

**COURSE DESCRIPTION** - Human genetics is a biology major course. It focuses on the major areas of human genetics - Mendelian genetics, molecular genetics (biochemical genetics), cytogenetics, complex genetics (multifactorial genetics) and population genetics. In addition to exams, students will be required to prepare short papers and to present a presentation (this will be done in pairs) on a topic related to human genetics.

**INSTRUCTOR:** Dr. Karen Kurvink  
Associate Professor of Biology  
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**COURSE INFORMATION:**

Lectures – MWF 1  
Collier Hall of Science 202  
Laboratory – Friday (12:45 – 3:45)  
Collier Hall of Science -303

**TEXT: HUMAN HEREDITY – Principles and Issues**, 8<sup>TH</sup> edition by  
Michael Cummings 2009

**COURSE GOALS:**

1. To interrelate historical and current human genetic topics in a way which will make them more meaningful.
2. To expose students to the research and diagnostic tools of human genetics.
3. To assist student in their development of appreciation, understanding, and/or empathy related to human difference.

**COMMENT:** Today's students will be the consumers of many of the new biotechnologies currently being developed. We will all need to make decisions about the application and regulation of these technologies. Awareness of human genetics should prepare students to better understand the social impact of science on one's personal life and on society in a more general sense.

**BASIC INFORMATION:**

1. Students are expected to attend both lecture and laboratory (attendance will be taken). If you must miss a class be sure to contact the professor immediately (preferably before the event occurs).
2. Students earn 20 points for attending and participating in each laboratory.

3. Students will play an active evaluation role in terms of the class presentations.
4. Misses exams require a written excuse. If a make-up exam is allowed, it will be an individual exam (usually a combined written/oral exam).

GRADING: The final letter grade will be based on earned points in the course divided by possible points available. The letter grade will be determined by this percentage:

|        |   |  |
|--------|---|--|
| 90-100 | A |  |
| 80-89  | B | + and – will be added at the discretion of the professor |
| 70-79  | C |  |
| 69-69  | D |  |
| below  | F |  |

OPTIONAL OPPORTUNITIES for earning additional points will be announced by the professor. Note: the same criterion will exist for these efforts (ie earned over possible points). These points will be added into the general course grade. Note: the maximum number of points which can be earned is 60 pts.

CONTRACTING: In recognition of student variability in interest and intellectual attributes, an opportunity for contracting is also available. Whether a student selects this option should also be based on available time (this is an add-on to the basic course design). Note: the maximum number of points which can be earned is 150 pts.

#### **ANTICIPATED POINT DISTRIBUTION**

|                   |                    |
|-------------------|--------------------|
| Unit exams        | 400 pts            |
| Short papers      | 300 pts            |
| Lab               | 260 pts            |
| Presentation      | 100 pts            |
| Optional extras   | maximum of 60 pts  |
| Optional contract | maximum of 150 pts |

## Tentative Lecture Schedule

|                              |  |                    |
|------------------------------|--|--------------------|
| Jan 19 (M)                   | Perspective on human genetics<br>Brief history<br>Tracing lineage – Y chromosome<br>Thomas Jefferson lineage<br>Cohen Y haplotype (CMH) – African Lemba tribe<br>Moravian archives | Chapter 1: 1-17    |
| Jan 21 (W)                   | Somatic cell division and the mitotic cell cycle<br>Germ cell division - meiosis   | Chapter 2: 18-43   |
| Jan 23 (F)                   | Mendel's laws  | Chapter 3: 44-69   |
| Jan 26 (M)                   | Single gene Mendelian patterns<br>autosomal dominant<br>autosomal recessive  | Chapter 4: 70-81   |
| <i>*Research paper 1 due</i> |  |                    |
| Jan 28 (W)                   | Single gene Mendelian patterns<br>X-linked dominant<br>X-linked recessive<br>Y-linked  | Chapter 4: 82-99   |
| Jan 30 (F)                   | Complex genetic conditions<br>Traditional methods of study   | Chapter 5: 100-114 |
| Feb 2 (M)                    | Complex genetic conditions<br>Association mapping  |                    |
| <i>*Research paper 2 due</i> |  |                    |
| Feb 4 (W)                    | Cytogenetics   | Chapter 6: 128-157 |
| Feb 6 (F)                    | Early development<br>Sex determination<br>Teratogens   | Chapter 7: 158-173 |
| <b>Feb 9 (M)</b>             | <b>UNIT EXAM 1</b> (Chapter 1-5) 100 pts   |                    |
| Feb 11 (W)                   | Role of hormones in sexual development<br>X-inactivation<br>Chimera  | Chapter 7: 173-187 |

|            |  |                      |
|------------|--|----------------------|
| Feb 13 (F) | Nucleic acids: DNA and RNA<br>Chromosome structure<br>DNA replication  | Chapter 8:188-209    |
| Feb 16 (M) | Protein synthesis  | Chapter 9: 210-230   |
|            | <i>*Research paper 3 due</i>   |                      |
| Feb 18 (W) | Genetic errors in metabolic pathways<br>PKU<br>Lactose intolerance   | Chapter 10:230-241   |
| Feb 20 (F) | Hemoglobinopathies   | Chapter 10:242-257   |
| Feb 23 (M) | Mutation at the molecular level  | Chapter 11:258-272   |
| Feb 25 (W) | Mutational repair  | Chapter 11:273-283   |
| Feb 27 (F) | <b>UNIT EXAM 2</b> (Chapter 6-10)  |                      |
| Mar 9 (M)  | Genes and cancer<br>Oncogenes<br>Tumor suppressor genes  | Chapter 12:284-292   |
| Mar 11 (W) | Cancer models<br>Specific chromosomal modifications  | Chapter 12:293-305   |
| Mar 13 (F) | Recombinant DNA technologies<br>Restriction enzymes<br>Vectors<br>Recombinant cloning<br>Recombinant libraries | Chapter 13:306-318   |
| Mar 16 (M) | Polymerase chain reaction<br>Southern blotting<br>DNA sequencing   | Chapter 13: 319-328  |
| Mar 16 (M) | Genetically modified organisms (GMOs)<br>Transgenic bacteria<br>Transgenic plants<br>Transgenic animals        | Chapter 14:330-340   |
| Mar 18 (W) | Genetic screening<br>Microarrays   | Chapter 14: 340-346  |
| Mar 20(F)  | DNA profiling  | Chapter 14: 346- 367 |

RFLPs  
Full DNA profiles  
DNA sequencing

|            |   |                      |
|------------|---|----------------------|
| Mar 23 (M) | Population genetics<br><i>*Research paper 4 due</i> | Chapter 19:546-467   |
| Mar 25 (W) | Microevolution                                      | Chapter 19:468-478   |
| Mar 27 (F) | Genetics of race/eugenics                           |                      |
| Mar 31 (M) | <b>UNIT EXAM 3</b> (Chapter 11-14; 19)              |                      |
| Apr 1 (W)  | Genomics/proteomics                                 | Chapter 15:358-375   |
| Apr 3 (F)  | Personal genome project                             | Chapter 15:375-376   |
| Apr 6 (M)  | Reproductive technologies                           | Chapter 16: 382-391  |
| Apr 8 (W)  | Gene therapy<br>Gene counseling                     | Chapter 16: 391- 401 |
| Apr 10 (F) | No class – Good Friday                              |                      |
| Apr 13 (M) | No class - Easter Monday                            |                      |
| Apr 15 (W) | Immunogenetics                                      | Chapter 17: 402-414  |
|            | <i>*Research paper 5 due</i>                        |                      |
| Apr 17 (F) | Immunogenetics                                      | Chapter 17: 415-429  |
| Apr 20 (M) | Behavior genetics                                   | Chapter 18: 430-445  |
| Apr 22 (W) | Behavior genetics                                   | Chapter 18: 446-455  |
| Apr 24 (F) | Guest lecture/lab                                   |                      |
| Apr 27 (M) | Guest lecture (or video)                            |                      |
| Apr 29 (W) | Presentations (done in pairs)                       |                      |
|            | <i>*Research paper 6 due</i>                        |                      |
| May 1 (F)  | Presentations (done in pairs)                       |                      |

**UNIT 4 EXAM** – Monday May 4<sup>th</sup> at 8:30 PM (during final exam period 1) (Chapters 15, 16, 17, 18)

### **TENTATIVE LABORATORY SCHEDULE**

- Jan 23            Karyotype preparation  
                         Introduction to routine chromosome cytogenetic analysis  
                         Mendelian genetic web sites - OMIM, genetests, and NORD  
                         Huntington's disease exercise  
                         Cystic fibrosis exercise  
                         Stem cell research regulation and ethical dilemmas
- Jan 30            Mitosis/meiosis  
                         A. Mitosis – prepare bone marrow sample of mouse or rat  
                         B. Meiosis  
                             - prepare testicular slides of mouse or rat  
                             - mammalian slides of spermatogenesis and oogenesis  
                         C. Sperm variation slides
- Set up and maintain somatic culture from rodent embryos
- Feb 6             Introduction to pedigree analysis for Mendelian genetic conditions  
                         Genetic problems  
                         Chromosome syndromes with primarily emphasis on Down syndrome  
                         A. Modern molecular cytogenetic procedures (FISH, chromosome painting)  
                         A. Prenatal diagnostic testing  
                             - amniocentesis  
                             - chorionic villi methods
- Feb 13            Prepare sex chromatin slides  
                         DNA isolation  
                         DNA molecular structure and associated terminology
- Feb 20            Protein synthesis  
                         DNA sequencing exercise – Sanger DNA sequencing  
                         Genetic engineering exercise
- Feb 27            Agarose electrophoresis  
                         A. Sickle cell screening  
                         B. Southern blotting of viral DNA
- Mar 13            Isolation of plasmid DNA  
                         Dermatoglyphics

- Mar 20      DNA fingerprinting  
Plasmid DNA electrophoresis and identification (from previous lab)
- March 27    Population genetics exercise  
Eugenics - Emphasis on US and Germany examples  
Evolution discussions
- April 3      Dog genetics and its association with human genetics  
Genetic counseling
- Apr 10      Good Friday – no lab
- Apr 17      Human chromosome cultures  
    A. Set up cultures (on Tuesday – April 14)  
    B. Harvest cultures (on Friday- April 17)  
    C. Preparation of slides
- Apr 24      Photographs of chromosomes
- May 1        Presentations on selected topics (done in pairs)

### **SHORT PAPER ASSIGNMENTS**

**(50 pts each)**

**Prepare a short (maximum 3 typed pages) paper on each of the following topics. Be sure to reference any information which is not considered to be general information in the body of the paper and to have a bibliography of at least three citations. The intent of these paper assignments is to provide opportunity for you to research and summarize a specific case (condition) related to the topics covered in lecture. Papers will be graded based on (a) content (b) reference and bibliography inclusion and (c) organizational design and accuracy.**

**Assignment 1.**      Select one of the following topics:

**Due date -** January 26th

- A. Your support or rejection of the possibility of Thomas Jefferson being the father of Sally Hemmings children based on Y chromosome analysis.
- B. Your support or rejection of the genetic evidence for a Jewish ancestry of the Lemba tribe based on Y chromosome analysis.

- C. What type of genetic information is collected by the Mormon church? Is any of this information available to scientists or the general public?

**Assignment 2.** Select one of the following topics related to stem cells:

**Due date:** Friday, Feb 2nd

- A. The scientific and ethical aspects of using somatic or embryonic stem cells for treatment of a selected human condition (disease or injury).
- B. Pros and cons of “banking” embryonic cord blood.
- C. Pros and cons of using somatic nuclear transfer and donor mammalian eggs in human IVF.

**Assignment 3.** Select one of the following topics related to genetic **chimerism**

**Due date:** Monday Feb 16

- A. association with ambiguous sexual development
- B. etiology and potential genetic identity “spin offs” of chimerism
- C. female heterozygosity for a selected X-linked recessive disease
- D. interspecies chimeras

**Assignment 4.** Select one of the following topics related to GM organisms.

**Due date:** March 23rd

- A. Prepare a personal position statement on consumption of GM food.
- B. What types of environmental pros and cons exist related to GMOs?
- C. Should global labeling of GM food be required?

**Assignment 5.** Select one of the following topics related to reproductive or genetic biotechnologies.

**Due date:** April 15th

- A. In what ways could the developing genetic and reproductive technologies be leading us into another period of eugenics?
- B. How do you think genomics/proteomics may influence your life in the future?
- C. What is associative gene mapping? How will it be important in the diagnosis and potential treatment of complex genetic diseases?

**Assignment 6.** Select a genetic condition related to immunogenetics, cancer genetics, or behavior genetics.

**Due date:: April 22<sup>nd</sup>**

For a **specific condition** related to any of these applied genetic topics indicate the current available technologies for diagnosis and/or treatment. Indicate, if possible, the importance of animal studies in the research and/or application of these technologies to humans.

**PRESENTATION GUIDELINES  
(100 pts/student)**

Oral presentations will be given during the last week of the course. The presentations will be researched and presented by pairs of students. The presentation will be 20 minutes in length and should include the following:

- (a) outline of topic (1 page which will be copied and given to the other students and the professor)
- (b) powerpoint presentation (approximately 15 minutes in length) – the last slide should contain references; a copy of the slides should be given to the professor before the presentation.
- © question/answer period (approximately 5 minutes)

The goal of these presentations is demonstrate the basic principles learned in the course in a more applied or specific situational case..

**Grading:** Both students will receive the same grade unless I have been advised otherwise.

The grade will be based on (a) content, (b) presentation effectiveness, and (c) class involvement.

Order:

Carolina taste papers

IND-16 Identifying Viral DNA by Rapid Southern Blotting - \$112.36

Package 3/4 \$63.81

IND-9 Producing a strain of E. coli that Glows in the Dark \$80.32

3-18 Plasmid DNA isolation Kit \$54.71

### **POSSIBLE OPTIONAL VIDEOS**

Harvest of Fear

### **POSSIBLE CONTRACTING IDEAS**

1. Books

Read and either prepare a short paper or discuss with professor

Possible considerations:

THE GENETIC STRAND by Edward Ball

THE UNFIT BY Elof Carlson

2. Survey

Type of articles related to human genetics in various type of journals,

newspapers.

Type of genetic information presented to the public via television and/or movies.

3. Counseling services (genetic and reproductive) available locally
4. Perinatal services available locally or in this region of the country.
5. Specific genetic disease review

Example: Tourette's syndrome – see video on reserve in library and research current information

Lecture 1

## **Y CHROMOSOME HAPLOTYPE AND HAPLOGROUP**

**Be able to define haplotype and haplogroup:**

**Haplotype – blocks of nucleotides which travel together  
(marker groups which have some degree of SNPs)**

**Haplogroup – wider grouping of haplotypes**

**Markers:**

Short tandem repeats  
SNPs (snips)

Unique markers for a population:

Duffy locus: FY\*O - black sub-Saharan Africa (Duffy-null variation)  
On chromosome 1  
Related to resistance to *Plasmodium vivex*

SR Y gene on short arms of Y chromosome

Kits: STR kits

- Power Plex Y
- Y -film

1. Jefferson's Y chromosome

**Nature** Nov 5 1998 article by Dr. Eugene Foster

Thomas Jefferson and Sally Hemings

Children: Thomas, Harriet, Beverly, daughter, Madison, Eston  
Y chromosome : T (K2) haplogroup  
25 markers (\$169 + 2 handling)  
Mark Jobling – Y chromosome spokesperson

2. Cohen Modal haplotype

1997 Michael Hammer – University of Arizona  
rabbinical haplotype

3. Morman genealogy

Book: **THE GENETIC STRAND** by Edward Ball 2007

Historical walk using hair DNA

1st ancient DNA sample – China in 1982 (questionable quality)

Two important molecular genetic technologies

1. PCR – Kary Mullis 1983 (Nobel Prize)

Amplify a DNA sequence

Dancing Naked in the Mind Field

Kary Mullis and Alec Jeffreys – National Inventors Hall of Fame

2. Sanger – DNA sequencing 1970s

Determine nucleotide sequence of short DNA fragments

dideoxy sequencing – manual

1982 – Leroy Hood – fluorescent dyes

1986 – 1st automated sequencer

1987 – thin tubes + laser scan + computer analysis (print outs)

## Places for DNA Analysis

1. Trace Genetics - DNA markers associated with Indian Ancestry  
database – several thousand DNA sequences of Native Americans  
blood quantum – documents and genealogies

2. Vita-Med – Canadian paternity testing

10 basic markers

3. European DNA 2.0

Predict Europea heritage

SEE – Southeastern Europe (Armenian, Jewish, Italia, Greek)

IB - Iberian (Spanish, Portuguese)

BAS – Basque (Spanish, French Pyrenee bordr)

CE – Continental European CC (German, Irish, English, Netherlands,  
French, Swiss and Italian)

NEE – Northeast European (Polish, Baltic, Swedish, Norwegian,  
Finnish, Russian)

Uses 1,349 European Ancestry Information Markers – autosomal markers (not  
Mt DNA or Y chromosome markers)

4. European DNA 1.0 – 1st pan genome test

5. Ancestry by DNA TM 2.5 –

percentage score of European 50% score

40% East Asian

15% Sub Saharan African

15% Native American

6. DNA Print Genomics, Inc. (Mark Shriver – Penn State University)

An applied genomic science company – development and marketing of  
Innovative genetic testing products and services

Applications: Forensic science

Genological research

Pharmeceutical development

7. International Hap/Map Project

Canada, China, Japan, Nigeria, UK, US

Multi-country effort to identify genetic similarities and differences in humans  
Disease, medications, environmental

Ethical aspects - challenge of historical cultural and social ideas

Race definition

Anonymous

Consent

Data release policy

Sample storage at Coriell Institute

Blast program – Gene walker program

SNPs

Megablast (database ties to total genome)

8. Mitotyping technologies – forensic DNA analysis – especially of hair samples and ancient DNA

Identifiler – STR analysis

814-861-0676

heteroplasmy – exists to some degree in all tissues

use 150 nucleotide sequence of 12S rRNA (identify species)

Search on <http://www.ncbi.nlm.nih.gov/BLAST>

Class 2

A brief review of HG history - On being normal or abnormal

Reference: **THE UNFIT – A History of a Bad Idea** by Elof Axel Carlson 2001

Web site:

## BOOKS

1. **JACOB'S LEGACY: Genetic view of Jewish History** by David Goldstein, 2008 ISBN 978-0-300-12583-2 \$26  
  
Discusses the Cohen Modal Haplotype (CMH) and its distribution to various groups including the Lemba over time.
2. **BLOOD MATTERS: From Inherited Illness to Designer Babies, How the World and I Found Ourselves in the Future of the Gene** by Masha Green 2008 ISBN 978-0151013623 \$25  
  
Covers Green's life as a BRCA-1 mutation carrier at risk for early-onset breast cancer.
3. **Spelling Love with an X: A Mother, a Son, and the Gene That Binds Them** by Clare Dunsford 2007 ISBN 978-0807072790 \$24  
  
Dunsford's son was born with fragile X syndrome. The book demonstrates how we can navigate life's events with grace.
4. **DAVENPORT'S DREAM: 21<sup>st</sup> Century Reflections on Heredity and Eugenics** Edited by Jan A. Witkowski and John R Inglis ISBN 978-08796 9756-3 \$55 Cold Spring Harbor Laboratory Press  
  
This is actually two books in one. It is a collection of essays about Davenport and his work and a reprint of his 1911 textbook HEREDITY IN RELATION TO EUGENIC...the first book on human genetics published in the US.
5. **IN PURSUIT OF THE GENE: From Darwin to DNA** by James Schwartz 2008 ISBN 978-0-674-02670 Harvard University Press \$30

This is an engaging account of the major discoveries and personalities in the History of inheritance up until the middle of the twentieth century. It includes information on HJ Muller who made major genetic and societal contributions.

6. **GENETIC WITNESS: Science, Law, and Controversy in the Making of DNA Profiling** by Jay D Aronson, 2007, ISBN 978-0-8135-4188-4, Rutgers University Press, \$24

DNA profiling is described and followed in legal cases involving paternity And forensic cases.

7. **THE MURDER OF NIKOLAI VAVILOV: The Story of Stalin's Persecution of One of the Great Scientists of the Twentieth Century** By Peter Pringle 2008 ISBN 978-0743264983, \$26

Good historical biography of a famous Russian geneticist who **fell out of favor during the reign of Josef Stalin.**

8. **ELIZBETH BLACKBURN AND THE STORY OF TELOMERES** by Catherine Brady 2007, ISBN 978-0262026222, \$30.

Story of Blackburn and her contributions to telomere biology. It is a good story about women in science and some of the scientific politics of that period of time.

9. **THE GENIUS FACTORY: The Curious History of the Nobel Prize Sperm Bank,** by David Plotz 2005 \$14.95

10. **TRACE YOUR ROOTS WITH DNA: Using Genetic Tests to Explore Your Family History** by Megan Smolenyak Smolenyak and Ann Turner 2004 \$16.95

Good book!

11. **THE GOD GENE : How Faith is Hardwired into our Genes** by Dean Hamer 2004 \$14.95

12. **THE AGILE GENE: How Nature Turns on Nurture** by Matt Ridley 2003

13. **ENOUGH- Slaying Huma in an Engineered Age** by Bill McKibben 2003 \$14.00

14. **Rosalind Franklin: The Dark Lady of DNA** by Brenda Maddox by Brenda Maddox 2002 ISBN – 978-0060184070 \$10

Or

**Rosalind Franklin and DNA** by Anne Sayre ISBN 987-0393320442,  
\$10

Bibliographies of her life and contributions to the research on DNA  
structure.

15. **ABRAHAM LINCOLN'S DNA AND OTHER ADVENTURES IN GENETICS**  
by Philip R. Reilly 2000 Cold Spring Harbor Laboratories \$16

Very good

16. **MUTANTS: On Genetic Variation and The Human Body** by Armand  
Marie Lenoir 2005 \$11

