

Genetic Diseases

INTRODUCTION

Oral presentation/skit (5-10 minutes) In lab Oct 12-14

This activity is designed to get you thinking about ethics and human impact of genetic diseases: how they effect carriers (people who carry the usually recessive disease genes but don't express the disease), how they effect the people with genetic diseases, the decisions that families must make, and the decisions that doctors must make when advising their patients.

You will work together in groups of 2-3. Each group will be given a particular scenario/genetic disorder. Or if you want to study a particular disease, talk to your TA so she can ensure there won't be two groups with the same disease. Each member of the group will choose the role of Doctor, genetic counselor, parents, sibling(s) or the affected individual as the case requires.

In cooperation with the other students, you need to research the disorder, make informed decision(s) based on consideration of the ethical issues involved, and give a 5-10 minute oral presentation to the class, preferably in the form of a skit. Your presentation will explain the scenario you were given, the symptoms of the disease, the ethical issues involved, and the decisions that were made by your group. You will defend your decision and answer questions posed by your peers. **Questions during the presentations are encouraged.**

What is a Genetic Counselor? Someone with a graduate degree in genetic counseling. They have knowledge of the genetics and biological symptoms of genetic diseases, as well as their psychological effects, and have good skills in communicating with both affected families and doctors. They provide information and psychological support to affected families and serve as patient advocates and refer individuals and families to community or state support services.

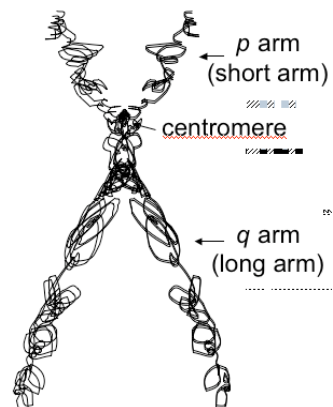
Where to find information?

Some genetic diseases are described in your textbook. A Google search will provide a lot of useful information. For many genetic diseases, there are family support associations with very useful information. You will need to decide what web pages provide credible information.

The National Center for Biotechnology Information (NCBI) also provides useful information on all common genetic diseases <http://www3.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM>

At the top of the page you can search OMIM (Online Mendelian Inheritance in Man) for genetic diseases by name or by OMIM number.

If I search for "breast cancer," this site will reveal genes that are known to be involved in breast cancer. For instance, if I click on [+113705](#) I can learn about the gene called "BREAST CANCER 1" (abbreviated as BRCA1). This was the first gene identified, which plays a role in someone's chance of developing breast cancer. The page says "Gene map locus [17q21](#)", which means that in humans, the gene is on the long (q) arm of chromosome 17, 21 centiMorgans (cM) away from the centromere. OMIM also provides references and links to the primary literature.



Some relevant things to consider:

What is the genetic basis of the disease? Is it dominant, recessive, sex-linked or other? Is it a single-locus disease or a multi-locus disease? What is the penetrance (does everyone with the gene(s) get the disease)? Does everyone with the disease have the same mutation(s)? What gene(s) are most commonly involved, or what kind of polypeptide do they encode?

What are the symptoms of the disease? Can patients live normal lives? Do they have normal life-spans? If someone knows they are a carrier, should they have children? Is the disease associated with particular groups of people? What is the financial burden to families with this disease?

EVALUATION of Genetic Diseases

Group members: _____

Oral Presentation (5-10 min; 30 points) :

Content (___/5 points)

Organization (___/5 points)

Creativity (___/5 points)

Encourage questions from class (___/5 points)

Answers to questions (___/5 points)

(if you don't know the answer, it's OK to admit that)

Visual aids (___/5 points)

● SCENARIOS

- Your 16-yr-old daughter is 6' tall. After some discussion about her health and some probing by the family physician into your family's history, you are referred to a genetic counselor. The physician suspects the possibility of Marfan's Syndrome. Your daughter is currently on the varsity basketball team, and the season has just gotten underway.
Your income: \$55,000 Insurance: HMO, 80% coverage.
- You and your wife have just lost a child to Tay-Sachs disease. You were referred to a genetic counselor before deciding to have more children.
Your income: \$75,000 Insurance: none
- You and your husband are in your early forties and have decided you would like to have another child. Your physician refers you to a genetic counselor to discuss concerns regarding Down's Syndrome.
Your income: \$150,000 Insurance: 80% coverage
- You and your partner are both African American. You have two children: the second child, a girl, is an albino; the first child, also a girl, is visually impaired. You would like another child and seek the advice of a genetic counselor.
Your income: \$90,000 Insurance: full coverage.
- You have one child, age 3, that has cystic fibrosis. You are three months pregnant with your second child; you and your husband separated a month ago. You have been referred to a genetic counselor.
Your income: \$35,000 Insurance: coverage through spouse's employer.
- You have just married. You and your spouse are healthy but your husband's brother has two children with sickle cell anemia and your sister has the same disease. You are thinking of having children and have sought the advice of a genetic counselor.
Your income: \$51,000 Insurance: none
- Your oldest child has PKU that was diagnosed at birth. You are unexpectedly pregnant with a second child and have been referred to a genetic counselor.
Your income: \$72,000 Insurance: through your employer jointly; your husband has just been laid off from his job.
- You have hemophilia; you and your spouse would like to have children. You are referred to a genetic counselor.
Income: You just lost your job due to missing so many days of work for hospital stays.
Wife's income as teacher's aide: \$18,000.
Insurance: none
- You and your wife both have achondroplasia. You have just built a house to suit your needs. You would like to have a family and have been referred to a genetic counselor.
Income: \$150,000. Insurance: HMO, 90% coverage
- Gloria, 19, is married to Robert, 21, and they wish to start a family. Both of Gloria's parents are healthy (Sonia, 39, and Todd, 40). However, Gloria's grandfather died at the age of 43 after being diagnosed with Huntington's Disease. Gloria and Robert have many questions and seek out a genetic counselor for information.
Income: \$52,000. Insurance: both, through employers

- Jim, 32, and Tammy, 28, have had two healthy children: Twila, age 3 and Terry, age 5. They have, however, recently discovered some background news about Tammy's family that concerns them. They have just found out that a brother of Tammy's, who was confined to a wheelchair by age 10, has Muscular Dystrophy. They would love to have a family of four children. Genetic counseling is available.
Income: \$80,000 Insurance: 50% coverage
- As a result of information learned in his high school biology class, Jim thinks he may have Klinefelter's Syndrome. His parents have never heard of this disorder and they seek out a genetic counselor.
Family income: \$92,000 Insurance: full major medical coverage.
- You and your wife have two children. The first is healthy. The second has spina bifida, and is paralyzed from the waist down. You desire more children and seek the advice of a genetic counselor.
Income: \$200,000 Insurance: full coverage
- Cindy, 38, is expecting her third child. She has two healthy children. Due to her age, her doctor suggests that amniocentesis be done at sixteen weeks post-conception. The karyotype reveals that the child has Turner's Syndrome. Cindy and her husband Stan are referred to a genetic counselor with this information in hand.
Income: \$75,000 Insurance: self-insured