INTER AMERICAN UNIVERSITY OF PUERTO RICO
METRO CAMPUS
SCIENCE AND TECHNOLOGY FACULTY
DEPARTMENT OF NATURAL SCIENCES
BIMEDICAL SCIENCES PROGRAM

SYLLABUS

## I. GENERAL INFORMATION

| | | |
|---|---|---|
| Course Title | **:** | **HUMAN GENETICS** |
| Code and number | : | BMSC 2210 |
| Credits | : | Three (3) |
| Academic Term | : | |
| Professor | : | |
| Office hours | : | |
| Office Telephone | : | |
| Email | : | |

## II. DESCRIPTION

Fundamental concepts of human genetics, from the perspective of structure, function and transmission of genes; including interaction gene-gene and gene-environment. Emphasis on the molecular aspects of human inheritance, genetic etiology of diseases and research techniques in human genetics. Prerequisite: BIOL 1102

## III. OBJECTIVES

After completing the course, students will:

Understand the basic concepts of genetics at all levels: molecular, cellular and population.

1.1 Distinguish the structure of DNA and RNA.
1.2 Describe the mechanisms of replication, transcription and translation.
1.3Relate the genetic code to protein synthesis
1.4 Point out the ways in which gene expression is regulated
1.5 Identify other expression regulation that cells may have.
1.6 Distinguish between the different ways in which genetic traits are passed on to new generations

2.1 Identify the foundations of the classical genetics.
2.2 Predict the different patterns of inheritance based on the results of different crosses. .

2.3 Explain how sex is determined in humans.
2.4 Predict the transmission of genetic conditions linked to autosomal or sex linked chromosomes.
2.5 Solve problems related to different patterns of inheritance.

3. Explain the changes that genetic materials undergo and factors that cause them to change.
   3.1 Point out the sources of genetic variations.
   3.2 Explain the mechanisms of genetic variations.
   3.3 Explain the relationship between genetic variation and adaptation.
   3.4 Apply concepts of genetics to the study of human populations and their relation to evolutionary processes.
   3.5 To know the relationship between the basic concepts of genetics, biotechnology and man

## IV. CONTENT

A. Introduction

1. Genetic studies heredity and variation
2. The historical impact of genetics in society
3. When the genetics started?

B. Cell Cycle

1. Stages of the Cell Cycle
2. Mitosis
3. Meiosis
4. Gametogenesis
   a. ovogénesis
   b. espermatogénesis

C. Transmission of genes

1. Mendel's Experiments
2. Ley I: Segregation
3. Ley II: Independent Assortment
4. Mendelian Heredity in humans
5. Other gene actions
   a. one allele over another
   b. incomplete dominance
   c. codominance
   d. gene interactions, epistasis
6. Examples in humans

**Test 1 - From the begining til here -**

D. Probability and Statistics

1. Definition of probability
2. Rules of probability
3. Definition of statistics
   a. corroboration of hypotheses
   b. chi square test

E. Sex linked and Sex influenced heredity

1. Sex determination in humans
   a. gene control in sex determination
   b. allosomic chromosome

2. Heredity linked to chromosome X
3. Differences between allosomic and autosomic inheritance.
4. Sex limited inheritance
5. Sex influenced inheritance
6. Holandric inheritance
7. Barr bodies and dose compensation
8. Family trees
   a. Construction
   b. Interpretation

9. The importance of the human genetic pedigrees

F. Linked genes in autosomic inheritance

1. Linkage in eukaryotes
   a. genetic recombination
   b. differences between linked and unlinked genes
   c. construction of genetic maps
      1. dihybrid crosses - single recombination; nonrecombinant
      2. tri-hybrid cross - single and double recombination; non-recombinant
      3. Human chromosome maps

G. Cytogenetics and chromosomal variations
1. Changes in the arrangement of chromosomes
   a. delection
   b. inversion
   c. traslocation
   d. duplication
2. Variations in number of chromosomes
   a. Euploidy (poliploid organisms)
   b. aneuploidy
   c. uniparental disomy
   **d.** fragile sites on chromosomes –**Until here –Test 2**

H. Molecular Genetics

1. Molecular structures of nucleic acids

   a. DNA
   b. RNA

2. Description of DNA replication

3. Transcription
   a. Description of the process
   b. RNA types

4. Translation
   a. Description of the process
   b. Effect upon cell metabolism
   c. Metabolism and genetic disorders

5. Changes in the DNA molecule

   a. Nature of mutations
   b. Detection
   c. molecular bases of mutations

      1. Substitution
      2. Delection
      3. Insertion

6. Repair mechanisms

I. Genes and cancer

1. Cancer and malignant tumors
2. Mutations that cause a predisposition to cancer
3. Tumor suppressor genes
4. Oncogenes and cell cycle
5. Oncogenes and cancer
6. genomic changes and cancer
7. chromosomal changes and cancer
8. The environment and cancer

**Until here test 3**

J. Genetics and the immune system

   a. Genetics of the antibodies
   b. Inheritance of blood types

1. ABO
2. Rh
3. transplants and HLA
4. Genetic Disorders

K. Behavioral genetics

1. Genetic model of behavior
2. The effect of a single gene on human behavior
3. A single gene and aggressive behavior
4. Genetics and its relationship with personality disorders and schizophrenia
5. La genética y el comportamiento social

L. Recombinant DNA techniques and cloning
1. Restriction endonucleases
2. Techniques of recombinant DNA
3. The human genome project
4. Genetic markers RFLP
5. Prenatal testing for genetic disorders
6. The gene transfer technology and its applications
7. Ethics and new technology

M. Population genetics

1. Hardy-Weinberg equilibrium
2. Using the Hardy-Weinberg equilibrium in human genetics
3. Genetic diversity in human populations

**Until here test 4**

## V. ACTIVITIES

A. professor lectures
B. Discussion of exercises
C. Discussion of articles

## VI. ASSESSMENT CRITERIA

A. Partial exams : 4 of 100 points each

B. Deliver reviews of one scientific paper in human genetics (50 pts) and answer case studies (50 pts) for a total of 100 pts.

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## VII. RESOURCES

### Textbook

Cummings, Michael R. ***''Human Heredity Principles and Issues''***, Any edition Brooks/ Cole Thomson Learning.

## VIII. BIBLIOGRAPHY


Gene link 2.0 CD-ROM
Laboratorios virtuales de genética molecular CD/Web Site

### Book

*"Human Molecular Genetics 2",* Strachan, Tom y Read, Andrew P. (1999). 2nd Ed. Wiley-Liss.

Pearl, E., Solomon et al. 2001. Biología. 5ta ed. México. Mc Graw Hill Interamericana.

Smith, A. 2000**.** *Biochemistry and molecular biology*. New York. Oxford University Press.

Griffiths, Wessler, Lewontin, Gelbart, Suzuki y Miller. 2005. *Intoduction to Genetic Analysis, New York, 8 th ed., W.H. Freeman and Company*


### Audiovisual Resources:

1. *Genetics and heredity: the blueprint of life.* Chatsworth, Ca.Aims Multimedia, c2000 (QH315. G454)
2. Serie de Genética Humana de Millar:
   a. "Pedigree Patterms"MP. 545x
   b. "Chromosome bandung Techniques"MP. 452x
   c. "Chromosome Abnormalities: The sex Chromosome"MP. 524x
   d. "Chromosome Abnormalities: The Autosome"MP.543x

**Electrónic Resources**

a. *Genomics and hits Impact on Medicine and Society*. A2001 Primer.
http://www.ornl./hgmis/publicat/primer2001/index.html.

*b. Human Genetics*
http://www.emc.maricopa.edu/faculty/farabee/BIOBK/BioBookhumgen.html

*c. The Human Genome: A Guide to Online Information*
http://www.cum.edu/gec/prof/geneeisi.html

d. Human Genetics Now
http://now.ilrn.com/cummings7

a. Human Heredity Companion Website for Students
http://biology.brookscole.com/cummings7

**Basic concepts of mitosis and meiosis**

**quiz 1 diferencias entre mitosis y meiosis**
http://highered.mcgraw-hill.com/sites/0072495855/student_view0/chapter28/animation__comparison_of_meiosis_and_mitosis__quiz_1_.html

**quiz 2 differences between mitosis and meiosis**
http://highered.mcgraw-hill.com/sites/0072495855/student_view0/chapter28/animation__comparison_of_meiosis_and_mitosis__quiz_2_.html

**control of cell cycle**
http://images.google.com.pr/imgres?imgurl=http://www.the-simple-homeschool.com/image-files/cyclincheckpoints.gif&imgrefurl=http://www.the-simple-homeschool.com/cell-cycle-control.html&usg=__PMG_hnZGb8FtBgBPLja8bCvdHvo=&h=585&w=600&sz=73&hl=es&start=2&itbs=1&tbnid=ih_Bd2UXPDgTRM:&tbnh=132&tbnw=135&prev=/images%3Fq%3Dmolecular%2Bcontrol%2Bcell%2Bcycle%26hl%3Des%26sa%3DG%26gbv%3D2%26tbs%3Disch:1

**control of cell cycle**
http://www.youtube.com/watch?v=QGx50C1w8YY&feature=player_embedded

http://www.cellsalive.com/mitosis.htm
http://www.cellsalive.com/meiosis.htm

http://www.cellsalive.com/cell_cycle.htm

http://cancer-gene.com/videos/mitosis-en-ancer.html

http://library.thinkquest.org/C004535/mitosis_animation.html

http://www.sumanasinc.com/webcontent/animations/content/meiosis.html

http://www.johnkyrk.com/meiosis.html

**how meiosis work**
http://highered.mcgraw-hill.com/sites/0072495855/student_view0/chapter28/animation__how_meiosis_works.html

**spermatogenesis 1**
http://highered.mcgraw-hill.com/sites/0072495855/student_view0/chapter28/animation__spermatogenesis__quiz_1_.html

**spermatogenesis 2**
http://highered.mcgraw-hill.com/sites/0072495855/student_view0/chapter28/animation__spermatogenesis__quiz_2_.html

**meiosis stages**
http://highered.mcgraw-hill.com/sites/0072495855/student_view0/chapter28/animation__stages_of_meiosis.html

**característics unique to meiosis**
http://highered.mcgraw-hill.com/sites/0072495855/student_view0/chapter28/animation__unique_features_of_meiosis.html

**Random orientation of chromosomes during meiosis**
http://highered.mcgraw-hill.com/olc/dl/120074/bio18.swf

**Animations of Mendel laws**
http://recursostic.educacion.es/secundaria/edad/4esobiologia/4quincena6/ventanas/leyes_mendel.htm

**Chi square**
http://www.youtube.com/watch?feature=endscreen&NR=1&v=mWVD0yAZFn4

**Sex linked inheritance**
http://www.learnerstv.com/animation/animation.php?ani=13&cat=biology

**Chromosome mapping in humans**
http://es.wikipedia.org/wiki/Citogen%C3%A9tica_humana

**El glosario hablado de genética**
http://itunes.apple.com/us/app/talking-glossary-of-genetics/id428340581?mt=8&ls=1
en español http://www.genome.gov/GlossaryS/

## IX. SPECIAL NOTES

### 1. *Servicios Auxiliares o Necesidades Especiales*

Students requiring special assistance or support services should I apply for it at the beginning of course or as soon as s/he acquires the knowledge of his/her needs, through the corresponding record in the Office of Professional Counselor, Mr. José Rodríguez, located in the Program University Counseling in the first floor.

### 2. *Honesty, fraud and plagiarism*

Dishonesty, fraud, plagiarism and other improper behavior with regards to academic work constitute a major infraction sanctioned by General Student Regulations. Major infractions, as provided in General Student Regulations may result in suspension from the University for a definite time more than a year or permanent expulsion from the University, among other penalties.

### 3. Using electronic devices

Cell phones and other electronic devices that could interrupt the teaching and learning process or disrupt a milieu for academic excellence will be disabled. Critical situations will be addressed, as appropriate. The use of electronic devices that allow access, store or send data during tests or examinations is prohibited.

**Revised in agosto of 2015**