FACULTY RESOURCE
CASE GUIDE

CASE: LISA
CASE DESCRIPTION:

Lisa is a healthy 19 year old woman who has come to the hospital to visit her 21 year old brother, Mark, who has cystic fibrosis (CF) and has been admitted with a lung infection. She has recently learned that she is pregnant but has not told anyone in her family because she is not married and in her first year of college. She has tried not to think too much about the pregnancy because she knows that her parents would not approve, but understands that she needs to start prenatal care soon because she plans to keep the baby and wants it to be healthy. She knows that her baby is at increased risk for having CF and is worried because she’s watched her brother struggle with the disease his whole life. She does not have any CF symptoms and she thinks she had a negative sweat test when she was a child, but is afraid to ask her parents because they would wonder why she wants to know. She has read about CF and knows that her brother’s test identified one \textit{CFTR} gene mutation that is fairly common in persons with CF, delta f508, but the other mutation was an unknown. She knows that some genetic tests are ordered during pregnancy and wonders if CF is one of them.

She also wonders what a negative CF test result would mean in her case since information about her partner is incomplete. She has no information about the baby’s father’s family health history, but she knows he wants her to have an abortion, which is why she’s not even talking to him right now; making it difficult to get that information. She knows she doesn’t want to have a baby with CF though, because she doesn’t want the baby to go through what her brother is going through, and doesn’t think she has the strength to raise a baby that’s sick so much. If she could find out if the baby has CF early enough, she might be able to terminate before her parents even found out about the pregnancy.

Lisa is also sensitive about her brother’s feelings. They have always been close. If she decides to have an abortion because of her fears about raising a baby with CF, she’s concerned he may feel hurt and unvalued because he has CF. Her parents already express guilt about having caused Mark to have inherited this illness. She definitely doesn’t want them to know about what she is considering, it would only make it harder.

CASE OBJECTIVES:

- Construct a three generation pedigree from the information provided by the client.
- Evaluate client’s comprehension of the possible implications of a germline CFTR mutation for cystic fibrosis.
- Evaluate client’s comprehension of the possible healthcare implications of an inherited CFTR mutation for her.
- Identify the ethical, legal and psychosocial issues associated with a possible CFTR mutation in the baby.
- Identify the ethical, legal and psychosocial issues associated with genetic testing for reproductive decisions.
- Describe the option for genetic testing for a known/unknown CFTR mutation in the family.
- Recognize when one’s own attitudes and values may affect care provided to clients.
- Identify three genetic and/or genomic resources on Cystic Fibrosis.
• Utilize applicable guidelines to provide recommended best care options (i.e., American Congress of Obstetricians and Gynecologists [ACOG]).
• Identify client who may benefit from specific genetic and genomic information and/or services based on assessment data.
• Describe one resource for finding genetic healthcare professionals to whom you could refer this patient.

SUGGESTIONS FOR HOW TO USE G3C:

This is an inpatient encounter with a family member of an admitted patient.

The student should be instructed to enter the virtual 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 student asks the client are located below the client video. Supplementary client materials including those that the healthcare provider gathers during the encounter are located by icons in the box to the right and can be viewed at anytime during the case.

Terminology:
Gene mutation: A change in the DNA sequence that makes up a gene. Gene mutations that are inherited from a parent are hereditary gene mutations. Some gene mutations can increase a person's risk for certain cancers.

Cystic Fibrosis (CF): Is an inherited disease of the mucus glands that affects many body systems. Common signs and symptoms include progressive damage to the respiratory system and chronic digestive system problems. Mutations in the CFTR gene cause cystic fibrosis.

CFTR (Cystic Fibrosis Transmembrane Conductance Regulator): A gene that when mutated is associated with CF and congenital absence of the vas deferens (CAVD) which causes infertility in men.

Autosomal Recessive Inheritance: Inheritance pattern associated with genetic conditions that occur only when mutations are present in both copies of a given gene (the gene inherited from the mother and the gene inherited from the father). CF is an autosomal recessive genetic condition.

Carrier: Inherited only one copy of the mutated gene from one parent, the other copy does not have a mutation. Carriers typically do not show signs and symptoms of CF.
SUGGESTED SUPPLEMENTAL STUDENT ACTIVITIES:

**Facts about Cystic Fibrosis (CF)**
Read about CF and have the student create a short summary that describes what they understand about the implications of having CF:


http://learn.genetics.utah.edu/content/disorders/whataregd/cf/

http://cysticfibrosis.about.com/od/cysticfibrosis101/a/CFgenetics.htm

http://www.genome.gov/10001213

- What is the Mendelian pattern of transmission associated with mutations in the CF gene?
  - CF is associated with the presence of two disease-causing mutations in the CFTR gene (i.e., autosomal recessive).
- What is the probability that Lisa has one of the same CF mutations as her brother? What are the possible outcomes of CF test results for Lisa?
  - At conception, each full sibling of an affected individual has a 25% chance of being affected, a 50% chance of being an asymptomatic carrier, and a 25% chance of being unaffected and not a carrier.
  - For an at-risk sibling who is known to be unaffected but has not yet undergone molecular genetic testing, the risk of being a carrier is 2/3.

**Genetics Specialist Career Opportunities**
Visit genomic careers site and identify two potential careers that would focus on genomics. Identify why at least two people were motivated to explore such career opportunities.
www.genome.gov/GenomicCareers/index.cfm

**Cystic Fibrosis Testing**
Read the guidelines identified below and have the student craft a response to whether or not CF testing will be offered as part of her prenatal visit work-up?


http://www.acog.org/Resources_And_Publications/Committee_Opinions/Committee_on_Genetics/Update_on_Carrier_Screening_for_Cystic_Fibrosis

http://www.cff.org/AboutCF/Testing/GeneticCarrierTest/

http://www.uptodate.com/contents/cystic-fibrosis-prenatal-genetic-screening (may require a subscription)
Read the FAQ identified below and have the student craft a response to whether or not CF testing can be done as part of the testing for the fetus?
http://www.acog.org/~media/for%20patients/faq171.ashx

**Family History**
Have the student create the pedigree using the Surgeon General's family history tool
https://familyhistory.hhs.gov
Is there anything in the family history besides CF that Lisa should be paying attention to?

Discuss with the student their perspective on the importance of ethnic background in understanding CF testing.

- More than 10 million Americans are carriers of one mutation of the cystic fibrosis (CF) gene, including one in 29 Caucasian Americans. In other races or ethnicities, one in 46 Hispanic Americans, one in 65 African Americans and one in 90 Asian Americans carry a mutation of the CF gene. The mutations screened by genetic testing vary according to a person's race or ethnic group, or by the occurrence of CF already in the family.

https://www.23andme.com/health/Cystic-Fibrosis/

http://www.acog.org/Resources_And_Publications/Committee_Opinions/Committee_on_Genetics/Update_on_Carrier_Screening_for_Cystic_Fibrosis

**Recent Advances in CF**
Explore the advances that have occurred with CF treatment by searching the internet, doing a literature search, checking out new drugs approved by the FDA, and/or visiting CF consumer sites. Create a summary of advances that could be shared with Lisa.
http://www.acog.org/~media/for%20patients/faq171.ashx

**Psychosocial and Ethics Assessment**
Read the article below and have the student identify other potential factors that may influence Lisa’s decision-making.

**Reference:**

Have the student take time to write down their feelings in response to Lisa’s comments about abortion. Describe how it makes them feel when she talks about termination of the baby. Then reflect on what they would say/not say to Lisa in response. How difficult is it to discuss such difficult subjects with clients and not share their values?
**Resource Reviews**
Have the student review these materials and provide information about whether or not they would you recommend these Resources for Lisa?


**SUGGESTED CLASSROOM DISCUSSION POINTS:**

- More than 10 million Americans are carriers of one mutation of the cystic fibrosis (CF) gene, including one in 29 Caucasian Americans. In other races or ethnicities, one in 46 Hispanic Americans, one in 65 African Americans and one in 90 Asian Americans carry a mutation of the CF gene.
- CF is associated with the presence of two disease-causing mutations in the CFTR gene (i.e., autosomal recessive).
- In some CF symptomatic individuals, only one or neither disease-causing mutation is detectable.
- The mutations screened by genetic testing vary according to a person's race or ethnic group, or by the occurrence of CF already in the family.
- At conception, each full sibling of an affected individual has a 25% chance of being affected, a 50% chance of being an asymptomatic carrier, and a 25% chance of being unaffected and not a carrier.
- For an at-risk sibling who is known to be unaffected but has not yet undergone molecular genetic testing, the risk of being a carrier is 2/3.
If the disease-causing CFTR gene mutation has been identified in the proband, it is most informative to test sibs by molecular genetic testing. Otherwise, sweat chloride testing should be performed.

- CFTR mutation carriers are generally asymptomatic.
- CFTR mutations are associated with CF and congenital absence of the vas deferens (CAVD) which results in infertility in males.

**WHICH ESSENTIAL COMPETENCIES DOES THIS CASE SCENARIO HELP TO TEACH?**

**Professional Responsibilities Domain**
- Recognize when one’s own attitudes and values related to genomic science may affect care provided to clients.
- Advocate for clients’ equitable and informed access to desired genomic services and/or resources including support groups.
- Incorporate genetic and genomic technologies and information into registered nurse practice.
- Advocate for the rights of all clients for autonomous, informed genomic-related decision-making and voluntary action.

**Professional Practice Domain**

**Nursing Assessment: Applying/Integrating Genomic Knowledge**
- Demonstrates an understanding of the relationship of genomics to the healthcare continuum, and quality outcomes.
- Demonstrates ability for health risk assessment 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.
- Conducts health and physical assessments which incorporate knowledge about genetic, environmental, and genomic influences and risk factors.
- Analyzes the history and physical assessment findings for environmental, and genomic influences and risk factors.
- Assesses clients’ knowledge, perceptions, and psychosocial responses to genomic information.
- Develops a plan of care that incorporates genomic assessment information.
Identification

The registered nurse:
- Identifies clients who may benefit from specific genomic information and/or services based on assessment data.
- Identifies credible, accurate, appropriate, and current genomic information, resources, services, and/or technologies specific to given clients.
- Identifies ethical, ethnic/ancestral, cultural, religious, legal, fiscal, and societal issues related to genomic information and technologies.
- Recognizes issues that affect the rights of all clients for autonomous, informed genomic-related decision-making and voluntary action. (e.g., military, vulnerable groups)

Referral Activities

The registered nurse:
- Facilitates referrals for genomic services for clients as needed.

Provision of Education, Care, and Support

The registered nurse:
- Uses health promotion/disease prevention practices that:
  - Consider genomic influences on personal and environmental risk factors.
  - Incorporate knowledge of 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).
- Provides clients with interpretation and/or access to interpretation of genomic information or services.
- Provides clients with credible, accurate, appropriate, and current genomic information, resources, services, psychosocial support and/or technologies that facilitate informed decision-making.
- Support patients and families to be informed decision-makers.
- Uses genomic-based interventions, treatments, and information to improve clients’ outcomes.
- Evaluate impact which may include effectiveness, safety, and psychosocial responses of genomic technology, information, interventions, and treatments on clients’ outcome.
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.
- 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.

*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.
ADDITIONAL SUGGESTED READING AND RESOURCES:

Cystic Fibrosis Foundation
http://www.cff.org

Testing for CF
http://www.cff.org/AboutCF/Testing/
http://www.cff.org/AboutCF/Testing/GeneticCarrierTest/

The mutations screened by the test vary according to a person's race or ethnic group, or by the occurrence of CF already in the family. More than 10 million Americans are carriers of one mutation of the CF gene, including one in 29 Caucasian Americans. In other races or ethnicities, one in 46 Hispanic Americans, one in 65 African Americans and one in 90 Asian Americans carry a mutation of the CF gene.

PubMed Summary

MayoClinic Summary
http://www.mayoclinic.com/health/cystic-fibrosis/DS00287

Genetic testing

Genetics Home Reference Summary

Dolan Learning Center
http://www.ygyh.org/cf/whatisit.htm

March of Dimes
http://www.milesforbabies.org/pnhec/159_525.asp
Lisa’s Family History

Mother: age 42, healthy
Father: age 44, dx diabetes age 44
Children: age 19, currently pregnant, no other health issues
Siblings: Brother, Mark, age 21, CF dx at birth
Maternal Grandmother: age 63, healthy
Maternal Grandfather: age 65, dx hypertension
Paternal Grandmother: age 65, dx 50
Paternal Grandfather: age 70, sx of dementia