Course Prefix and Number: BLGY 244  
Credit Hours: 3

Course Title: Introduction to Human Genetics

Course Prerequisites: Biology 101 or Biology 225 or Biology 230


Course Description:
General principles of genetics, heredity, and genetic analysis. Emphasis is placed on inheritance of human disease.

Learning Outcomes:

At the end of this course students will:
A. apply knowledge of anatomy and physiology to processes of human reproduction and inheritance;
B. apply the concepts and rules of genetics to appreciate the inheritance of diseases and disorders;
C. recognize the characteristics of selected genetics conditions; and
D. appreciate the social and ethical considerations associated with advances in human genetics.

To achieve the learning outcomes, the student will
1. describe the development of genetics as a discipline (A)
2. explain the concept of genes, the basic way they work, and how they are transmitted from parents to offspring (B)
3. describe methods employed to study genes (B)
4. communicate the many ways in which the eugenics movement has had an impact on society and the impact on our lives of biotechnology and the Human Genome Project (C)
5. describe the structure and organization of cells in higher organisms (A)
6. describe chromosomes as cellular organelles that carry genetic information (A)
7. explain mitosis and the cell cycle and the significance of mitosis and the genetic control of the cell cycle (A)
8. explain the process of meiosis and the production of genetic variety among gametes (A)
9. contrast the similarities and differences between spermatogenesis and oogenesis (A)
10. perform Mendel’s monohybrid crosses and utilize the principle of segregation, the concept of dominance and recessiveness, and the presence of genes in pairs. (B)
11. perform the dihybrid cross and utilize the accompanying principle of independent assortment. (B)
12. apply Mendelian principles to the inheritance of traits in humans. (B)
13. explain the fact that many genes have multiple alleles and not all alleles can be described as simply dominant or recessive. (B)
14. utilize the symbols and conventions used in constructing a pedigree and explain the reason for doing pedigree analysis in human genetics. (B)
15. discuss the difficulties inherent in studying genetic traits in humans, relying on indirect, observational methods rather than direct, experimental methods. (B)
16. list the six modes of inheritance of human traits and the guidelines for determining the mode of inheritance of a trait. (B)
17. describe the expectations for the behavior of autosomal recessive traits in a pedigree, and examples of autosomal recessive traits in humans. (B)
18. predict the expectations for the behavior of autosomal dominant traits in a pedigree, and examples of autosomal dominant traits in humans. (B)
19. describe the expectation and behavior of X-linked (dominant and recessive) and Y-linked traits in pedigrees, and examples in humans. (B)
20. discuss mitochondrial inheritance and examples. (B)
21. utilize knowledge of variations in gene expression, such as penetrance and expressivity, that modify the phenotype of a gene. (B)
22. compare the distinction between continuous and discontinuous traits. (B)
23. discuss the varying role of environment in determination of phenotypes. (B)
24. list the characteristics of traits that demonstrate continuous variation, including threshold effects and regression to the mean. (B)
25. define the meaning and measurement of heritability, especially via twin studies. (B)
26. discuss the complex relationship between genotype and environment in multifactorial traits. (B)
27. define terms used to describe chromosome morphology. (A)
28. describe the normal human karyotype and how it is produced. (A)
29. contrast the concepts of polyploidy and aneuploidy. (B)
30. describe the mechanisms (cytokinesis failure and double fertilization) that give rise to polyploidy, and the inviability of the polyploidy condition. (B)
31. explain the mechanism (nondisjunction) that gives rise to aneuploidy and the very poor viability of the aneuploid condition. (B)
32. describe details of several autosomal trisomic conditions, including Down syndrome, and the risk factors that predispose to aneuploid conditions. (B)
33. explain the basis of aneuploidies of the sex chromosomes, specifically Turner Syndrome, Klinefelter’s syndrome, and XYY syndrome. (B)
34. discuss two methods of prenatal diagnosis: amniocentesis and chorionic villus sampling. (B)
35. compare variations in chromosome structure including deletions, translocations, and fragile sites, and their consequences. (B)
36. utilize knowledge of the male and female reproductive systems to describe gamete formation. (A)
37. list the major feature and the stages in human development (events of the three trimesters). (A)
38. define a teratogen and how alcohol consumption leads to teratogenic effects. (B)
39. discuss factors that determine sex, the sex ratio, and how it changes throughout life. (A)
40. explain details of sexual differentiation and the role of the Y chromosome in this process. (A)
41. compare how sex can be defined at the chromosomal, gonadal, and phenotypic levels, and how these are not consistent in some individuals. (B)
42. discuss the concept of dosage compensation by X-inactivation. (B)
43. discuss the historical evidence proving that DNA is the genetic material and providing information about the structure of DNA. (B)
44. describe the chemical composition and structure of the components of DNA. (A)
45. describe the Watson-Crick model of DNA. (A)
46. describe chromosome structure from the level of DNA to the metaphase chromosome. (A)
47. discuss the mechanisms of DNA replication. (A)
48. compare sex-limited and sex-influenced traits and imprinted genes. (A)
49. explain how genetic information stored in DNA is used to synthesize proteins. (A)
50. discuss the nature and characteristics of the genetic code. (A)
51. describe the process of transcription and the processing of messenger RNA. (A)
52. explain the process of translation, including initiation, elongation, and termination. (A)
53. describe the polypeptide product, including the importance of correct folding into proteins and the functions that proteins perform in the cell. (A)
54. discuss the organization of a typical human (eukaryotic) gene, with promoter, introns and exons, and terminator (A)
55. explain the concept that proteins are the end products of genes. (A)
56. discuss the fact that proteins that function as enzymes participate in metabolic pathways. (A)
57. explain the idea that mutations can produce defects in enzymes that affect metabolic pathways, producing phenotypic effects due to either lack of a necessary product or accumulation of a toxic pathway substance; details of PKU provide a useful example. (A)
58. list functional capacities of proteins in other roles, including receptors and transport.
59. describe the phenotypic effects of mutations that affect the amino acid sequence of receptor proteins, and of transport proteins such as the hemoglobin defects sickle cell anemia and the thalassemias. (A)
60. compare the role of gene differences in pharmacogenetics and ecogenetics. (B)
61. explain how mutations are detected in humans. (B)
62. discuss the methods used in measuring the rate of mutation in humans, and the mutation rate that is assigned as an average for human genes. (B)
63. explain the ways in which various types of radiation and chemicals act as mutagens. (B)
64. discuss the molecular basis of mutations, including base substitutions, deletions and insertions, and trinucleotide repeats, including the phenotypic consequences of changing the number of such repeats. (B)
65. summarize the importance of DNA repair systems. (A)
66. explain the concept that mutations of specific genes are the causes of cancer and the application of this concept to several forms of familial cancer. (B)
67. explain the concept of proto-oncogenes and tumor suppressor genes as normal components of the genome that can be oncogenic when mutated or disrupted. (B)
68. compare the relationship between genomic instability in general and cancer, and particularly the roles of DNA repair mechanisms and chromosome breaks with specific forms of cancer. (B)
69. list methods of epidemiology. (A)
70. explain the role of environmental agents in causing somatic mutations leading to cancer. (B)
71. discuss the importance of behavior in controlling risk of cancer. (B)
72. describe the concept of cloning organisms, cells, and molecules. (A)
73. explain the use of restriction enzymes, ligase, and vectors in the construction of cloned DNA sequences and genetic libraries. (B)
74. describe means of finding a specific gene in a library, blotting and DNA sequencing, and how they are used in analysis of cloned sequences. (B)
75. explain the phenomenon of genomic imprinting and its role in the expression of genetic disorders. (B)
76. discuss the various situations in which individuals and embryos are tested for genetic disease. (B)
77. explain the use of DNA profiles in the law and forensics to identify individuals. (B)
78. participate in a discussion of ethical issues raised by biotechnology. (C)
79. describe the purpose and progress-to-date of the Human Genome Project and the technological approaches used in genomics to map, identify, and sequence genes. (B)
80. explain how genomics, bioinformatics, and proteomics are defined, how they have revolutionized the study of human genetic disorders, and how they hold the promise of developing better treatments including ethical concerns and future potential of human genomics. (B, C)
81. explain how contraceptive techniques for the male and female work and the available techniques for correcting specific types of male and female infertility. (A)
82. discuss the goals, history, and methods of gene therapy and the ethical issues inherent in assisted reproductive technologies and gene therapy. (B)
83. describe the profession of genetic counseling. (B)
84. describe the inflammatory response and the complement system and how the structure and function of antibodies and the mechanism of antibody formation, including the genetic recombination that generates distinctive antibodies. (A)
85. describe the immune response, including similarities and differences between antibody-mediated and cell-mediated, and the memory function of both. (A)
86. discuss the role of genetics and the immune system in transplantation. (A)
87. apply genetics and clinical significance to the ABO and Rh blood groups. (B)
88. list diseases of the immune system, autoimmune diseases, and allergies. (B)
89. discuss the idea that most behavior traits represent complex phenotypes with multifactorial inheritance and the range of methods used to study human behavior genetics. (B)
90. describe the use of animal model systems to establish that aspects of behavior are under genetic control and to provide estimates of heritability. (B)
91. describe single gene defects that bring about altered behavior in humans. (B)
92. discuss the complex nature of behavior traits such as bipolar illness and schizophrenia, and other multifactorial traits. (B)
93. describe the need for new and innovative approaches to the study of the genetic basis of human behavior. (B)

Course Requirements

In order to receive a grade of “C” the student must earn 70% of the total possible points for the courses and achieve all of the following course requirements.

- minimum average of 70% on tests including the mandatory final exam
- satisfactory participation in case studies and other activities
students enrolled in this course in an online format will be required to take the final exam in a proctored environment; on the BPCC campus or with an approved proctor

Course Grading Scale:

A- 90% or more of total possible points and meets all course requirements
B- 80% or more of total possible points and meets all course requirements
C- 70% or more of total possible points and meets all course requirements
D- 60% or more of total possible points and meets all course requirements
F- less than 60% of total possible points or does not meet all course requirements

Attendance Policy: The college attendance policy is available at http://www.bpcc.edu/catalog/current/academicpolicies.html

Nondiscrimination Statement

Bossier Parish Community College does not discriminate on the basis of race, color, national origin, gender, age, religion, qualified disability, marital status, veteran's status, or sexual orientation in admission to its programs, services, or activities, in access to them, in treatment of individuals, or in any aspect of its operations. Bossier Parish Community College does not discriminate in its hiring or employment practices.

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Reviewed by J Coston/April 2017