to identify at risk patients and refer them for assessment and evaluation to a Gastroenterology clinic prior to the screening age.

Health Promotion

Colorectal cancer is the third most common cause of cancer diagnosed in the men and women in the United States (ACS, 2019). Colon cancer can be prevented by colonoscopy and the removal of adenomatous colon polyps before they progress to an adenocarcinoma. Patients who have been identified with a genetic variant have an increased lifetime risk of the development of colon cancer at a younger age. The current screening guidelines vary on state law and are regulated by the American Cancer Society who has lowered the screening age to 45 years of age. The US Preventive Services Task Force (***USPSTF***) recommends starting screenings at age 50. Because colon polyps have no signs or symptoms, patient who develop symptoms typically have an advanced adenoma or adenocarcinoma. Identification of patients who are at higher risk for the development of colon cancer based on their genetics would allow for colonoscopy to be performed at a younger age on a yearly basis to prevent colon cancer. This would promote health by reducing the disparity of colon cancer by early recognition of at-risk patients.

Patient Centered Care

Patient centered care incorporates patient concerns with evidenced based practice. The purpose of this project is to ultimately find a tool that can identify patients who are at an increased risk for colon cancer and other genetically linked cancers. When genetic variants are confirmed it allows the patient and the provider to strategize and personalize care.

Quality and Safety

This project had no safety issues as it is a retrospective correlational study. There were no safety issues as a result of this project. The project was a correlational study designed to evaluate a relationship between a history of hereditary cancer syndromes based on a questionnaire that is filled out by each new patient and the presences of early onset adenomas found on colonoscopy. The same guidelines and principals set by the American Gastroenterology Association were adhered to regarding quality and patient safety. After completion of this project the results will be disseminated.

Informatics and Innovation

Information and innovation were used to collect and analyze data. The electronic medical record (EMR) that was utilized is called GMED. This EMR was tailored specifically for gastroenterology practices and imports data collected from colonoscopy, pathology, and chart notes into one system. The data that were collected and imported into a password protected excel document to ensure patient privacy. Once enough information was completed to sustain a reliable g-power the data were imported into SPSS software for analytical analysis.

Teamwork and Collaboration

Teamwork and collaboration were exhibited by having an open line of communication with my office personal at the University. The project was derived from data that is stored in an EMR. No assistants helped with input of the data. The original data from the hereditary cancer screening questionnaire was filled out by the patient, scanned into the chart by the front desk personnel at the office, and the colonoscopy and pathology report was interfaced into GMED once the procedure was complete and the pathologist had verified the results. Multiple people were involved in the project related to their general work roles. The office manager, overseeing

physician, clinical nurse, fellow colleagues, and faculty advisors were part of the collaborative effort to make the project a reality.

Professionalism

As healthcare evolves there is a greater need for nurses to reach the highest potential in their education and career. One day nurses can exhibit this is by improving processes that impact the care of their patient population. Nurses can have a voice in legislation that will improve policy and procedures that impact patient care. This project was focused specifically on another tool to identify patients who are at increased risk for developing adenomatous colon polyps with an end goal to reduce the prevalence of colorectal cancer. After completion of this project, and faculty review, this will be submitted to the Society of Gastroenterology Nursing Association for publication in their journal to educate colleagues on the important findings. Professionalism is being a leader in one's area of specialty and advocating not only for patients, other nurses, but for improved policy and procedures. Use of these research findings as well as other information obtained as part of the DNP program will augment professional practice in the role of a doctorally prepared advanced practice nurse.

Appendix E

The Essentials of Doctoral Education for Advanced Practice Nursing

Essential I. Scientific Underpinnings for Practice

The conceptual framework used for the design of this project as the American Association of Critical Care Nurses Synergy Model. This model recognizes that synergy is a unique relationship between nurses and patients (ANCC, 2017). The center of the model includes the patient's characteristics and the nurse's competencies. Surrounding the circle is the patient, system, and nurse exhibiting how change in function, behavior, resources, and physiological changes ultimately affect the synergist relationship between the nurse and patient.

The purpose of this scholarly project was to find another tool that will be helpful identifying patients who are at an increased risk for developing early onset adenomas, factor that can contribute to the development of early onset colon cancer in genetic variants. Genetic variants can be tested with DNA testing by saliva or blood or by tissue sample from a cancer. There are twenty-six genes that have been associated with the development of eight different cancers. People who carry the genetic variant have a 50% chance of passing the genetic variant to their children which makes hereditary cancer screening questionnaires relevant in studying if they are a reliable predictive tool to identify patients who are at risk. In this research, not only were genetic factors taken into consideration, but age, BMI, and gender were also evaluated. The goal of this project was making a positive change in gastroenterology which could prevent colon cancer by early identification.

Essential II. Organizational and Systems Leadership for Quality Improvement and Systems Thinking

One of the responsibilities of a doctoral prepared advance practice nurse is to use leadership skills to reduce healthcare dipartites. Colon cancer is the third leading cause of cancer in both men and women (ACS, 2018). In 2019, CMS MIPS registry published quality benchmarks for each specialty of medicine. Measure #185: Colonoscopy Interval for Patients with a History of Adenomatous Polyps- Avoidance of Inappropriate Use is one that is considered a high priority measure. The performance measure is based on patients over the age of 18 years of age having colonoscopy interval of every three years with a personal history of colon adenomas. Performance measures are not met if the patient undergoes an unnecessary procedure before three years for adenomas detection rate.

The purpose of this project was to assess if answering positive to a hereditary cancer screening tool has a statistically significant correlation with patients who are diagnosed with premature colon adenomas. Patients are offered genetic testing. Those that test positive to genetic variants would be excluded from QI Measure #185, therefore not affecting reported data. Individuals who are offered genetic testing and test negative for genetic variants with the presence of adenomas below the screening age would be subject to American Gastroenterology Guidelines and would not require unnecessary repeat colonoscopy for three years pending the prep was adequate, there is no presence of inflammatory bowel disease, and the adenoma was completely removed (Gastroenterology MIPS Quality Measures and Improvement Activities, 2019).

Essential III. Clinical Scholarship and Analytical Methods for Evidence-Based Practice

A problem was identified at the research practice site was of concern to the research, the attention of the primary researcher, the practice manager, and the attending physician. Many of the patient referrals that were undergoing diagnostic colonoscopy evaluation for rectal bleeding, abdominal pain, change in bowel habits, constipation, and diarrhea were incidentally found to have adenomas that had no correlation or relationship to their symptoms. Genetic testing has been widely available and utilized for breast cancer patients and in oncology after a patient has been diagnosed with cancer and the tumor has been excised. The capability of genetic testing exists and can be used as a tool to strategize and individualize patient screening exams. A review of literature was conducted and analyzed at was found that patients who have genetic variants develop adenomas at a younger age that have a much faster adenoma to adenoma-carcinoma carcinoma rate. This project will be analyzed data from the office database and input it into SPSS for statistical analysis. The findings were disseminated to improve healthcare outcomes in gastroenterology care.

Essential IV. Information Systems/Technology and Patient Care Technology

Technology and information systems were utilized throughout this project. Initially all the data was taken from the office EMR, GMED. The data were manually entered a password protected excel document for patient privacy. Once the data collection was complete the patient name was replaced with a case number and were entered SPSS for statistical analysis. Upon completion of the projected the results of the data were presented to assigned faculty advisor in a word document and power point presentation. Individuals who answer positive to a genetic screening questionnaire were flagged and genetic testing was discussed and offered.

Essential V. Health Care Policy for Advocacy in Health Care

At the doctoral level it is expected that nurses engage in politics and have a voice in policy development. I am a member of the Society of Gastroenterology Nurses Association, American Critical Care Nurses Association, and the American Nurses Association. Something that is unique to my field of work is that colonoscopy screenings vary on insurance coverage based on whether the state recognizes the recommendations by the American Cancer Society or the U.S. Preventative Services Task Force (USPSTF). The difference is vast. The American Cancer Society has lowered the overall age of colonoscopy screening to begin at age 45 while the USPSTF still recommends start at age 50. This is confusing for patients and providers. Based on my personal research I found that Tennessee was regulated by the American Cancer Society, and although this is not being publicly reported by insurance companies, they are covering screenings at age 45 as preventative with 100% coverage.

This is an example of how nurses can make an impact at the legislative level. Knowledge on state laws is pivotal to patients receiving affordable care. My goal would be that if this project is successful that I will have the information published in a peer reviewed journal and petition for legislators to allow genetic testing to be covered as a primary preventative strategy under all health care plans.

Essential VII. Clinical Prevention and Population Health for Improving the Nation's Health

The goal of this project is to reduce the health disparities related to colorectal cancer by identifying if a hereditary cancer screening questionnaire is a viable took to recognize patients who may have early onset adenomas. If so, this would decrease the mortality rate of colorectal cancer as polyps would be removed from patients who are at increased risk. In the least, patients

who are shown to be at risk will be given the option for genetic testing to confirm that they have or do not have a genetic variant putting them at a much greater risk for the early development of colon cancer. Certain ethnicities are at risk for colorectal cancer at an early age such as African American. Being male, being over the age of 50 years old, smoking, obesity, and a diet high in red meat are all risk factors. While risk factors pose a threat, genetic variants most certainly carry more weight when it comes to the probability of someone developing a cancer. Therefore, this project can potentially affect population health, as it can give us another tool to find those patients who are asymptomatic but are at increased risk due to their family history of genetically inherited cancers

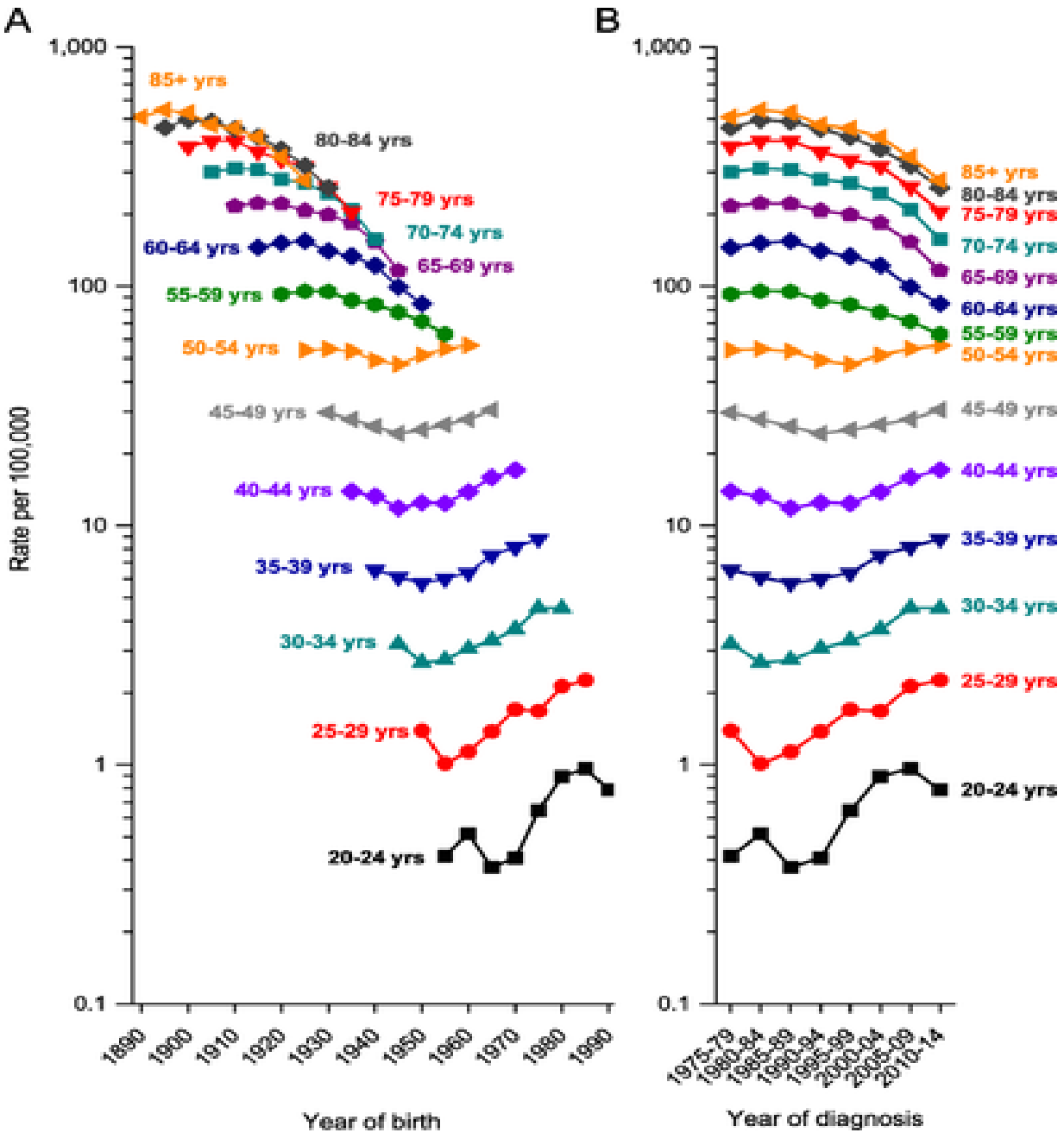
Essential VII. Advanced Nursing Practice

My doctorate is focused on Acute Care Specialization. I see primarily patents who have GI disorders in clinic and in the hospital setting. One of the most despairing news to tell a patient is that they have cancer. Particularly with patients who are young and would never be on the radar for cancer screening modalities. Pancreatic cancer, until this year, had no formal cancer screening protocols. Thanks to M.D. Anderson we now have a framework that we can utilize to screen people for pancreatic cancer based on their family history. This is the same for colorectal cancer screening. As APN's it is our duty to advocate, educate, and facilitate optimal care to ensure good patient outcomes. Conceptual and analytical skills will be used to provide statistically significant information substantiating the need to screen all individuals who may be at risk for the early development of hereditary cancers. As a nurse practitioner, it is my duty to guide my patients and provide them with the most up to date information so that as a team we can make decisions on their care together. As a DNP, I have an obligation to disseminate my

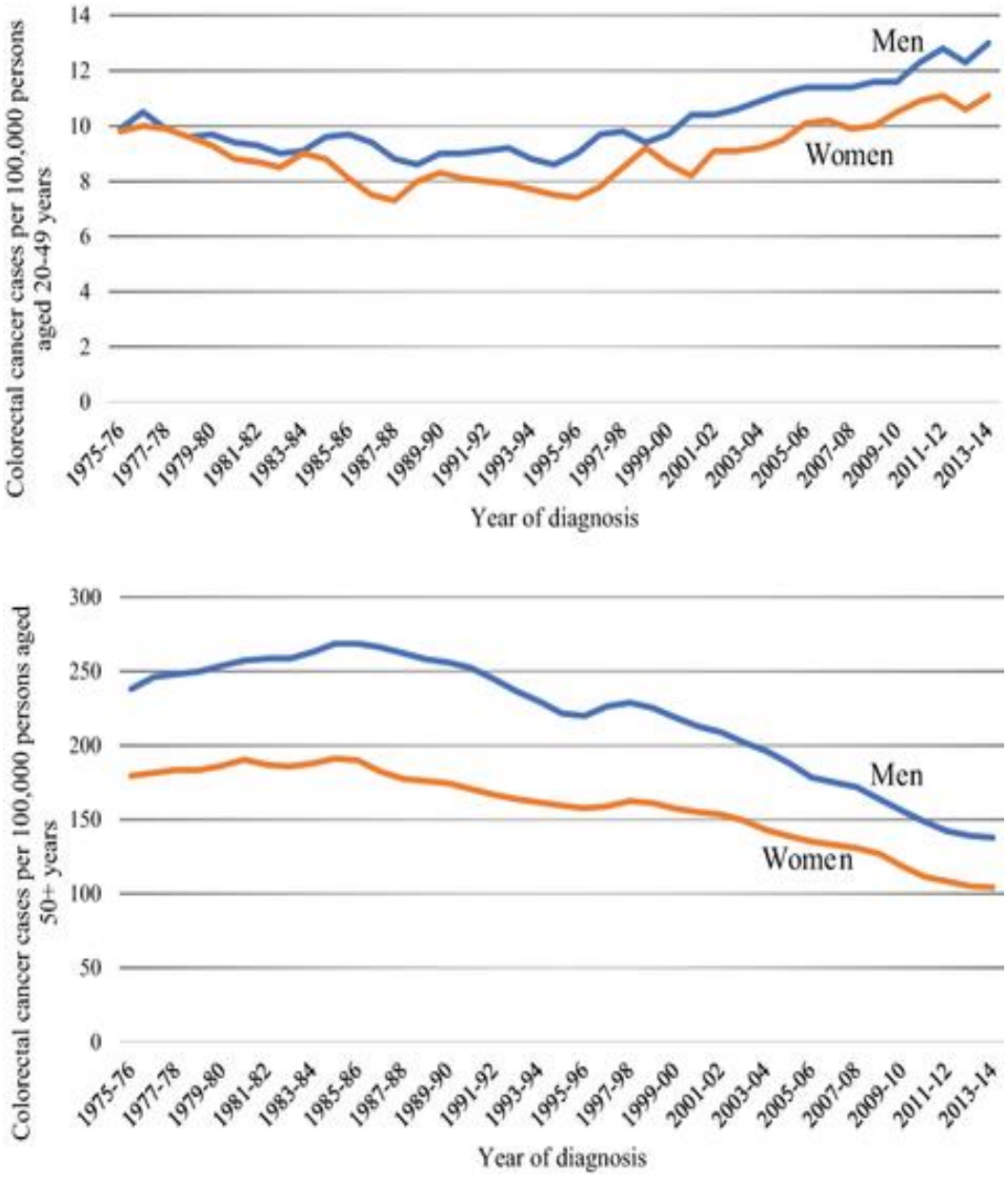
findings with other providers so that their patient population can also take advantage of the analytical data.

Appendix F

Data showing trends in Colorectal Cancer Incidence Rates by Age and Year of Birth, and by Age and Year of Diagnosis, United States, 1975 to 2014. Data Source Surveillance, Epidemiology, and End Results (SEER) program, National Cancer Institute



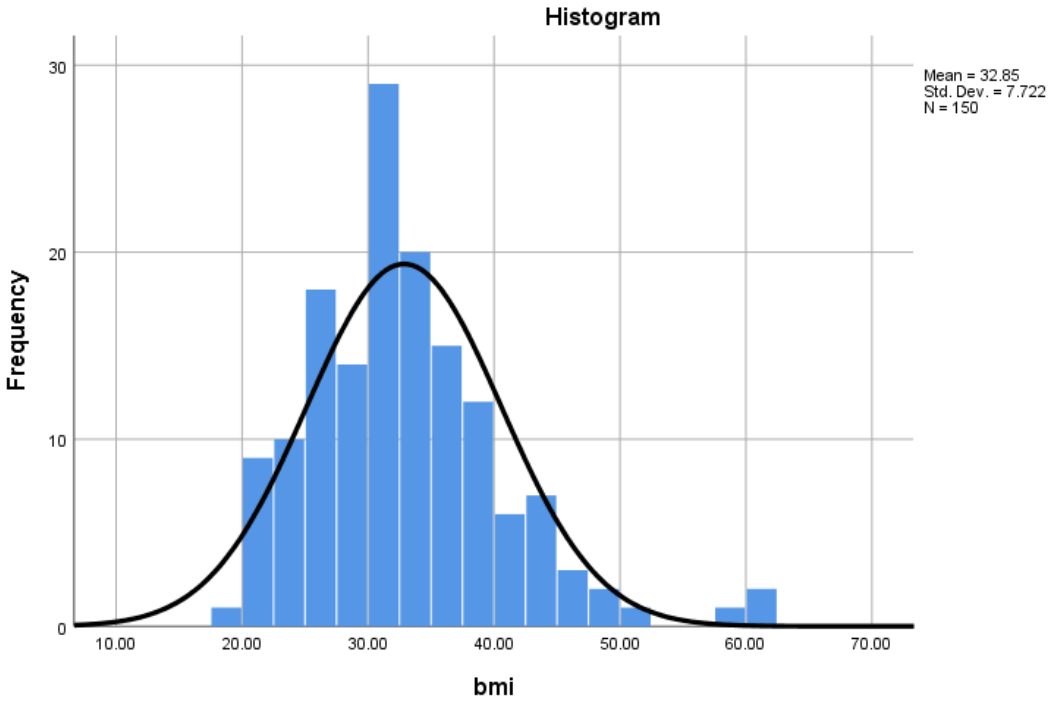
Trends in Colorectal Cancer Incidence Rates by Age (Ages 20-49) and Ages 50+) and Sex, 1975 to 2014. Rates are adjusted for delays in reporting and are plotted as a 2-year moving average. Data source: Surveillance, Epidemiology, and End Results Program, National Cancer Institute.



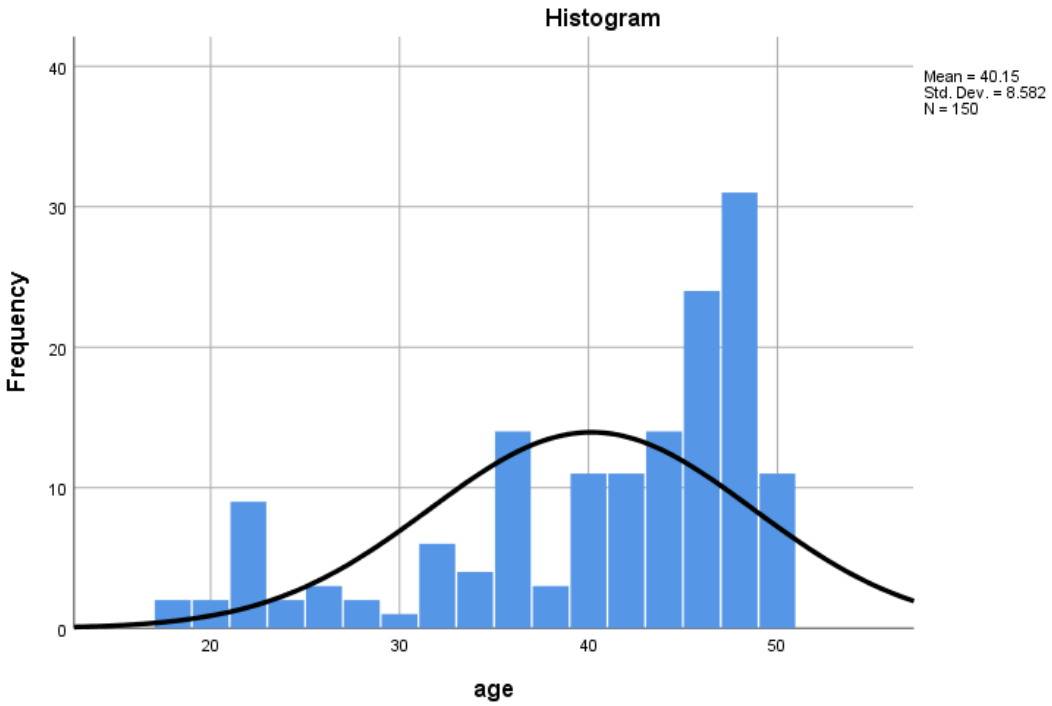
Appendix G

Statistical Charts

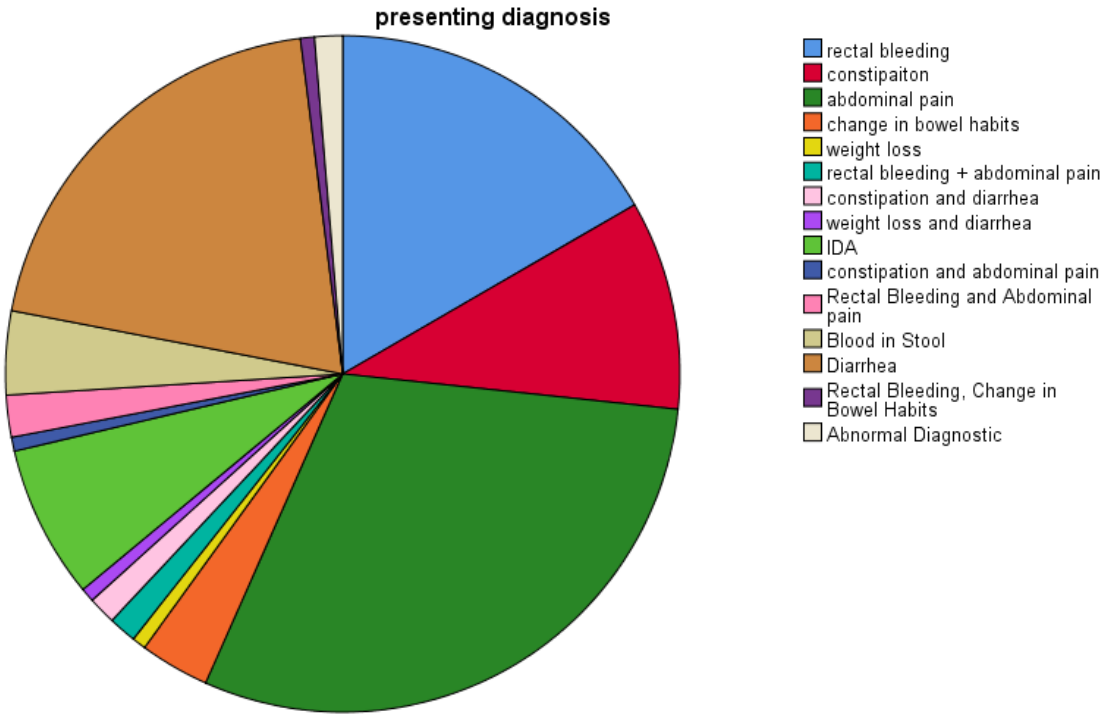
BMI



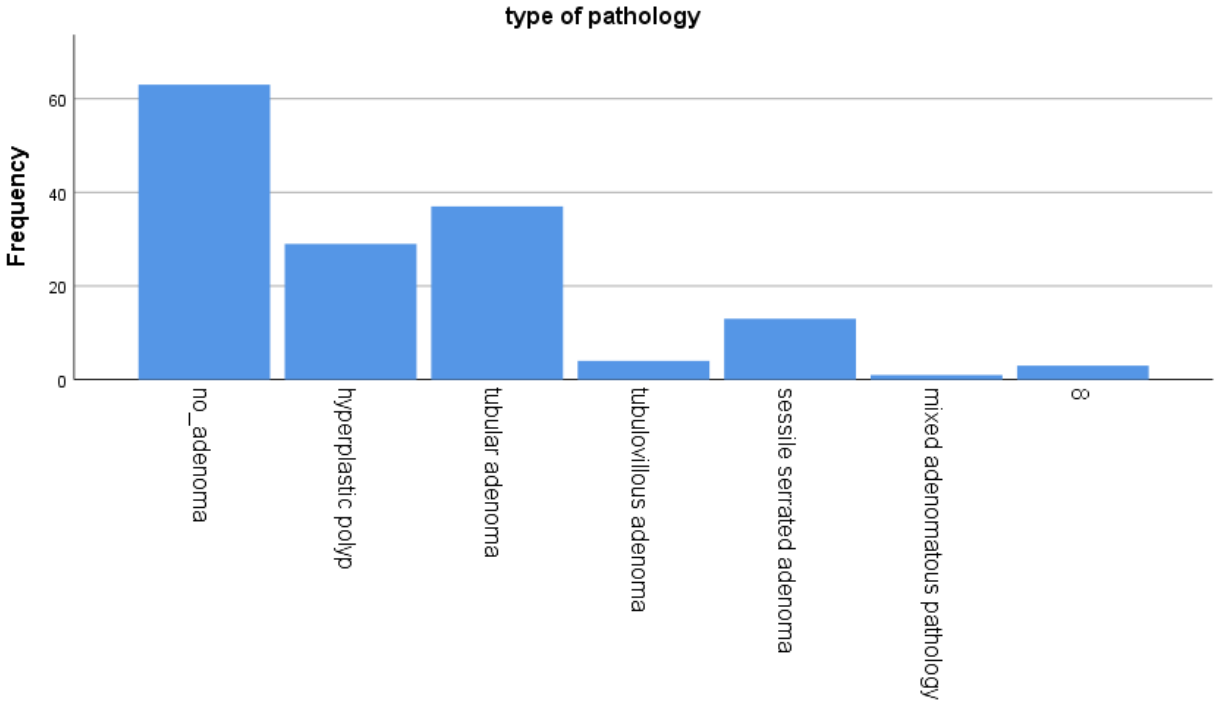
Age



Presenting Diagnosis



Pathology


type of pathology
Frequency
60
40
20
0
no_adenoma
hyperplastic polyp
tubular adenoma
tubulovillous adenoma
sessile serrated adenoma
mixed adenomatous pathology
8

Location of Polyp