FACULTY RESOURCE
CASE GUIDE

CASE: MARIA
CASE OBJECTIVES:

- Describe two benefits of learning the genetics and genomics of coronary artery disease.
- Identify at least two personal beliefs or values related to genetics/genomics that may influence client care.
- Construct a three generation pedigree from the information provided by the client/family.
- Identify red flags consistent with an inherited susceptibility to coronary artery disease.
- Evaluate client’s comprehension of the possible implications of their family history for healthcare and family decisions.
- Identify the ethical, legal and psychosocial issues associated with a family history of coronary artery disease.
- Describe the option for using genetic/genomic information for determining drug dosages.
- Identify three genetic and/or genomic resources.

SUGGESTIONS FOR HOW TO USE G3C:

This is an ambulatory care clinical encounter of a new client who presents to a local family practice as a New Client because she had a full genome scan performed by a direct to consumer testing company and results revealed an increased risk of cardiovascular disease and heart attack. Further assessment including taking a detailed family history reveals a family history cardiovascular disease which also impacts her risk. She is a healthy female, 39, married for 15 years, vegetarian, marathon runner, avid exerciser, good body weight (low BMI), information seeker, has a boy-teenager and a pre-teen girl, both living with her husband and her. They live in a large city, downtown. Maria works at very stressful job as an accountant and is very computer literate. She does not have a routine primary care provider because she is otherwise healthy.

The student should be instructed to enter the clinic and begin by reviewing the case materials located in the clients folder. When ready the student progresses to the client encounter and begins by selecting a question to ask the client. Additional student activities associated with the specific questions the students ask the client are located below the client video. Supplementary client materials including those that the nurse 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 STUDENT ACTIVITIES:

Review the Genomic Compass Report
- Identify additional results that may have clinical relevance to this client.
- Review the evidence associated with the Glaucoma and Stomach Cancer findings and discuss the clinical relevance of these findings.

Pedigree Construction
- Construct a three generation pedigree using My Family Health Portrait
- Report which side of the family has cardiovascular disease in contrast to which side of the family has diabetes?
Family History Assessment
- Identify the Red Flags significant for a family history of cardiovascular disease

Reference:

Values Clarification Exercises
Note: Journaling may assist students during the viewing of G3C scenarios to explore how they view issues raised, how they feel about the patients perspective, how their own values/beliefs might influence patients decisions, and recommendations for interventions.
- Consider personal values influencing clinical decision making
- Discuss their perspective on direct to consumer genetic testing

References:
Weaver et al. (2012). Direct to Consumer genetic testing: What are we talking about? Journal of Genetic Counseling, 14, 596-593.

Skirton et al. (2012). Direct to consumer genetic testing: a systematic review of position statements, policies and recommendations. Clinical Genetics, [Epub ahead of print]

Ethical, Legal and Social Implications
- Identify critical issues associated with documentation of genetic test results in a medical record.

References:


- Describe current privacy protections for genetic information within your state and at the Federal level.
List the state regulations that protect clients undergoing genetic testing in your state and compare those protections with current Federal protections.

Reference:
Genome Statute and Legislation Database
http://www.genome.gov/PolicyEthics/LegDatabase/pubsearch.cfm

SUGGESTED CLASSROOM DISCUSSION POINTS:

- Coronary artery disease results from an interaction of genetic and environmental factors.
- Relatively uncommon genetic disorders can dramatically increase the risk of heart disease and its occurrence at an earlier age.
- A family history may identify individuals who would benefit from screening and risk reducing interventions.
- Single Nucleotide Polymorphisms (SNP) multiplex tests currently have limited predictive value for cardiovascular disease risks but this may change over time.
- Interventions are effective at reducing morbidity and mortality for high risk individuals.

WHICH ESSENTIAL 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.

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.
- Assesses clients’ knowledge, perceptions, and responses to genetic and genomic information.
- 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 interpretation of selective genetic and genomic information or services.
- 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 (e.g., a client with a genetic predisposition for high cholesterol who can benefit from a change in lifestyle that will decrease the likelihood that the genetic risk will be expressed).

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.
- 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.
- Provide anticipatory guidance to assist clients in the decision-making process related to genetics/genomics.
- Assess the clinical and psychosocial outcomes, including benefits, limitations, and risks of genetic/genomic information and/or therapies, for clients.
- Support client coping and client use of genetic/genomic information in promoting health, reducing risk, managing symptoms, and/or preventing illness.
Nurses with graduate degrees functioning in APRN roles also

- Communicate results of genetic/genomic screening and/or testing at a level that clients can understand.

**Clinical Management**

*All nurses with graduate degrees in nursing*

- Apply knowledge about the interaction of genetic/genomic and environmental factors to the care of clients.
- Make appropriate referrals to genetic professionals or other health care resources.

*Nurses with graduate degrees functioning in APRN roles also*

- Manage care of clients, incorporating genetic/genomic information and technology (e.g., risk-based genetic screening and testing, prescription of pharmacogenomic-based drugs, gene-targeted therapy, and use of genetic/genomic information in symptom management).

**Ethical, Legal and Social Implications (ELSI)**

*All nurses with graduate degrees in nursing*

- Facilitate ethical decision-making related to genetics/genomics congruent with the client's values and beliefs.

**SUGGESTED ADDITIONAL READING:**


MARIA SUGGESTED RESOURCES:

Evidence Reviews
EGAPP statement when finalized-currently under development:
Use of Genomic Profiling to Assess Risk for Cardiovascular Disease and Identify Individualized Prevention Strategies
http://www.egappreviews.org/workingrp/topics_review_cvd.htm

Background on Coronary Artery Disease
NHBLI What is Coronary Artery Disease

Direct to Consumer Genetic Testing
Secretary’s Advisory Committee on Genetics, Health and Society Report
ASHG Statement of Direct to Consumer Genetic Testing
Genetic Alliance Promotion of Genetic Testing Directly to Consumers
http://www.geneticalliance.org/ws_display.asp?filter=policy.tmarket
Genetics and Public Policy Center Issue Brief Direct-to-consumer genetic testing: empowering or endangering the public?
American College of Medical Genetics Statement on Direct-to-Consumer Genetic Testing
http://www.acmg.net/AM/Template.cfm?Section=Policy_Statements&Template=/CM/ContentDisplay.cfm&ContentID=2975

Genetics of Cardiovascular Disease
CDC Genomics and Population Health: Genetics and the Prevention of Coronary Heart Disease
http://www.cdc.gov/genomics/about/reports/2003/chap07.htm
US Preventative Services Task Force Recommendations
http://www.uspreventiveservicestaskforce.org/uspstf/uspsacad.htm

Family History Assessment

Surgeon General’s Family History Tool  
http://www.hhs.gov/familyhistory/

Guidelines and Tools to Assess Family History for Common Diseases  
http://www.genome.gov/27527602

NIH State-of-the-Science Conference Statement: Family History and Improving Health  

What Constitutes a Significant Family History of Cardiovascular Disease  

Risk Assessment  
Risk Assessment Tool for Estimating 10-year Risk of Developing Hard CHD (Myocardial Infarction and Coronary Death)  

Reynolds Risk Score  
http://www.reynoldsriskscore.org/home.aspx

Clinical Practice Guidelines  
Cardiovascular Risk Reduction-Adults  

American Heart Association and American College of Cardiology Foundation Joint Guidelines  
http://circ.ahajournals.org/content/124/22/2458  
http://circ.ahajournals.org/content/122/25/e584.full.pdf

Genome-Wide Scans  

Pharmacogenetics and Pharmacogenomics  
Realizing the Potential of Pharmacogenomics: Opportunities and Challenges

One Size Does Not Fit All: The Promise of Pharmacogenomics

Clarification of Optimal Anticoagulation Through Genetics (COAG)
http://www.clinicaltrials.gov/ct2/show/NCT00839657


Genetic Discrimination
http://www.nursingworld.org/ MainMenuCategories/ANAMarketplace/ANAPeriodicals/OJIN/ TableofContents/Vol142009/No2May09/Articles-Previous-Topics/The-Genetic-Information-Nondiscrimination-Act-GINA.aspx

Genetic Discrimination
http://www.genome.gov/10002077