

SCIENCE EDUCATION FOR NEW CIVIC ENGAGEMENTS AND RESPONSIBILITIES

SENCER

HUMAN GENETICS

KIM FINER

(KENT STATE UNIVERSITY STARK CAMPUS)

The SENCER Model Series 2002

Human Genetics

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What are the capacious civic questions or problems addressed in the course?

The course is organized around this key question: How will advances in Human Genetics, particularly data generated by the Human Genome Project, impact society? Six years ago Human Genetics was taught as a traditional genetics “content” course to nursing students. However, the huge volume of information generated by the Human Genome Project has changed the course dramatically. In addition to a basic understanding of genetic principles, students are now also challenged with ethical, legal, moral, and social questions that new information and technologies have raised.

What basic science is covered in this course and how is it linked to public policy questions?

Scientific Principles and Topics	Linkage to Public Policy
History of the discipline (Genetics).	Positive and negative eugenics Laws against race mixing, sterilization laws, laws restricting immigration
Transfer of genetic material (Mendel and pedigrees).	Genetic privacy, workplace or military testing, families as research subjects.
Nature of genetic defect (molecular and chromosomal level).	Genetic testing, ethics of testing, <i>in utero</i> testing, selective reduction of multiples.
Sex determination.	Olympic sex testing; Do the parts (sex) make the person?
Gene mapping, technology, laboratory manipulation of genetic material.	Genetically modified foods, cloning of sheep or people, therapeutic or experimental use of fetal tissues, stem cells.
Human Genome Project	Is it worth the cost? Private vs. government efforts; The right to patent genetic information.
Genetics testing	Ethics of testing; The right to know or not to know; Why test if there is no therapy? Insurance coverage and legislation

Cancer; cell cycle regulation.	Genetics vs. environmental influences. Prophylactic treatment (mastectomy); Insurance coverage and costs.
Histocompatibility, transplantation, blood groups.	Transplants: who and how much. Organ-farming.
Genetic disease.	Treatment, gene therapy, costs, application, risks. Government control of gene therapy. Patenting of disease genes.

What strategies does the course use to both advance science education and foster civic engagement?

Following completion of this course, it is expected students will have the tools to make informed decisions regarding the impact of genetic advances on society as well as their own personal lives. The course is designed to introduce the student to the discipline of Human Genetics by interweaving classical genetics concepts with major genetic “issues” including genetic diversity, the human genome, biotechnology, and genetic disorders. It is hoped that through mastery of course content and completion of course activities, students will gain a critical appreciation of how scientists approach and solve problems. They will also have the tools to make informed decisions regarding the impact of genetic advances on society, as well as their own personal lives. Students currently enrolled in the course include majors in Biology, Nursing, English, Women’s Studies, and the Exploratory Option.

Course activities and teaching tools have included the use of problems and case studies, poster presentations, term papers, position papers, and class discussions. In a recent semester the course centerpiece was a portfolio composed of ten journal entries, each one a synopsis and critique of an article found in the popular press concerning genetic advances. Students must include in their synopsis 1) the genetic principles which apply to the news item and 2) an opinion of the application or value of the new information or technological advance to society. The portfolio also includes a reflective essay describing the ways in which student thinking, reading, and writing have improved, and how sharply their critical thinking skills have developed, as well as an evaluation of how their content learning has improved. In addition to mastering content, a major goal of this course is to encourage students to understand that although the science of genetics appears exact and technologically advanced, there are still many areas in which opinions and judgments must be formed as to the application and use of the scientific advances. A Human Genetics course offers the perfect subject matter through which to learn and evaluate the social consequences of science. My hope is that students in the course will gain an understanding of their responsibility and role in an informed community.

Human Genetics: BSCI 30050

General Information

Instructor:	Dr. Kim R. Finer
E-mail Address	Kfiner@stark.kent.edu
Phone:	330-244-3434
Office:H	434 Main
Office Hours:	Tues., Thurs., 9:30 AM-10:30AM Mon., Wed., 2:30 PM-3:00 PM, and as arranged
Class Time:	Mon, Wed. 3:30PM-4:45PM
Course Credit:	3 hours

Course Description

This course is designed to introduce the student to the discipline of Human Genetics by interweaving classical genetic concepts with major genetic issues including genetic diversity, the human genome, biotechnology and genetic disorders. It is hoped that through mastery of course content and completion of course activities, students will gain a critical appreciation of how scientists approach and solve problems. Following completion of the course, students will have the tools to make informed decisions regarding the impact of genetic advances on society as well as their own personal lives.

Text

Required: *Human Heredity, Principles and Issues*, Fifth Edition, 2000, Michael Cummings, Brooks/Cole Thomson Learning.

Textbook web site:

<http://www.brookscole.com/biology/member/student/humheredity/index.html>

Class Requirements

Attendance is assumed. Students are responsible for lecture materials missed in their absence. Missed exams may be made up only if the instructor has been informed of the absence and found the excuse acceptable **on** or **before** the day of the exam. Make-up exams will be taken (regardless of date originally scheduled) during the last week of classes in the Testing Center (lower level, Library). It is necessary to complete **all** assignments to receive a passing grade in this course.

Course Goals and Objectives

The objective of the student will be to master the following classical genetic concepts: DNA and RNA replication, chromosomal structure, cell division, Mendelian inheritance, sex linkage, mutation, gene mapping, cloning, and gene regulation. These concepts will then be applied to specific genetic topics. To meet the above objective it will be the goal of the student to master the material such that he or she will be able to answer the review questions found at the end of each chapter in the book.

Lecture Schedule

Date	Topic	Reading
Jan. 14	Introduction, Eugenics	Hand-out
16	History, DNA Structure and Function	1,8
21	MLK Holiday	
23	Transcription and Translation	9
28	Translation Cont., Chromosomes, Structure	9, 2
30	Mitosis and Meiosis	2,
Feb. 4	Mitosis and Meiosis, /case	2,
6	Mendel, Principles and Crosses	3,
11	Mendel Continued	3,
13	Autosomal inheritance, Pedigrees	4
18	Exam I	
20	Sex Linkage, Pedigrees	4
25	Sex Linkage	4
27	Gene Mutation	11
Mar. 4	Chromosomal Aberrations	6
6	Chromosomal Aberrations	6
11	Sex determination, case	7
13	Exam II	
18-	Gene Mapping	12,13
20	Gene Mapping, Technology	12,13
25, 27	<i>Manipulation of the genetic material</i>	12, 13
April 1	Spring Break	
3	Human Genome Project	13
8	EXAM III	
10	Human Genome Project	13, Handout
15	Human Genome Project, Genetic testing, Ethics	19
17	Regulation of Expression, Cell Cycle, Cancer	14
22	Cancer	14
24	Histocompatibility, Blood Groups	15
29	Human Biochemical Genetics	10
May 1	Human Biochemical Genetics	10
8	Final Exam, 3:30 PM	

Grading

Mini Exam I	50 points
Mini Exam II	50 points
Exam III	80 points
Final Exam	100 points
Portfolio assignment	70 points
Activities (cases/short papers/problems)	70 points
Attendance and class participation	10 points
Total	430 points

Final Grade Determination

90-100%	A
80-89%	B
70-79%	C
60-69%	D
0-59%	F

Final Grades will not be curved. The grade you receive will be the grade you **earn**.

Students with Disabilities

In accordance with University Policy, if you have a documented disability, you may request accommodations to obtain equal access and to promote your learning in this class. Please contact the Office of Student Services to acquire the name of the campus representative to whom documentation should be submitted. After your eligibility for accommodations is determined, you will be given a letter which, when presented to instructors, will help us know best how to assist you.

Withdrawal Policy

Refer to the KSU Undergraduate Catalog for information .
Course withdrawal deadline: Mar. 23, 2002

Academic Misconduct Policy

The use of intellectual property of others without attributing them is considered a serious academic offense. Cheating or plagiarism will result in receiving a failing grade for the work or the course. Repeat offenses will result in dismissal from the University.

Beeper, Telephone, and Pager Policy.

As a courtesy to your fellow classmates, I request that you turn off **all electronic devices** before you enter the classroom. If an emergency arises outside of the classroom that affects you, have your contacts call the switchboard (330-499-9600) and tell them it is an emergency. Student Services will immediately send someone to the classroom for you. If by any chance, you forget to turn off your telephone or pager, please **exit the classroom with the device as soon as possible** so as not to disturb the rest of the class.

Caveat

This syllabus serves only as a guide. Dates, times, and topics may change due to unforeseen circumstances.

Tips for Being Successful in this Course.

1. Sit in the front.
2. Attend class. While an emergency may arise during the semester which may cause you to miss class, frequent absenteeism is directly reflected in test grades. Don't rely on someone else's notes. You need to be in class.
3. Work problem after problem after problem. Attempt all of the problems at the end of the chapters. This is the best way to be sure you have mastered the material.
4. Read the book chapter within a day of the class lecture. Use the book to help you understand the material and fill in any holes in your notes. Better yet, rewrite your notes using the lecture and the book to organize your thoughts. If you need additional help come see me during office hours.
5. Use the textbook website. There are some interactive activities that will not only help you learn but are actually fun!
5. Do not wait until the night before the exam to begin your preparations for the test. You simply cannot learn this material in a short period of time. Also do not wait until the day of the test to visit me during office hours. If you are having difficulty understanding the concepts see me as soon as possible.
6. Accept responsibility for your education. I will give you 100% effort in the classroom, and to be successful, you must do the same.

Eugenics Movement Analysis

(This activity will take place at the beginning of the semester and be repeated at the end)

Articles are from the Image Archive on the American Eugenics Movement
<http://www.eugenicsarchive.org>

Eugenic Laws Against Race Mixing
Eugenic Sterilization Laws
Eugenic Laws Restricting Immigration

- Summarize five key points/positions in the assigned eugenics piece.

- Identify 3-5 incorrect genetic assumptions made in the article

- What societal influences or prejudices at the time supported the position of these eugenics practitioners?

**Case Study:
How much should we know?**

In 1971, an ultrasound scan of Mrs. W revealed large cysts in her kidneys and liver consistent with autosomal dominant polycystic kidney disease. Four years later her kidneys failed and she became a dialysis patient. Mrs. W's father and two brothers had died from kidney failure.

After Mrs. W began dialysis treatment, her four sons also underwent ultrasound scanning. The scans revealed they each had polycystic kidney disease although at the time they were healthy.

Three years later Mrs. W's 32 year old son developed abdominal pain and the first symptoms of kidney disease. Not wanting to undergo dialysis, he committed suicide leaving behind his wife and 6 yo son.

When Mrs. W's other 30 yo son learned of his condition, he sold his half completed new house, not wanting any financial burden. He is now under medical treatment, but not yet on dialysis.

A 25 yo son and his fiancée dissolved their engagement. 15 years later he died suddenly of a ruptured intracranial aneurysm, a complication that occurs in 10% of patients with this form of polycystic kidney disease. He had known about the aneurysm, but had refused surgical treatment.

The fourth brother, a 21 yo, left college and took a well paying job vowing to stay single and enjoy life while it lasted. Although he has developed hypertension, he is now 42 and still enjoys life.

Draw a pedigree of this family as it relates to polycystic kidney disease.

Question for discussion:

Should Mrs. W have kept the true nature of her disease to herself and not told her sons?

"A Right to Her Genes"

by

Susannah Gal

Department of Biological Sciences
State University of New York at Binghamton
and

Jessie W. Klein

Science Department
Middlesex Community College

"But, doctor, what should I do?"

Michelle was sitting in her OB-GYN's office, having just confronted him with the dilemma she was facing.

"My mother died of breast cancer when I was little and now I find out that her mother, my grandmother, has bone cancer and my grandmother's brother and my grandfather both have lung cancer. My mother was 40 when she died and I'm almost that now myself. Should I have my breasts removed to prevent me from getting breast cancer?"

The doctor tried to calm her down and clarify the family tree. As far as Michelle knew, her mother was the oldest of four girls and two boys in the family and the only one to have cancer. In fact, her grandfather also had a brother and two sisters, and none of them showed any signs of cancer.

"It's not strong enough evidence to suggest you should remove your breasts," the doctor said. "Actually, breast cancer susceptibility is not linked to lung cancer. Lung cancer, especially, is usually linked to environmental factors, like smoking or exposure to asbestos. Does your father smoke?" the doctor asked.

"Yes," Michelle replied. "But I don't want to get cancer and have my two little boys watch me die the way I did with my mother."

The doctor suggested Michelle gather more information about her family tree and then come back and he would contact a genetic counselor about the possibility of genetic testing.

A month later, Michelle went to see her doctor again after having talked with her grandmother extensively about the family. She had uncovered two very interesting family facts. First, the man she thought was her mother's father was not her biological grandfather, but his brother. Her real grandfather had no history of cancer. Second, Michelle found out her mother had an estranged older sister, Anne, who had recently

been diagnosed with ovarian cancer. The doctor now felt the family connection was strong enough to advise Michelle that there was a possibility of a cancer gene linkage in the family.

The doctor contacted Laura, a genetic counselor in the area, and found out that the most likely candidates were the breast cancer genes BRCA1 and BRCA2. Mutations in one of these two genes are found in 90% of tumors from breast and ovarian cancer. Women with hereditary mutations in one of these genes are 30 times more likely to develop breast cancer by age 50, while men have a three times higher risk of developing prostate or colon cancer if they inherit this mutation. The genetic counselor clarified that a variety of studies have suggested that mutations in BRCA1 are more often linked with breast and ovarian cancer, while mutations in BRCA2 are most often linked only with breast cancer. The fact that Michelle's grandmother, the mother of both affected women, had bone cancer and not breast or ovarian cancer might be the result of a metastasis from the original site of the tumor in the breast. Thus, Laura felt this family was a candidate for a BRCA1 mutation.

With Michelle's consent, the doctor arranged to have the breast biopsy sample from her mother sent to a DNA testing facility, Myriad Genetics in Salt Lake City, Utah. A month later they found out that the sample was a poor one, making it difficult to get any information. The testing facility thought that the sample might not have been prepared or stored properly and thus the DNA in it was degraded beyond usefulness. So, there was no data to indicate that Michelle's mother's cancer was due to a mutation in BRCA1. No other samples existed of Michelle's mother's tumor or other tissue from which they could do another analysis.

Michelle's doctor contacted Laura, the genetic counselor, to discuss the case. Laura felt that in order to make any assessment, a DNA sample from an individual with cancer was needed. This sample could then be used as a comparison for the sample, namely Michelle's, with unknown cancer risk. If the DNA from an individual with cancer showed a mutation in the BRCA1 gene, Laura explained, then they would look for the same mutation in Michelle's DNA. Ideally, the mother's DNA would have provided them with this information, but without it, a DNA sample from another close relative with cancer was essential. Otherwise, they wouldn't be able to be certain that a DNA change in Michelle's BRCA1 gene was necessarily linked to cancer. DNA from the estranged, cancer-carrying aunt would be very useful and could be obtained from a sample of her blood.

Laura had a lengthy conversation with Michelle in which they went over Michelle's complicated family situation. Laura then wrote a letter to Anne, the aunt who lived in a remote town two hours away, in which she explained the process and asked for her informed consent to give a blood sample to get DNA. After two weeks had gone by with no reply, Laura tried contacting Michelle's aunt a second time. Again, she received no reply. After the third time, Laura finally got the aunt on the phone. Anne reluctantly agreed to meet with Laura for coffee--not in her home, but in a restaurant in her small town--and as long as Laura paid for it.

Laura met Anne at the restaurant at the appointed hour. Anne was accompanied by a younger woman who turned out to be her daughter. The daughter read the menu for her mother and Laura suspected that the older woman was illiterate.

After introducing herself, Laura said: "I'm here representing your niece, who would like to arrange to get some information from you to help her decide if she has a cancer gene."

Anne looked confused. "What's a gene?" she asked.

Laura explained: "Our bodies are made up of small parts called cells that contain a substance called DNA. DNA is the blueprint for our cells and controls everything about us. DNA is the same in all your cells. It is passed from one generation to the next, provided from both mother and father at fertilization. So, the DNA found in Michelle should be similar to your DNA, since the two of you are related. Changes in your family's DNA may have made several of its members more susceptible to cancer and would explain why several of you have gotten the disease. This would include your sister, Michelle's mother, who died from breast cancer. Michelle would like to know if there is such a change in your DNA and also in her own DNA. If there is such a mutation, Michelle might then decide to remove either her breasts or her ovaries to prevent her from getting cancer. Michelle would like you to give a blood sample at your doctor's so that DNA can be isolated and the gene sequence determined."

Anne was silent for a moment and then asked Laura, "Who would pay for this? I haven't got any money."

"I don't know that yet. Who covers your medical bills?" asked Laura.

"My medical bills are covered by Medicaid," Anne said.

Laura knew that the BRCA1 test costs in the neighborhood of \$2500 for the complete information about this gene and that Medicaid would not pay for it.

Anne's daughter interjected belligerently, "Why are you doing this, Mom, when they ignored you for so many years and haven't given us any money since you left?"

Create a family tree using standard symbols for the people mentioned in the case and include some designation to indicate those affected with cancer.

- What part of the blood sample would be used for the DNA analysis and why?
- What techniques can be used to determine whether Anne's DNA has a mutation in the BRCA1 gene?
- Why might the degradation of the DNA from Michelle's mother make the information obtained unusable? Are there other approaches one could use to get around that problem?
- What do each of the people in this scenario want to get out of this situation?
- What problems do you foresee with the different agendas--the different needs--of the people involved in this case?
- What are ways that one could solve these problems?

**Human Genetics
Spring 2001
Portfolio Assignment**

Requirements:

Month	Due	Point value
January	1 summary	10
February	2 summaries	10 each (20)
March	2 summaries	10 each (20)
April	1 summary	10
May	Reflective essay/Completed portfolio	10 points

Each summary includes a synopsis and critique of a news article (a copy of which is attached to the summary) describing a discovery, advance, therapy, or ethical issue in genetics. The summary should also include a brief discussion of any genetic concepts that you may have learned in class, or found in the text which apply to your selected article.

Summaries should be approximately 2 pages in length.

Completed portfolio includes the following components:

Table of contents

Reflective essay

6 summaries (modified as suggested)

The two page reflective essay serves as an introduction to the summaries in your portfolio. The objective of this essay is to reflect upon how your thinking processes, opinions, reading and writing skills have been impacted by this activity.

Questions you may want to answer in the reflective essay:

How have your thinking/writing/reading skills improved as a result of your work on this project? How might this practice at analyzing/summarizing help you in future course work? How did the subject matter in the course relate to your overall learning and ability to read and analyze your selected articles? How will the advances which you read about impact you, your community, or the human race? Did any of your readings change the way you think about scientific research or discovery?

The complete portfolio should be bound in a three ring binder or other bound format. The completed portfolio is due May 2 at 5:00 PM

Any source of print or electronic information may serve as a portfolio article.
 Suggested resources: newspapers, magazines, Info trac
 Articles should be relatively recent (written within the last three months)

Grading Rubric for Portfolio Summaries

Point Value	Rank	Content	Opinion	Connection	Mechanics
10	Exemplary	Summarizes >90% of content	Expresses logical (supported) opinion regarding article	Incorporates more than one concept from course	Minimal (3) major grammatical or punctuation errors
7	Good	Summarizes 70-80% of content	Expresses opinion (not supported) regarding topic	Incorporates one concept from course	>5 G & P errors
4	Adequate	Summarizes less than 70% of content	No opinion, or unsupported opinion	Does not incorporate course content	>8 G & P errors