FACULTY RESOURCE
CASE GUIDE

CASE: GABE
CASE DESCRIPTION:

Gabe is a 55 year old Ashkenazi Jewish male. He feels pretty healthy and seldom sees his primary care provider unless he has a problem. Prior to his recent colonoscopy, he has never had a screening test for colorectal cancer because he feels fine. Several weeks ago he noticed a small amount of blood in the toilette bowl after a bowel movement. He thought it was just hemorrhoids and was not concerned but his wife who insisted he go to see his Primary Care Provider (PCP) right away. Gabe was referred to a gastroenterologist for a colonoscopy and was found to have 11 adenomatous polyps, mostly on the right side of the colon. He is presenting to his PCP for an evaluation and to discuss the colonoscopy results. Due to his polyposis and Ashkenazi Jewish heritage, Gabe is at risk for a hereditary colorectal cancer syndrome and is referred for consideration of testing.

CASE OBJECTIVES:

- Describe components of the cancer risk assessment process.
- Construct a three generation pedigree from the information provided by the client/family.
- Identify red flags consistent with an inherited susceptibility to colorectal cancer.
- Identify indications for a genetics referral related to polyposis.
- Identify one resource for colorectal cancer screening recommendations based on the patient's cancer risk level and colonoscopy results.
- Identify two ethical, legal or psychosocial issues associated with having a possible hereditary colorectal cancer syndrome.
- Describe two resources to assist with providing patient education regarding:
  - Patient's level of colorectal cancer risk
  - Impact of being ethnicity on colorectal cancer risk
  - Health behaviors that may reduce colorectal cancer risk
- Able to utilize applicable guidelines to provide recommended best care options (i.e., USPSTF, NCCN, EGAPP).
- Describe one resource for finding genetic healthcare professionals to whom you could refer this patient.

SUGGESTIONS FOR HOW TO USE G3C:

This is an ambulatory care outpatient clinical encounter of a client who presents to his primary care provider to learn the results of a colonoscopy test to evaluate rectal bleeding. Gabe is a healthy Ashkenazi Jewish male, 55, married with three children, two sons ages 17 and 20 and a daughter age 15. He is a business man and owns several auto repair shops. Several weeks ago he noticed a small amount of blood in the toilette bowl after a bowel movement. He feels pretty healthy and seldom sees his primary care provider unless he has a problem. Prior to his recent colonoscopy, he has never had a screening test for colorectal cancer because he feels fine. He thought it was just hemorrhoids and was not concerned. This happened a couple more times the next few days. He happened to mention this to his wife who insisted he go to see his Primary Care Provider (PCP) right away. His wife usually manages the health care appointments for the family. Colonoscopy results revealed 11 adenomatous polyps found during the colonoscopy,
mostly on the right side of the colon. Further assessment reveals that Gabe is at potential risk for a hereditary colorectal cancer syndrome associated with polyposis and he is referred to a genetics specialist for further evaluation.

The learner should be instructed to enter the virtual clinic and begin by reviewing the case materials located in the clients folder. When ready the learner progresses to the client encounter and begins by selecting a question to ask the client. Additional learner activities associated with the specific questions the learners ask the client are located below the client video. Supplementary client materials including those the user gathers during the encounter are located by icons in the box to the right and can be viewed at anytime during the case.

SUGGESTED SUPPLEMENTAL LEARNER ACTIVITIES:

**Cancer Risk Assessment**
- Describe how to conduct a cancer risk assessment and evaluate Gabe's rectal bleeding.

**Reference:**
National Cancer Institute PDQ® Cancer Genetics Risk Assessment and Counseling: Components of the Cancer Risk Assessment Process Section

**Colorectal Polyps**
- Describe how to explain colon polyps to Gabe.

**Reference:**

**Risk Factors for Hereditary Colorectal Cancer Syndrome Associated with Polyposis**
- Review Gabe's pathology report and identify anything that might be a risk factor for a hereditary colorectal cancer syndrome associated with polyposis.
- Describe what questions they might ask Gabe to evaluate his risk factors for a hereditary polyposis syndrome associated with colorectal cancer risk.

**References:**
Gabe's pathology report
National Cancer Institute PDQ® Genetics of Colorectal Cancer:

**Colorectal Cancer Risk Factors**
- Identify colorectal cancer risk factors based on the Gabe's personal and family health history.
- Describe the impact of Gabe's ethnic background on his colorectal cancer risk
References:
The American Cancer Society: Colorectal Cancer: What Are the Risk Factors

American Cancer Society: Diet and Physical Activity: What's the Cancer Connection?
http://getyourscreentest.com/docroot/PED/content/PED_3_1x_Link_Between_Lifestyle_and_CancerMarch03.asp

Systematic Family History Collection
Read: National Cancer Institute: Determining Cancer Risk, Analysis of the Family History
http://www.cancer.gov/cancertopics/pdq/genetics/risk-assessment-and-counseling/HealthProfessional/page3#Section_174
- Have the learner describe how to systematically collect and analyze Gabe's family health history.

Pedigree Construction
- Construct a three generation pedigree for Grace based on her family history information using the Surgeon General's My Family Health Portrait Tool at www.hhs.gov/familyhistory
- Describe how drawing out the pedigree helps identify patterns in the health history and why it is important to use standard pedigree symbols.

Reference:

Learner Instructions for Using the My Family Health Portrait Tool at www.hhs.gov/familyhistory:
Although this tool can be used by health care providers, the primary purpose is for an individual to create a family history diagram based on their own family history. The health information and family history questions use lay language and are asked in the format one asks of an individual. For example, "How many sisters do you have." The learner should answer the questions from the perspective of the person in the case study. For "Date of Birth" on the initial screen, subtract the age of the individual in the case study from the current year for the year of birth and use 01/01 for the month and year. Unless stated otherwise in the case study, assume no one in the case study is adopted, born a twin, or has parents related other than marriage.
*Note: My Family Health Portrait version 2.4.2 draws the same pedigree symbol for both identical and fraternal twins. This will be corrected in future versions.
Colorectal Cancer Risk Calculation Using the NCI Tool

- Describe whether the NCI Tool may be underestimating Gabe's colorectal cancer risk and if so why.

Colorectal Cancer Risk Calculation Results: The information the learner needs to calculate Gabe's colorectal cancer risk includes the following: race, ethnicity, age, height, weight, vegetable consumption, history of colonoscopy and polyps, NSAID medication use, exercise pattern, tobacco use, family history of colorectal cancer.

Gabe's colorectal cancer risk calculations are as follows: 5 year risk is estimated to be 0.5% with a range of 0.3%-0.6% compared to an average risk of about 0.5%, 10 year risk is estimated risk is 1.2% with a range of 0.9% -1.6% compared to an average risk of about 1.1%, and lifetime risk is estimated to be about 5.9% with a range of 4.5% and 7.7% compared to an average risk of about 5.9%. Limitations of this risk calculation program is that Gabe's Ashkenazi Jewish heritage is not considered and the program does not consider the number of polyps a person has which is likely to result in Gabe's colorectal cancer risk being under estimated.

Colorectal Cancer Screening Guidelines for Polyposis

- Identify the appropriate colorectal cancer guidelines for Gabe based on his polyposis.
- Identify colorectal cancer screening guidelines for Gabe were he to be diagnosed with Attenuated Familial Adenomatous Polyposis.

Reference:
Guidelines for colonoscopy surveillance after polypectomy: A consensus update by the US Multi-Society Task Force on Colorectal Cancer and the American Cancer Society.

SUGGESTED CLASSROOM DISCUSSION POINTS:

- Individuals who are Ashkenazi Jewish have an increased risk of colorectal cancer.
- A personal and/or family history of polyposis can be an indication of a hereditary colorectal cancer syndrome.
- Colorectal cancer screening recommendations for individuals with polyposis can vary depending on factors such as the number and type of poyps, age of the individual, family history, and whether the person has a hereditary colorectal cancer syndrome.
- Colonoscopy with polypectomy can potentially prevent colorectal cancer in individuals with polyposis.
- Having a hereditary polyposis syndromes has implications for other family members.
WHICH ESSENTIAL GENETIC/GENOMIC COMPETENCIES DOES THIS CASE SCENARIO HELP TO TEACH?

**Professional Responsibilities Domain**

- Recognize when one’s own attitudes and values related to genetic and genomic science may affect care provided to clients.
- Examine competency of practice on a regular basis, identifying areas of strength, as well as areas in which professional development related to genetics and genomics would be beneficial.
- Incorporate genetic and genomic technologies and information into registered nurse practice.
- Advocate for the rights of all clients for autonomous, informed genetic-and genomic-related decision-making and voluntary action.

**Professional Practice Domain**

**Nursing Assessment: Applying/Integrating Genetic and Genomic Knowledge**

- Demonstrates an understanding of the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness.
- Demonstrates ability to elicit a minimum of three generation family health history information.
- Constructs a pedigree from collected family history information using standardized symbols and terminology.
- Collects personal, health, and developmental histories that consider genetic, environmental, and genomic influences and risks.
- Develops a plan of care that incorporates genetic and genomic assessment information.

**Identification**

- Identifies ethical, ethnic/ancestral, cultural, religious, legal, fiscal, and societal issues related to genetic and genomic information and technologies.

**Provision of Education, Care, and Support**

- Provides clients with credible, accurate, appropriate, and current genetic and genomic information, resources, services, and/or technologies that facilitate decision-making.
- Uses health promotion/disease prevention practices that:
  - Consider genetic and genomic influences on personal and environmental risk factors.
  - Incorporate knowledge of genetic and/or genomic risk factors
WHICH ESSENTIAL GENETIC/GENOMIC COMPETENCIES FOR NURSES WITH GRADUATE DEGREES DOES THIS CASE SCENARIO HELP TO TEACH?

**Professional Practice**

**Risk Assessment and Interpretation**

*All nurses with graduate degrees in nursing*
- Identify clients with inherited predispositions to diseases as appropriate to the nurse’s practice setting.

*Nurses with graduate degrees functioning in APRN roles also*
- Analyze a pedigree to identify potential inherited predisposition to disease.
- Use family history and pedigree information to plan and conduct a targeted physical assessment.
- Interpret the findings from the physical assessment, family history, laboratory findings, diagnostic tests, and/or radiology results that may indicate genetic/genomic disease, disease risk, or the need for a genetics/genomics referral.

**Genetic Education, Counseling, Testing, and Results Interpretation**

*All nurses with graduate degrees in nursing*
- Incorporate clients’ attitudes, values, and beliefs rooted in varying ethnic, cultural, social, and religious backgrounds when communicating genetic/genomic information.
- Provide genetic/genomic information that is appropriate to client's level of health literacy and numeracy.
- Educate clients about possible risks, benefits, and limitations of genetic testing and/or therapy.
- Support client coping and client use of genetic/genomic information in promoting health, reducing risk, managing symptoms, and/or preventing illness.

**Clinical Management**

*All nurses with graduate degrees in nursing*
- Make appropriate referrals to genetic professionals or other health care resources.

**Ethical, Legal and Social Implications**

*All nurses with graduate degrees in nursing*
- Facilitate ethical decision-making related to genetics/genomics congruent with the client's values and beliefs.
SUGGESTED READING AND RESOURCES FOR THE GABE CASE HISTORY:

**Colorectal Cancer Risk Factors, Prevention and Screening: Patient Resources**

American Cancer Society: Diet and Physical Activity: What's the Cancer Connection?

American Cancer Society: What Are the Risk Factors for Colorectal Cancer? (includes information about hereditary colorectal cancer risk factors, polyposis, and ethnic, racial background risk factors)

Genetics Home Reference: APC Gene

National Cancer Institute: What You Need to Know About Cancer of the Colon and Rectum.

National Cancer Institute: Colorectal Cancer Screening, Patient Version
(English and Spanish: includes risk factors and screening information)

National Cancer Institute: Colorectal Cancer Prevention.
http://www.cancer.gov/cancertopics/pdq/prevention/colorectal/Patient/page3


**Family History: Patient Resource**

U.S. Surgeon General's Family History Resources to Create Your Own Family History Diagram to take to your health care provider.
http://www.hhs.gov/familyhistory/

**Family History: Health Professional Resources**


U.S. Surgeon General's Family History Resources to Create Your Own Family History Diagram (available in English and Spanish, both web and printable versions). http://www.hhs.gov/familyhistory/portrait/index.html


**Colorectal Cancer Risk Assessment: Health Professional Resources**


National Cancer Institute: Colorectal Cancer Prevention: Health Professional Version http://156.40.134.52/cancertopics/pdq/prevention/colorectal/HealthProfessional

Clinical Guidelines: Colorectal Cancer Screening: Health Professional Resources


Ethnic Issues: Health Professional Resources

Genetics of Colorectal Cancer: Health Professional Resources


http://www.ncbi.nlm.nih.gov/pubmed/16454848 (abstract only)


National Cancer Institute: Genetics of Colorectal Cancer
http://www.cancer.gov/cancertopics/pdq/genetics/colorectal/HealthProfessional


Genetics of Colorectal Cancer Patient Resources

Review Genetics Home Reference "What is a gene mutation and how do mutations occur?" and think about how you would answer Gabe's question.

National Cancer Institute: Gene Testing slide show
http://www.cancer.gov/cancertopics/understandingcancer/genetesting
National Cancer Institute: Understanding Gene Testing online pamphlet
http://www.accessexcellence.org/AE/AEPC/NIH/

National Institutes of Health Genetics Working Group
Genetic Testing: What it Means for Your Health & for Your Family's Health
http://www.genome.gov/Pages/Health/PatientsPublicInfo/GeneticTestingWhatItMeansForYourHealth.pdf
Gabe's Family History

- Lila, wife, age 50 year old Ashkenazi Jewish woman who is dedicated to her family. She is not employed. She manages the household, volunteers at the high school her children attend, and is involved with activities at the Jewish synagogue to which the family belongs. She also volunteers for the local chapter of the Jewish Federation. She is also the health care manager for the family keeping track of routine appointments and health screening tests.

- Nathan, his 20 year old son, attends college and does not live at home.

- Noah, his 17 year old son is a junior in high school.

- Naomi, his 15 year old daughter is a freshman at the same high school. Both are involved in high school activities and also in youth activities through the Jewish synagogue. She has a history of asthma.

- Mother: age 78, asthma, arthritis, otherwise healthy
- Father: age 80, hypertension, memory problems, otherwise healthy

- Brother age 58, healthy, never had a colonoscopy or any type of colorectal cancer screening

- Brother age 62, had a colonoscopy at age 60 and several "growths" were found that were not cancer

- Maternal uncle: age 72, healthy

- Maternal aunt: age 73, memory problems but otherwise healthy

- Maternal uncle: age 78, diagnosed with hypertension, had some sort of "colon growths"

- Maternal grandmother: died of "female cancer" at about age 50

- Maternal grandfather: died at 67 in a farming accident

- Paternal aunt: age 77, arthritis

- Paternal uncle: age 82, has congestive heart failure

- Paternal grandmother: died at 92 of "old age"

- Paternal grandfather: died at age 87 of heart failure
Gabe's Cancer Pedigree

Paternal Side
Ashkenazi Jewish

87
CHF

d. 92

77
arthritis

82
CHF

80
hypertension

Maternal Side
Ashkenazi Jewish

d. 87
accident

d. female
cancer 50

78
asthma
arthritis

72

73

78
hypertension
colon growths

62
colon
growths 60

58

55
polyps 55

15
asthma

17

20

Male

Female

Deceased

Cancer

Married

Siblings