Jordan University of Science and Technology
Faculty of Applied Medical Sciences
Allied Medical Sciences Department

LM441 Diagnostic Molecular Biology And Cytogenetic - JNQF Level: 7

First Semester 2023-2024

Course Catalog
3 Credit Hours. It is a theoretical course designed to provide students with a comprehensive understanding of the theoretical principles of molecular biology and cytogenetics and their applications at the diagnostic level. The topics of this course are Modes of inheritance, Pedigree analysis, Genetic Variation and Mutation Analysis, DNA Sequencing and Next-Generation Sequencing (NGS), application of molecular and cytogenetic diagnostic techniques in the health service including knowledge about common genetic diseases, clinical features, screening, and available diagnostic tests, population screening, cellular pathology and how to interpret and report the results of the tests. This course will also cover genetic counselling and ethical and regulatory considerations.
Teaching Method: On Campus

Text Book	
Title	Molecular Diagnostics: Fundamentals, Methods, and Clinical Applications
Author(s)	Lela Buckingham and Maribeth L. Flaws
Edition	3rd Edition
Short Name	1
Other Information	2019

Instructor	
Name	**Dr. MOHAMMAD AL SHBOUL**
Office Location	-
Office Hours	Sun : 10:00 - 12:00 Mon : 10:00 - 12:00 Tue : 11:00 - 12:00 Wed : 11:00 - 12:30
Email	maalshboul@just.edu.jo

Class Schedule & Room

Section 1:
Lecture Time: Mon, Wed : 13:00 - 14:30
Room: NB49

Tentative List of Topics Covered		
Weeks	**Topic**	**References**
Week 1	Introduction to Diagnostic Genetics and Cytogenetics ? Overview of diagnostic genetics and its significance in healthcare	From **1**
Week 2	Basic principles of genetics: genes, alleles, and inheritance patterns (I) Constructing and analyzing family pedigrees Mendelian genetics: autosomal dominant, autosomal recessive	From **1**
Weeks 3, 4	Basic principles of genetics: genes, alleles, and inheritance patterns (II) Constructing and analyzing family pedigrees Mendelian genetics: X-linked inheritance Non- Mendelian genetics: mitochondrial inheritance, genomic imprinting and multifactorial Penetrance and expressivity of genetic traits	From **1**
Weeks 5, 6	Genetic Diseases and Variants Overview of genetic diseases: single gene disorders, chromosomal abnormalities, multifactorial disorders ? Identification and classification of genetic variants ? Role of genetic variants in disease susceptibility and pathogenesis	From **1**
Weeks 7, 8	Sequencing Techniques in Diagnostic Genetics, Principles of DNA sequencing: Sanger sequencing, Introduction to next-generation sequencing (NGS) technologies, Applications of sequencing in disease diagnosis and research	From **1**
Weeks 10, 11	Cytogenetics and Chromosomal Abnormalities: Structure and function of chromosomes, Techniques in cytogenetic analysis: karyotyping, fluorescence in situ hybridization (FISH), Common chromosomal abnormalities and their clinical implications	From **1**
Weeks 12, 13	Genetic Testing and Interpretation: Types of genetic and cytogenetic testing: diagnostic testing, predictive testing, carrier testing, prenatal and preimplantation genetic testing, Laboratory methods for genetic testing: PCR, DNA sequencing, microarrays, and others, Interpretation of genetic test results and reporting	From **1**
Week 14	Case Studies and Practical Applications: Analyzing selected case studies in diagnostic genetics and cytogenetics, Interpretation the tests results	From **1**
Week 15	Pharmacogenetics and Personalized Medicine: Introduction to pharmacogenetics: genetic variations affecting drug metabolism and response ? Applications of pharmacogenetics in drug development and treatment, Integration of personalized medicine approaches in clinical practice	From **1**
Week 16	Genetic Counseling and Ethical Considerations: Principles of genetic counseling: patient education, risk assessment, informed consent	From **1**

Mapping of Course Outcomes to Program Outcomes and NQF Outcomes	**Course Outcome Weight (Out of 100%)**	**Assessment method**
Construct, analyze and interpret family pedigree charts and understand the patterns of inheritance, including Mendelian inheritance, X-linked inheritance, and multifactorial inheritance [10SLO1] [10L7K1]	10%	First Exam

Differentiate between different classes of variants and the functional effect [10SLO1] [10L7K1]	10%	First Exam
Understand Genetic Variation and Mutation Analysis [10SLO1] [10L7K1]	10%	First Exam
Discuss the different methods of DNA sequencing and their diagnostic purpose [10SLO1] [10L7K1]	10%	First Exam, Second Exam
Discuss examples of various genetic diseases, clinical features, diagnosis and treatment [10SLO1] [10L7K1]	10%	Second Exam
Understand the purpose of pharmacogenetics and personalized Medicine [10SLO1] [10L7K1]	10%	Second Exam
Explain the types of chromosomal abnormalities and their association with diseases [10SLO1] [10L7K1]	10%	Second Exam
Analyze pedigree charts [10SLO1] [10L7S2]	10%	First Exam
Interpret karyotyping and genetic results [10SLO1] [10L7S2]	10%	
Debate the ethical, legal, and social implications (ELSI) of genetic testing [10SLO1] [10L7S2]	10%	
To review examples of case studies related to genetics and chromosomal abnormalities. [1SLO1] [10L7C2]	10%	
Highlight the importance of genetics and cytogenetic testing [10SLO1] [10L7C2]	10%	

Relationship to Program Student Outcomes (Out of 100%)											
SLO1	SLO2	SLO3	SLO4	SLO5	SLO6	MSLO1	MSLO2	MSLO3	MSLO4	MSLO5	MSLO6
120											

Relationship to NQF Outcomes (Out of 100%)		
L7K1	L7S2	L7C2
70	30	20

Evaluation	
Assessment Tool	**Weight**
First Exam	30%
Second Exam	30%
Final Exam	40%

Policy

Attendance	Students are expected to attend all classes. A student should not miss more than 20% of the classes during a semester. Those exceeding this limit will receive a failing grade regardless of their performance and the grade in this course will be considered (zero), but if the absence is due to illness or a compulsive excuse accepted by the dean of the college.
Make-up Exam	Make-up exams are entitled to students who miss the exam with an accepted legal or medical excuse endorsed by the instructor within 24 hours after the scheduled exam (please review university regulations for more details).

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