HUMAN GENETICS
Biology 3700
Syllabus Spring 2022
Professor ZMG Sarwar Jahangir
Ph.D. in Cellular Molecular and Developmental Biology
Department of Biological Sciences
From NCBI

Human Chromosomes
22 autosomes + 2 sex chromosomes
DNA sequencing incomplete

> 1000 kb | 250 - 1000 kb | < 250 kb
Draft sequencing | Heterochromatin

1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y
Access-Ability Services (AAS) serves as a liaison and resource to the KCC community regarding disability issues, promotes equal access to all KCC programs and activities, and makes every reasonable effort to provide appropriate accommodations and assistance to students with disabilities. Please contact this office if you require such accommodations and assistance. Your instructor will be glad to make the accommodations you need, but you must have documentation from the Access-Ability office for any accommodations.

Access-Ability Services is always looking to hire student aides/federal work study students to help provide certain services for our students with disabilities, if you are interested please stop by D205 to find out more.

Disability Related Services D205

Office Hours:

- Monday: 9am-5:00pm
- Tuesday: 9am-8:00pm
- Wednesday: 9am-8:00pm
- Thursday: 9am-5:00pm
- Friday: 9am-5:00pm

By email: AAS@kbcc.cuny.edu  By phone: 718-368-5175
STUDENT RESPONSIBILITIES & ACADEMIC INTEGRITY

Absence Policy
Attendance is required to do well in the course. You are expected to attend all lectures, exams, and presentations. **Excessive absences will result in an F grade** regardless of the legitimacy of the absence. As defined by the University Policy for Attendance a student is considered excessively absent if they have missed more than 15% of the total instructional hours for a class. Therefore, if you miss more than 12 hours total, in any combination of lecture exams and presentations, an F grade is automatically assigned.

A student absent for <15% in total must take the initiative for completing the incomplete course work and is responsible for all covered material and assigned work. The student must discuss absences with the professor prior to an anticipated absence or immediately following a missed activity session for advice.

Cell Phones and Beepers
The use or ringing of cell phones and beepers in the classroom during class sessions is a disruption of class and a violation of the Henderson Rules. Violation of this policy may result in a disciplinary referral.

Written Assignments
You will prepare three case study reports as in the schedule. You will be completing case studies in person. In addition, two students jointly will be making a 10 minutes presentation during the last third of the semester, as scheduled. That will carry 10 points for the final grade. The presentation will be based on human genetics on current issues and future directions.

Plagiarism (Academic Integrity)
You may find the CUNY and KCC’s Academic Integrity Policies are in:
- Kingsborough Community College Catalogue
- Kingsborough Community College Student Handbook
- [www.kingsborough.edu/Academic_Integrity_Policy.pdf](http://www.kingsborough.edu/Academic_Integrity_Policy.pdf)

“Plagiarism as a violation of academic integrity is the intentional use of another’s intellectual creation(s) without attribution. Determination and penalty—ranging from grade reduction to course failure—is at the sole discretion of the faculty member.”

In addition, your instructor may inform you of his or her policy regarding academic integrity at the beginning of the semester.

Assessment
You will be given a 10-20 minutes assessment test at the last part of the semester in order to test the effectiveness of teaching and learning in this course. Your instructor will decide of the content of this test and may assign a credit value, if appropriate.
COURSE DESCRIPTION

It is a three-credit course meeting three hours per week.

This non-majors Biology offering encourages students to become more “science literate” by learning and relating how current topics are constantly molding and influencing our changing world, specifically in the field of genetics. We will read, examine and critique current newspaper articles as well as use the Internet for our studies. Lectures will be augmented by selected readings from the newspapers and/or primary literature, as applicable to the topic. Class discussions and case studies will extend our lecture topics of human heredity including gene therapy, somatic nuclear transfer and stem cells, thereby allowing an extensive and comprehensive treatment of them.

The end of term ethic debate requires students to utilize course material (textbook, class discussions, and literature sources) to formulate and present their view/opinion on a topic the class chooses. Your support or disagreement will be written, presented and (re)evaluated in the format of a class debate. The course grade calculation includes a portion for submission of Internet Assignments (samples attached) which will require you to either locate or access web sites utilized by students, researchers and teachers to procure specific genetic information then to answer specified questions.

The Big Picture … MAIN COURSE OBJECTIVES

• To enrich our understanding of human heredity through exploration of the many aspects involved [a survey through the molecular, cellular and organismal levels].
• To understand how normal and abnormal cellular processes affect humans at all these levels.
• To learn what current ideas, issues and trends involve human inheritance.
• To become aware of, and to be able to discuss ethical, legal and social issues in human genetics and the implications of these developments.

TEXTBOOK

Reading Assignments: OER (Open Education Resources) online – Blackboard.

• Biology OER- https://openlab.citytech.cuny.edu/bio-oer/gene-expression/
• Help me understand genetics: https://ghr.nlm.nih.gov/primer
• Public domain – provided as PowerPoints.

ADDITIONAL MATERIALS

COURSE GRADE CALCULATION

Written Examinations (20pts x 3) = 60%
Internet Assignments and Case Reviews (4pts x 2.5) = 10%
End-of Term Ethics Debate Presentation = 10%
Final Examination = 20%
Total = 100%

Notes:

- There are no make-up examinations. A missed examination will be assigned a grade of zero. In accordance with KCC’s Attendance Policy, excessive absences will result in course grade reduction.
- Two lateness’s are equivalent to one absence.
- In accordance with the college’s policies on academic integrity, any student identified participating in cheating; plagiarism, etc. will be subjected to disciplinary actions.
- An extra credit assignment worth 5 points may be available on request. Please see the instructor during the week of 9-10. The assignment will not be accepted after its due date.
- Utilization of mobile any mobile or electronic devices is prohibited during lecture and examinations.
### Assigned Reading - OER (Open Education Resources) online;
Instructor: Professor ZMG Sarwar Jahangir, Kingsborough Community College, Biological Science.

<table>
<thead>
<tr>
<th>Date</th>
<th>Major Lecture Topics</th>
<th>Assigned Reading</th>
<th>Helpful reading</th>
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</table>
| Mar 15 & 17   | **Case Study 1.** Assignment given on Mar 15;  
| Mar 22 & 24   | **Case study 1 due on Mar 22.**  
| Mar 29 & 31   | **1st Exam on March 29 Includes topics covered before.**  
| Apr 05 & 07   | **Case Study 2.** Assignment given on Apr 05.  
| Apr 12 & 14   | **Case study 2 due on Apr 12.**  
| Apr 28 & May 03 | **2nd Exam on Apr 28. On topics covered after the 1st Exam.**  
**Case Study 3.** Assignment given on May 03;  
Topic 8: Genetics of Behavior;  
| May 12 & 17   | **Case study 3. Due on May 12.**  
Topic 11: Gene Expression and Epigenetics; | OER              | 199-211, 212-235 |
<table>
<thead>
<tr>
<th>Date</th>
<th>Event Description</th>
<th>Link</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>May 24</td>
<td>Term End Student Presentation.</td>
<td><img src="https://ghr.nlm.nih.gov/search?query=mutation" alt="Link" /></td>
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<tr>
<td>May 26 &amp; 31</td>
<td>Term End Student Presentation.</td>
<td><img src="https://ghr.nlm.nih.gov/search?query=Biotechnology" alt="Link" /></td>
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<tr>
<td>Jun 02 &amp; 07</td>
<td>Term End Student Presentation.</td>
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<tr>
<td>June 09 Thu</td>
<td>Final Examination; Cumulative: 9:00 AM – 11:00 AM.</td>
<td><img src="https://ghr.nlm.nih.gov/search?query=Biotechnology" alt="Link" /></td>
<td><strong>Good luck!</strong></td>
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**Grading:** There will be three written exams each carrying 20% X 3 = 60% for the final grade. In addition, there will be three Case Studies each carrying 3.33% x 3 = 10% and a Term End Presentation carrying additional 10% for the final grade. The final exam will carry 20%. Thus, three lecture exams, three case studies, one term end presentation and the final exam will add to 100% for the final grade.

Grades and % scores (tentative): A+ = 97-100; A, 94-96; A-, 90-93; B+ = 87-89; B, 84-86; B-, 80-83; C+ = 77-79; C, 74-76; C-, 70-73; D+ = 65-69; D, 60-64; F, ≤ 59

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1 Introduction: Overview of Genetics
Levels of genetics
What are genes and how do they work?
How are genes transmitted from parents to offspring?
How do scientists study genes?
Most genes do not function alone
Applications of genetics

https://openlab.citytech.cuny.edu/bio-oer/gene-expression
At the conclusion of this section’s material students will:
Understand the historical and current (modern) applications and approaches used in the field of human genetics.
Understand how genetic studies and practices include ethical, legal and social issues.

2 Cells
Cell components
The cell cycle (cell division) and cell death (apoptosis)
Cell to cell interactions (signal transduction)
Mitosis is essential for growth and cell replacement
Stem cells and cell specialization

OER https://ghr.nlm.nih.gov/primer
At the conclusion of this section’s material students will:
Understand that cells are the fundamental unit of living organisms, and be able to describe how each cellular component functions.
Be able to describe mitotic cell division, and explain its role in cell replacement.
Be able to state what stem cells are, how they function, and where they exist
3 DNA and Chromosomes
DNA Structure and Replication
DNA carries genetic information
Discovery of the structure of DNA (Watson & Crick model)
DNA contains two polynucleotide chains
RNA is a single-stranded nucleic acid
From DNA molecules to chromosomes
DNA replication depends on complementary base pairing


At the conclusion of this section’s material students will:
Know and distinguish the relationship between DNA, chromatin and a chromosome.
Recognize and identify parts of the DNA double helix.
Deduce proper results of DNA replication when given an example segment.
Be able to describe how genetic information is maintained

4 Gene Action: From DNA to Protein
DNA, not protein is the hereditary molecule
The link between genes and proteins
Genetic instructions are stored in DNA
The genetic code: the key to life
Tracing the flow of genetic information from nucleus to cytoplasm
Transcription produces genetic messages
Translation requires the interaction of several components
Polypeptides fold into three-dimensional shapes to form proteins
Protein structure and function are related


At the conclusion of this section’s material students will:
Describe how the information encoded in DNA specifies protein products.
Be able to trace the flow of genetic information from the DNA in the nucleus to the protein product in the cytoplasm.
Distinguish between transcription and translation and describe similarities and differences.
Explain the steps entailed for final protein product formation.
5  Meiosis and Development
The reproductive system
Meiosis
Gamete Maturation (spermatogenesis and oogenesis)
Equalizing the expression of X chromosomes in males and females
Prenatal Development
How is sex determined?
Defining sex in stages: chromosomes, gonads, and hormones
Mutations can uncouple chromosomal sex from phenotypic sex
Sex-influenced and sex-limited traits
Birth Defects (teratogens)
Maturation and Aging (accelerated aging, longevity)

At the conclusion of this section’s material students will:
Be able to describe which parts of the human reproductive system are involved in sex determination and development.
Know the sequence of sex determination from conception to determination of genetic sex, then gonadal sex, then phenotypic sex.
Understand and describe dosage compensation, and the difference between sex-influenced and sex-limited inheritance.
Name and describe teratogens that influence development
Discuss accelerated aging and longevity

6 Reproductive Technologies
Fertility and Sub fertility
Male fertility, female fertility, infertility tests
Assisted reproductive technologies
Donated sperm, donated uterus, in vitro fertilization,
ICSI Oocyte banking
Pre-implantation genetic diagnosis
Potential therapies to correct many disorders (gene therapy) Genetic counseling assesses reproductive risks
Extra Embryos

At the conclusion of this section’s material students will:
Be able to define and describe male and female fertility and infertility test
Be able to describe various assisted reproductive technologies utilized as childbearing options.
Be able to describe and discuss ethical issues in reproductive technology
Discuss pre-implantation technologies and potential therapies
Describe how/why surplus embryos are made and utilized
7 Chromosomes
Portrait of a chromosome – chromosome parts
Karyotypes – constructing and analyzing karyotypes
Visualizing chromosome
Variations in chromosome number (polyploidy, aneuploidy)
What are the risks for autosomal trisomy?
Variations in chromosome structure
(deletions, duplications, translocations, inversions)
Other forms of chromosomal abnormalities

https://ghr.nlm.nih.gov/chromosome

At the conclusion of this section’s material students will:
Be able to quantitatively and qualitatively describe the characteristic human chromosomal complement.
Name and describe sex chromosome aneuploidies and their consequences
Name and describe structural alternations within chromosomes
Identify and differentiate between normal and abnormal karyotypes and describe a condition to which they correspond.
Be able to describe several human syndromes based on the karyotype provided.

8 Single-Gene Inheritance
Following the inheritance of one gene – segregation
Mendel’s experiments
Single-gene inheritance in humans
  Mendel’s first law
Following the inheritance of two genes – independent assortment
  Mendel’s second law
Pedigree analysis


Beyond Mendel’s Laws
When gene expression appears to alter Mendelian ratios
  Multiple alleles, epistasis, pleiotropy, penetrance, expressivity
Maternal inheritance and mitochondrial genes
  Linkage

https://openlab.citytech.cuny.edu/bio-oer/genetics/co-dominance-and-multiple-alleles/
At the conclusion of this section’s material students will:
Describe a method of how traits are inherited
Describe how many basic genetic concepts we know about genetics was first identified in pea plants.
Describe Gregor Mendel’s experimental methodology utilizing pea plants to study one and more than one gene simultaneously.

Explain how Gregor Mendel’s experiments explain the separation and assortment of genes (alleles).

Explain how meiosis explains Gregor Mendel’s experimental results.

Describe Mendelian inherence in humans.

Be able to distinguish between autosomal dominant, autosomal recessive, and sex-linked dominant and recessive traits.

Explain holandric and maternal inheritance, illustrating with an example for each.

Explain how most human traits are controlled by more than one gene. Give several examples.

Provide a few examples of exceptions to Gregor Mendel’s laws.

Know how to interpret and design a pedigree.

9 Multifactorial Traits

Genes and the environment mold most traits
Polygenic traits
Fingerprint patterns
Height, hair color, skin color
Heart health
Weight


At the conclusion of this section’s material students will:

Explain how most human traits are controlled by >1 gene and giving several examples.
Distinguish between polygenic and multifactorial traits.
Describe how height, hair and eye color are inherited.
Relate gene expression to environmental influence (e.g.: heart health and weight).

10 Genetics of Immunity System

The importance of cell surfaces
The immune system – components and systems
Immune system responses: non-specific and specific defenses
Physical barriers, innate v. acquired immunity
Blood types, transplantation
Abnormal immunity – autoimmunity, allergies
Altering immune function – vaccines, transplants


At the conclusion of this section’s material students will:
Be able to distinguish between antibodies and antigens.
Be able to discuss how the immune system defends the body against infection.
Distinguish between general and specific defenses against infection.
Be able to discuss how antibodies are manufactured in the body during infection.
Describe blood types and their importance in blood transfusions and immune reactions between mother and fetus.
Be able to describe immune system disorders such as allergies and autoimmune reactions.

11 Genetics of Cancer
Cancer is genetic not usually inherited
Characteristics of cancer cells
Origins of cancer cells - cancer begins in a single cell
Cancer is a disease of the cell cycle
Cancer genes
A series of genetic changes causes some cancers
Brain tumors, colon cancer
Chromosome changes, hybrid genes, and cancer
Environmental; causes of cancer (carcinogens, cancer-environmental links)
Evolving cancer diagnosis and treatment


At the conclusion of this section’s material students will:
Be able to explain why cancer is considered a genetic disease.
List the steps that occur manifesting in cancer, from a single mutated cell to the disease.
Describe the mutations and steps in colon cancer.
Describe several common genetic changes which occur in cancer cells.
Distinguish between Inherited susceptibility and sporadic cancers
Name and describe potential contributing factors in cancer (e.g.: colon, lung)

12 Allele Frequencies
The importance of knowing allele frequencies (sec. 14.1)

DNA Profiling (sec 14.4)
Privacy (sec 14.5)

At the conclusion of this section’s material students will:
Be able to discuss the importance of knowing allele frequencies
Learn how DNA profiling is utilized in forensics and disasters
Discuss challenges to genetic privacy.

THEN...
And at the very end of the semester, many years later…
Be able to discuss human inheritance with confidence.