

**Activity 2.1.1 Chronicles of a Genetic Counselor**

Introduction

Both of the Smith brothers are faced with difficult questions regarding the health of their future offspring. James and his wife will soon be having a new baby. Aaron and his wife are hoping to become parents. Tests that screen for abnormalities in the genes can provide information about their children before they are conceived or before they are born. They both hope that a genetic counselor can offer advice and help them navigate their reproductive choices. A genetic counselor can help a family understand the risks of having a child with a genetic disorder, the medical facts about an already diagnosed condition, and other information necessary for a person or a couple to make decisions suitable to their cultural, religious and moral beliefs.

Scientists have worked tirelessly to decode our genetic code. The goals of the Human Genome Project were to determine the sequence of the three billion base pairs that make up human DNA, identify all of the genes, and devise a method to store and analyze all of the data. The completion of this project in 2003 gave us the “parts list” for a human being. While scientists are still working to figure out how all of these parts fit together, they have learned the specific function of many genes in our genetic code, as well as how these genes determine traits and sometimes even signal disease.

Our genome consists of over three billion base pairs. Surprisingly, less than one percent of these A’s, C’s, G’s and T’s differ from person to person. Differences in our DNA help code for our unique appearance, personality, traits, and even our susceptibility to disease. Genetic diseases and disorders are illnesses that originate in our chromosomes and DNA. These genetic changes can be passed down from parent to child. Modern biology has given scientists the tools to examine changes in our DNA and to test for the presence of thousands of these genetic diseases.

*Genetic testing* is the use of molecular methods to determine if someone has a genetic disorder, will develop one, or is a carrier of a genetic illness. Genetic testing involves sampling a person’s DNA and examining the chromosomes or genes for abnormalities. The abnormality may be large – a large piece of a chromosome or a missing chromosome may be viewed under a microscope. Other abnormalities may be the result of a single base pair change – substitutions, deletions, or additions – in a single gene. Information from the Human Genome Project is expanding the uses of genetic testing to examine not only reproductive risks, but adult-onset conditions such as Huntington’s disease, mental illness, and even cancer.

In this activity, you are in the final steps of your training to become a genetic counselor. As a practical exam, you are asked to chronicle the case of one of your clients. Your notes on the case should showcase your expertise, your compassion, and your ability to provide clear, understandable options and advice. You will be assessed on the depth of your research, the sensitivity you show towards your client and the thoroughness of your analysis and recommendations. It is your final exam and you want to impress.

Equipment

* Computer with Internet access and presentation software
* Activity 2.1.1 Student Response Sheet
* Genetic counseling case file
* Laboratory journal
* Career journal

Procedure

Part I: Introduction to Genetic Testing and Screening

1. Obtain a Student Response Sheet from your teacher.
2. Fill out the Genetic Testing Pre-Survey on the Student Response Sheet.
3. Discuss your responses with the class as directed by your teacher.
4. Take notes as the teacher presents the Genetic Disorders presentation. Record information on the Student Resource Sheet as the teacher reviews the slides and shows various video clips.
5. Work with a partner to match each of the disorders listed in Step 6 to one of the classifications of disorders presented in the presentation. Be specific about how this disease is inherited or passed on and whether or not this disorder is the result of abnormalities in a gene or on a chromosome. Think about how these abnormalities affect function and describe how the disease affects function.
6. Use the Internet to investigate the following genetic disorders.

* Duchenne Muscular Dystrophy
* Cystic Fibrosis
* Huntington’s Disease
* Down Syndrome
* Leber hereditary optic neuropathy
* Alzheimer’s Disease

1. Compile your research on the Student Response Sheet.

Part II: Genetic Counseling

1. Use the Internet to research a career in genetic counseling. In your career journal, write a one paragraph job description for a genetic counselor. Cite your sources properly!!
2. Using the list of genetic disorders you have investigated in Part I, brainstorm how a genetic counselor might help in the case of two of the disorders. Record your answers in your career journal.
3. Watch the NOVA – Cracking the Code video clip Chapter 3 of 12: One Wrong Letter available at <http://www.pbs.org/wgbh/nova/genome/program.html>. Pay attention to the inheritance of the Tay Sachs disease.

http://www.pbslearningmedia.org/resource/tdc02.sci.life.gen.onewrong/one-wrong-letter/

http://www.pbs.org/wgbh/nova/genome/media/2809\_q056\_03.html

1. In your career journal, create a small pedigree for the family in the video which shows how the disease was passed from parent to offspring.
2. Explain how a genetic counselor may have helped the family showcased in the video. Write your response in your career journal.
3. Discuss with your classmates, as directed by your teacher, if the video clip changed any of your responses on the Genetic Testing Pre-Survey.
4. Obtain a genetic counseling case file from your teacher. Each file provides background information on a person or a family that is seeking your advice and expertise. You will work on the case as a member of a team.
5. Silently read the genetic counseling case. Your team’s job is to investigate the disease or disorder in question and decide how to counsel the family. Remember, you are not telling them what to do. You are providing information and options as well as answering their questions.
6. Research the genetic disorder/condition of concern. Learn all you can about the cause(s) of the disorder, the pattern of inheritance, the role genetic testing can play in this case, the symptoms and prognosis of the disease, as well as the severity of the conditions. Use the websites listed below or other reliable sources you may find to begin your research:

* On-line Mendelian Inheritance in Man (OMIM) <http://www.ncbi.nlm.nih.gov/sites/entrez?db=omim>
* Dolan DNA Learning Center – Your Genes, Your Health <http://www.ygyh.org/>

1. Take notes in your laboratory journal as you research and discuss the illness. Cite your sources.
2. Think about how you would counsel your patient. Your job is to produce the case notes or chronicles of a genetic counselor. A colleague should be able to pick up your notes, read your recommendations and be able to counsel the family. Impress your employer with how much you know! These notes should include:
   * A brief description of the disorder or disease in question. REMEMBER: You will be speaking to a family that may have little or no background in science or genetics. Be clear and descriptive and use appropriate and well-explained medical terminology.
   * A pedigree showing inheritance or a karyotype of the disorder as applicable to the case. Make sure to explain to the family what this diagram tells them about the likelihood of disease.
   * A list of at least three questions the couple or patient is most likely going to ask you about the situation. Provide a brief response to each question using a different color pen or text.
   * A case recommendation. Using information you have learned about the case, provide recommendations to the patient or family. Make sure to reference their personal situation and to provide options. Refer to the available types of genetic testing that are applicable to the case. Your job is to help the patient or family understand their risks and their reproductive options and to discuss options for testing, treatment, or living with this disease. Be sure to discuss potential ethical or moral issues associated with genetic testing and screening. Your recommendations should help the parents or the patient make well-informed decisions related to his or her own health and the health of their offspring.
3. Present your case to the class in an informal 5-10 minute presentation. Speak to the class as you would your client. You may use up to four presentation slides or visuals to display key information about your case.
4. As other groups present their cases, write down the name of the disorder and at least three bullet points about the case in your laboratory journal.
5. Describe what you would do if you were faced with the situation presented in each case. Write your response in your laboratory journal under the bullet points. If you are comfortable discussing your feelings on the case, share your thoughts with the class.
6. Type up the case notes for the case your team worked on and return your formal report in the original case file.

Conclusion Questions

1. How would a genetic counselor use a pedigree to help his or her clients understand genetic diseases?
2. Compare and contrast preimplantation genetic diagnosis and fetal testing. Does one type of test raise more ethical concerns than the other? Explain your thinking.
3. Explain why parents may be fearful or apprehensive about genetic testing.
4. How might factors such as income and access to medical insurance, influence a couple’s genetic testing and reproductive decisions?
5. Why are studies of identical twins raised in different environments extremely valuable in understanding the causes of a disease?
6. Recall the molecular tools you studied in PBS and HBS – Gel Electrophoresis, PCR, and Restriction Analysis. How do you think these techniques and procedures can be used to test human genes?

Web Portfolio

Document your work in Part II: Genetic Counseling

1. Discuss a career in genetic counseling
2. Do a write up of the genetic counseling case file (be creative)