

Course Specification

— (Postgraduate Programs)

Course Title: Human Genetic Diseases
Course Code: BIO-6014
Program: Master of Science in Applied Biology (Cell and Molecular Biology)
Department: Biology
College: Science
Institution: Imam Mohammad Ibn Saud Islamic University
Version: 1
Last Revision Date: <i>Pick Revision Date.</i>



Table of Contents

A. General information about the course:	3
B. Course Learning Outcomes (CLOs), Teaching Strategies and Assessment Methods:	Error! Bookmark not defined.
C. Course Content:.....	Error! Bookmark not defined.
D. Students Assessment Activities:	7
E. Learning Resources and Facilities:.....	7
F. Assessment of Course Quality:.....	8
G. Specification Approval Data:.....	8

عبد المحسن بن أحمد العزيمي، عبد الله بن محمد بن الناصر

عبد المحسن بن أحمد العزيمي، عبد الله بن محمد بن الناصر



A. General information about the course:

1. Course Identification:

1. Credit hours: (Lecture 2+ Lab 0)

2. Course type

A. ☐ University ☐ College ☐ Department ☒ Track

B. ☐ Required ☒ Elective

3. Level/year at which this course is offered: (undetermined level)

4. Course general Description:

This graduate course is an advanced course on the molecular genetics of human disease. It provides the students with an in-depth understanding of the major concepts in Human Genetics. The course focuses on the different types of genetic disorders along with illustrations of significant examples such as autosomal dominant, autosomal recessive, X-linked dominant, and X-linked recessive. The course emphasizes a contemporary overview of genetic variations within populations and the forces that shape them. This involves the changes in the frequencies of genetic variation in populations over space and time. The course also covers the identification and assessment of mutations that contribute to the development of abnormal phenotypes

5. Pre-requirements for this course (if any):

None

عبد المحسن بن أحمد العنزي

6. Co-requisites for this course (if any):

None

7. Course Main Objective(s):

The objective of this graduate course is to introduce the students to the modern approaches used to identify genetic factors in health and disease, and analyze variation within populations. The students through studying this course get familiar with concepts and methods such as complex traits, population studies, the molecular basis of genetic disease and Pedigree analysis.

عبد المحسن بن أحمد العنزي

2. Teaching Mode: (mark all that apply)

No	Mode of Instruction	Contact Hours	Percentage
1	Traditional classroom	√	100%
2	E-learning		
3	Hybrid - Traditional classroom - E-learning		
4	Distance learning		

3. Contact Hours: (based on the academic semester)

No	Activity	Contact Hours
1.	Lectures	30
2.	Laboratory/Studio	-
3.	Field	-
4.	Tutorial	-
5.	Others (specify).....	
	Total	30

B. Course Learning Outcomes, Teaching Strategies, Assessment Methods

Code	Course Learning Outcomes	Code of CLOs aligned with program	Teaching Strategies	Assessment Methods
1.0	Knowledge and understanding			
1.1	List the different types of genetic disorders and Clarify the theoretical basis of population genetics	K1	Two credits weekly lecture	-Quizzes -Assignments -Presentations -Written exams
1.2	Explain how variations in the coding and non-coding components of the human genome can result in genetic diseases	K2	Two credits weekly lecture	Quizzes -Assignments -Presentations -Written exams

Code	Course Learning Outcomes	Code of CLOs aligned with program	Teaching Strategies	Assessment Methods
1.3	Recognize the significance of genetic testing in the diagnosis, prevention, and treatment of genetic diseases	K3	Two credits weekly lecture	Quizzes -Assignments -Presentations -Written exams
2.0	Skills			
2.1	Interpret and analyze the results of common genetic tests	S1	-Two credits weekly lecture -Tutorials	-Assignments -Presentations -Written exams
2.2	Investigate the frequency of genetic diseases by applying the Hardy-Weinberg equilibrium	S2	-Two credits weekly lecture -Tutorials	-Assignments -Presentations -Written exams
2.3	Relate between the encountered diseases and genetic factors	S3	-Two credits weekly lecture -Tutorials	-Assignments -Presentations -Written exams
3.0	Values, autonomy, and responsibility			
3.1	Discuss the latest advances in genomic technologies and their applications to the identification of genetic diseases	V1	Group discussions	-Presentation -Reports
3.2	Appraise the implications of genetic disorders on patients, families, tribes, and society	V2	Group discussions	-Presentation -Reports
3.3	Demonstrate the ability to evaluate and critically review literature describing the recent research into human genetic	V3	Group discussions	-Presentation -Reports

C. Course Content:

No	List of Topics	Contact Hours
1.	Fundamental Concepts in Human Genetics: Genetic Terminology, genetic information, sex chromosomes, autosomes, homologous chromosomes, replication, Human Genome, pedigree	2
2.	Molecular Basis of Genetic Disease: Genetic Diseases and Faulty DNA Repair Diseases caused by chromosomal aberrations Diseases associated with single-gene Mendelian inheritance Diseases associated with single-gene non-Mendelian inheritance Diseases caused by multifactorial inheritance	3
3.	Inheritance of Traits in Humans and Pedigree Analysis: The inheritance of chromosomes—Meiosis Symbols Used in Pedigrees Analysis of Pedigrees Autosomal Recessive Traits Autosomal Dominant Traits X-Linked Recessive Traits X-Linked Dominant Traits Y-Linked Traits	2
4.	Studying Twins and Adoptions: Concordance in Twins A Twin Study of Asthma Adoption Studies	2
5.	Rare diseases and Common diseases: AA amyloidosis, Adrenoleukodystrophy (ALD), Ehlers-Danlos syndrome, Mitochondrial diseases, Usher syndrome, Chromosomal disorders, Multifactorial disorders, Monogenic disorders The Saudi Genome Project: -Pathogenic Variants	3
6.	Genetic Counseling and Genetic Testing: Genetic Counseling Genetic Testing Interpreting Genetic Tests Direct-to-Consumer Genetic Testing	3



	Genetic Discrimination and Privacy Risk of Consanguinity on Genetic Diseases	
7.	Gene Pool of a Population: Allelic frequencies, phenotypic frequencies, genotypic frequencies	2
9.	Hardy-Weinberg Law: Genotypic Frequencies at Hardy–Weinberg Equilibrium Closer Examination of the Assumptions of the Hardy–Weinberg Law Implications of the Hardy–Weinberg Law Extensions of the Hardy–Weinberg Law Testing for Hardy–Weinberg Proportions Estimating Allelic Frequencies with the Hardy–Weinberg Law	3
10.	Genetic Variation in Populations and their Analysis: -Nonrandom Mating -Evolutionary Forces: mutations, genetic recombination, gene flow or gene migration, genetic drift, natural selection	2
11.	Populations substructure	2
Total		30

D. Students Assessment Activities:

No	Assessment Activities *	Assessment timing (in week no)	Percentage of Total Assessment Score
1.	Midterm exam-1	5 th week	20%
2.	Midterm exam-2	10 th week	20%
3.	Assignments, presentations, data search, participation	Throughout the course duration	20%
...	Final exam	16 th week	40%

*Assessment Activities (i.e., Written test, oral test, oral presentation, group project, essay, etc.)

E. Learning Resources and Facilities:

1. References and Learning Resources:



Essential References	-Benjamin A Pierce. (2020). Genetics: a conceptual approach. 7 th edition W.H. Freeman. ISBN-13-978-1319216801.
Supportive References	https://www.ucl.ac.uk
Electronic Materials	
Other Learning Materials	

2. Educational and Research Facilities and Equipment Required:

Items	Resources
facilities (Classrooms, laboratories, exhibition rooms, simulation rooms, etc.)	Classroom
Technology equipment (Projector, smart board, software)	Projector, smart board
Other equipment (Depending on the nature of the speciality)	

F. Assessment of Course Quality:

Assessment Areas/Issues	Assessor	Assessment Methods
Effectiveness of teaching	Student	Indirect
Effectiveness of students' assessment	Faculty	Direct
Quality of learning resources	Faculty	Direct
The extent to which CLOs have been achieved	Faculty	Direct
Other		

Assessor (Students, Faculty, Program Leaders, Peer Reviewer, Others (specify))

Assessment Methods (Direct, Indirect)

G. Specification Approval Data:

COUNCIL /COMMITTEE	BIOLOGY DEPARTMENT COUNCIL
REFERENCE NO.	23
DATE	23/8/1445