Form: Course Syllabus	Form Number	EXC-01-02-02A
	Issue Number and Date	2/3/24/2022/2963 05/12/2022
	Number and Date of Revision or Modification	
	Deans Council Approval Decision Number	265/2024/24/3/2
	The Date of the Deans Council Approval Decision	2024/1/23
	Number of Pages	06

1.	Course Title	Medical Genetics
2.	Course Number	0504321
3.	Credit Hours (Theory, Practical)	2 Theory
	Contact Hours (Theory, Practical)	28 Lectures
4.	Prerequisites/ Corequisites	--
5.	Program Title	Doctor of Medicine
6.	Program Code	05
7.	School/ Center	School of Medicine
8.	Department	Physiology and Biochemistry
9.	Course Level	Bachelor
10.	Year of Study and Semester (s)	Third year/ Second Semester
11.	Program Degree	Bachelor
12.	Other Department(s) Involved in Teaching the Course	--
13.	Learning Language	English
14.	Learning Types	☐Face to face learning ☐Blended ☐ Fully online
15.	Online Platforms(s)	☐Moodle ☐Microsoft Teams
16	Issuing Date	January 2024
17.	Revision Date	May 2025

18. Course Coordinator:

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19. Other Instructors:

None

20. Course Description:

A- Course Description:

This course covers the study of chromosomes and heredity, genetic linkage, chemistry of the gene, mitosis and meiosis, gamete formation, mechanisms of transfer of genetic traits, genetic map, sex determination, sex-linked characteristics, human genetic disorders and their diagnosis and management, and genetic engineering.

B- Aims:

The aims of this course are to provide students with a thorough understanding of genetic principles and applications. Students will learn about chromosomes, heredity, and genetic diversity. They will explore chromosomal aberrations, Mendelian and non-Mendelian inheritance, and sex-linked traits. The course covers topics like risk assessment, human genetic variation, and biochemical genetics. Specialized areas such as cancer genetics will be explored. By the end of the course, students should be equipped to interpret genetic information, assess risks, and understand key advancements in genetics.

21. Program Intended Learning Outcomes: (To be used in designing the matrix linking the intended learning outcomes of the course with the intended learning outcomes of the program)

PLO's	***National Qualifications Framework Descriptors***		
	Competency (C)	**Skills (B)**	**Knowledge (A)**
1.	☐	☐	☒
2.	☐	☒	☐
3.	☒	☐	☐
4.	☐	☐	☒
5.	☐	☒	☐
6.	☒	☐	☐
7.	☐	☐	☒
8.	☒	☐	☐

* Choose only one descriptor for each learning outcome of the program, whether knowledge, skill, or competency.

22. Course Intended Learning Outcomes: (Upon completion of the course, the student will be able to achieve the following intended learning outcomes)

Course ILOs #	The learning levels to be achieved						Competencies
	Remember	Understand	Apply	Analyse	Evaluate	Create	
1.	✓	✓					Recognize the principles of chromosomes, heredity, genetic linkage, and the chemistry of the gene.
2.	✓	✓	✓	✓	✓		Interpret human genetic variation, understanding its implications and how it contributes to the diversity observed in clinical genetics.
3.	✓	✓	✓	✓	✓		Analyze and interpret genetic information to assess clinical implications, including the diagnosis and management of human genetic disorders.
4.		✓	✓	✓	✓	✓	Distinguish between Mendelian and non-Mendelian

							inheritance and apply this knowledge to identify features of autosomal dominant and autosomal recessive pedigrees and diseases.
5.		✓	✓	✓	✓	✓	Assess and communicate risks associated with genetic traits and disorders, integrating knowledge from risk assessment sessions.
6.		✓	✓	✓	✓	✓	Demonstrate the ability to integrate genetic knowledge into clinical practice, construct appropriate diagnostic and therapeutic

							strategies, considering environmental, social, cultural, and psychological factors.

23. The matrix links the intended learning outcomes of the course -CLO's with the intended learning outcomes of the program -PLOs:

PLO's * / CLO's	1	2	3	4	5	6	Descriptors		
							A	B	C
1	✓						✓		
2						✓		✓	
3			✓						✓
4		✓					✓		
5								✓	
6				✓	✓				✓
7							✓		
8									✓

***Linking each course learning outcome (CLO) to only one program outcome (PLO) as specified in the course matrix.**

****Descriptors are determined according to the program learning outcome (PLO) that was chosen and according to what was specified in the program learning outcomes matrix in clause (21).**

24. Topic Outline and Schedule:

Week	Lecture	Topic	Student Learning Outcome (SLO)	Descriptors **	Learning Types (Face to Face/Blended/ Fully Online)	Platform Used	Synchronous / Asynchronous Lecturing	Evaluation Methods	Learning Resources
1	1.1	**Genetic Diversity, Laws of Segregation, and Independent Assortment**	Define and explain the principles of genetic diversity. Describe the laws of segregation and independent assortment.	K K	Fully online	Teams	Asynchronous Lecturing	Written exam	A
	1.2	**Karyotyping, Chromosome Structure, and Nomenclature**	Describe the theoretical foundations of karyotyping, including the principles and methodologies involved in the analysis of chromosome structure Label the structures of chromosomes	K K	Fully online	Teams	Asynchronous Lecturing	Written exam	A
2	2.1	**Autosomal Chromosomes and Numerical Chromosomal Aberrations**	Determine key components of chromosomes, including telomeres, centromeres, and unique DNA sequences, demonstrating knowledge of their structural organization. Analyse numerical chromosomal aberrations resulting from nondisjunction during meiosis, encompassing aneuploidy and polyploidy. Identify and explain the clinical implications of specific chromosomal abnormalities, such as Down syndrome (Trisomy 21), Trisomy 18 (Edward syndrome), and Trisomy 13 (Patau syndrome), integrating both structural and numerical perspectives.	K S K	Fully online	Teams	Synchronous Lecturing	Written exam	A
	2.2	**Sex Chromosomes**	Explain the consequences of nondisjunction of sex chromosomes, detailing conditions such as Klinefelter syndrome (XXY) and Turner syndrome (X0), demonstrating knowledge	K	Fully online	Teams	Asynchronous Lecturing	Written exam	A

			of sex chromosome aneuploidies and their prevalence. Describe the Chromosomal Basis of Sex Explain the significance of sex chromosomes in genetic disorders.	K K					
3	3.1	**Structural Chromosomal Aberrations**	Explain the genetic basis and manifestations of disorders caused by structurally altered chromosomes, such as cri du chat syndrome and chronic myelogenous leukemia (CML), demonstrating knowledge of the underlying chromosomal abnormalities and associated clinical outcomes. Demonstrate the ability to analyze reciprocal translocations, like the Philadelphia chromosome in CML, explaining the consequences of the BCR-ABL gene fusion. This involves interpreting the molecular mechanisms and their role in unregulated cell division leading to cancer. Apply knowledge of meiotic outcomes in carriers of balanced reciprocal and Robertsonian translocations, predicting the potential gametes and the risk of producing offspring with chromosomal abnormalities. This involves interpreting the outcomes based on the type of translocation and understanding the implications for offspring development.	K S S	Fully online	Teams	Asynchronous Lecturing	Written exam	A
	3.2	**Translocation, Aneuploidy, Mosaics, and Chimera**	Explain reciprocal and Robertsonian translocations, detailing the stability of derivative chromosomes during mitosis and their impact on chromosomal structure. Describe origins of triploidy and tetraploidy, differentiating dispermy,	K K	Fully online	Teams	Asynchronous Lecturing	Written exam	A

			meiotic faults, and endomitosis. Analyze clinical manifestations, distinguishing diandric and digynic triploidy types.	S					
4	4.1	**Contrasting Mendelian and Non-Mendelian Inheritance**	Describe the general features of Mendelian patterns of single gene inheritance. Construct and interpret pedigrees using standard nomenclature. Analyze scenarios involving pleiotropy, epistasis, and polygenic inheritance.	K K/S S	Fully online	Teams	Synchronous Lecturing	Written exam	A
	4.2	**Autosomal Dominant Pedigrees & Diseases**	Identify features of autosomal dominant pedigrees and associated diseases. Contrast and construct pedigrees with autosomal dominant mode of inheritance	K S	Fully online	Teams	Asynchronous Lecturing	Written exam	A
5	5.1	**Autosomal Recessive Pedigrees & Diseases**	Identify features of autosomal recessive pedigrees and associated diseases. Contrast and construct pedigrees with autosomal recessive mode of inheritance	K S	Fully online	Teams	Asynchronous Lecturing	Written exam	A
	5.2	**X-Inactivation**	Explain X-inactivation in females.	K	Fully online	Teams	Asynchronous Lecturing	Written exam	A
6	6.1	**Sex-Linked Pedigrees & Diseases**	Analyze features of sex-linked pedigrees and associated diseases	S	Fully online	Teams	Synchronous Lecturing	Written exam	A
	6.2	**Phenotypic Expression**	Explain the concepts of penetrance and expressivity in the context of phenotypic expression. Differentiate between allelic and locus heterogeneity. Provide examples of genetic disorders demonstrating allelic heterogeneity, like cystic fibrosis, and locus heterogeneity, such as congenital deafness. Analyze scenarios where mutations at different loci result in similar phenotypes. Define and distinguish	K S	Fully online	Teams	Asynchronous Lecturing	Written exam	A

			between sex-limited and sex-influenced phenotypes. Demonstrate proficiency in recognizing the influence of environmental, social, cultural, and psychological factors on the manifestation of genetic traits, refining clinical decision-making processes.	K/S C					
7	7.1	**Imprinting, and Dynamic Variants**	Recognize non-Mendelian inheritance and identify disorders that deviate from Mendelian patterns, such as mitochondrial inheritance, unstable trinucleotide repeats, and imprinting. Determine the mechanisms and characteristics associated with non-Mendelian inheritance. Explain the concept of unstable trinucleotide repeats and their role in disorders with anticipation, such as Fragile X Mental Retardation syndrome, Huntington's disease, myotonic dystrophy, and others. Analyze the impact of trinucleotide repeat expansions on gene function and phenotype.	K K K S	Fully online	Teams	Asynchronous Lecturing	Written exam	A
	7.2	**Pedigree Practice**	Demonstrate the ability to construct accurate pedigrees using standard symbols and nomenclature. Recognize and analyze common inheritance patterns, including autosomal dominant, autosomal recessive, X-linked dominant, and X-linked recessive. Apply understanding of genetic diversity principles and laws of segregation and independent assortment to real-world clinical scenarios.	S S C	Fully online	Teams	Synchronous Lecturing	Written exam	A
8	**Midterm exam**								
9	9.1	**Risk Assessment I**	Understand population-level risk assessment for	K	Fully online	Teams	Asynchronous	Written exam	A

			genetic disorders. Explore the impact of familial relationships and carrier status on genetic risk. Constructing and interpreting Punnett squares for different genetic scenarios.	S S			Lecturing		
	9.2	**Risk Assessment II**	Determine the probability of specific genetic events, such as the likelihood of having all male or all female children in a family, the probability of inheriting a disease like Huntington's, or the chance of having children with a particular genetic condition.	S	Fully online	Teams	Asynchronous Lecturing	Written exam	A
10	10.1	**Risk Assessment Practice**	Apply Mendelian inheritance principles to predict the likelihood of specific genetic outcomes in families. Explore scenarios involving carriers of genetic disorders, such as cystic fibrosis, and calculate the probabilities of having affected, carrier, or unaffected children.	S S	Fully online	Teams	Synchronous Lecturing	Written exam	A
	10.2	**Human Genetic Variation I**	Understand the distinctions between mutation, polymorphism, and variant according to the ACMG 2015 guidelines. Explore the various categories of genetic variation, including genome mutations, chromosome mutations, and gene mutations, while assessing their frequencies and implications. Navigate genetic databases for genes and variants. Utilize online resources to access and interpret genetic information. Develop the ability to use approved reference sequence types (c., g., m., n., o., p., and r.) for reporting variants at	K K S S S	Fully online	Teams	Asynchronous Lecturing	Written exam	A

			different molecular levels.						
11	11.1	**Human Genetic Variation II**	Describe different types of genetic variation and delineate their potential impact on the protein and mRNA levels. Summarize the molecular events involved in splicing, including the role of snRNPs and spliceosome components. Demonstrate the ability to consider socio-cultural, psychological, and environmental factors when interpreting and applying genetic knowledge.	K K C	Fully online	Teams	Asynchronous Lecturing	Written exam	A
	11.2	**Clinical variation**	Learn about types (conductive, sensorineural, mixed) and severity levels of hearing loss, distinguishing between central auditory dysfunction and other types. Recognize and differentiate between syndromic and nonsyndromic hearing impairments. Describe genetic patterns and associating specific syndromes with hearing loss. Demonstrate the ability to integrate genetic knowledge into clinical practice by formulating tailored diagnostic approaches for diagnosis of hearing loss. Determine the genetic basis of Usher Syndrome, including its types and molecular components. Explore different approaches in conducting molecular genetic testing used in the diagnosis of Usher Syndrome type 1	K K K C K S	Fully online	Teams	Asynchronous Lecturing	Written exam	A

			(USH1). Apply genetic insights to clinical scenarios, illustrating the integration of genetic knowledge into the diagnosis of Usher Syndrome. Identify the genetic basis of Treacher Collins Syndrome (TCS) and the role of TCOF1, POLR1C, and POLR1D genes in its development. Analyze the phenotypic variability in TCS, including incomplete penetrance and the range of craniofacial anomalies	C K S					
12	12.1	**Population genetics**	Define and explain the key concepts of population genetics, including allele frequencies, gene flow, genetic drift, and natural selection. Analyze patterns of genetic variation within populations and understand the factors influencing genetic diversity. Apply the Hardy-Weinberg equilibrium to predict allele frequencies and understand the conditions necessary for equilibrium.	K S S	Fully online	Teams	Synchronous Lecturing	Written exam	A
	12.2	**Precision medicine**	Define precision medicine and understand its principles, emphasizing the focus on individual variability in genes, environment, and lifestyle. Understand the use of genomic data in precision medicine, including the interpretation of genetic variations and their implications for personalized treatment. Implement clinical and genomic data to formulate personalized treatment plans, considering genetic predispositions to diseases and drug responses.	K K S	Fully online	Teams	Asynchronous Lecturing	Written exam	A

13	13.1	**Biochemical Genetics I**	Define Inherited Metabolic Diseases (IMD) and articulate the biochemical processes involved in major food conversion and degradation for excretion.	K	Fully online	Teams	Asynchronous Lecturing	Written exam	A
	13.2	**Biochemical Genetics II**	Explain the organization of biochemical reactions into pathways, emphasizing their interrelation in physiological activities, with a focus on energy production and the urea cycle.	K	Fully online	Teams	Asynchronous Lecturing	Written exam	A
14	14.1	**Cancer Genetics I**	Describe the fundamental characteristics of cancer as a genetic disease. Explain the main classes of cancer, focusing on carcinomas, sarcomas, and hematopoietic/lymphoid neoplasms. Analyze the genomic basis of cancer development, including the concepts of driver and passenger gene variants. Differentiate between the random mutations observed in tumors and the specific mutations in driver genes crucial for cancer progression.	K K S S	Fully online	Teams	Synchronous Lecturing	Written exam	A
	14.2	**Cancer Genetics II**	Explain the Two-Hit Theory of Tumor Suppressor Gene Inactivation, emphasizing its application to retinoblastoma and other hereditary cancers. Understand the significance of both inherited and somatic mutations in the inactivation of tumor suppressor genes. Analyze the concept of loss of heterozygosity (LOH) in the context of retinoblastoma and	K K S	Fully online	Teams	Asynchronous Lecturing	Written exam	A

			hereditary cancer syndromes. Explore the various genetic, epigenetic, and genomic mechanisms that can lead to LOH and its role in the second hit hypothesis.	S					
** K: Knowledge, S: Skills, C: Competency									

25. Evaluation Methods:

Opportunities to demonstrate achievement of the ILOs are provided through the following assessment methods and requirements:

Evaluation Activity	Mark	Topic(s)	SLOs	Descriptors**	Period (Week)	Platform
Midterm exam	40	Genetic Diversity, Laws of Segregation, and Independent Assortment Karyotyping, Chromosome Structure, and Nomenclature Autosomal Chromosomes and Numerical Chromosomal Aberrations Sex Chromosomes, Structural Chromosomal Aberrations Translocation, Aneuploidy, Mosaics, and Chimera Contrasting Mendelian and Non-Mendelian Inheritance Autosomal Dominant Pedigrees & Diseases Autosomal Recessive Pedigrees & Diseases X-Inactivation Sex-Linked Pedigrees & Diseases Phenotypic Expression Imprinting, and Dynamic Variants Pedigree Practice	1.1/ 1.2/ 2.1/ 2.2/ 3.1/ 3.2/ 4.1/ 4.2/ 5.1/ 5.2/ 6.1/ 6.2/ 7.1/ 7.2	K S	8th week	Exam builder/online
Final exam	60	Risk Assessment I Risk Assessment II	9.1/ 9.2/ 10.1/ 10.2/ 11.1/ 11.2/ 12.1/ 12.2/ 13.1/ 13.2/ 14.1/ 14.2	K S	15th -16th week	Exam builder/online

		Risk Assessment Practice Human Genetic Variation I Human Genetic Variation II Clinical variation Population genetics Precision medicine Biochemical Genetics I Biochemical Genetics II Cancer Genetics I Cancer Genetics II				
** K: Knowledge, S: Skills, C: Competency						

*** According to the instructions for granting a bachelor's degree.**

****According to the principles of organizing semester work, tests, examinations, and grades for the bachelor's degree.**

Mid-term exam specifications table*

*(The tableS will be completed on separate forms by course coordinators prior to conduction of each exam according to Accreditation and Quality Assurance Centre procedures and forms).

No. of questions/ cognitive level						No. of questions per CLO	Total exam mark	Total no. of questions	CLO/ Weight	CLO no.
Create **%10**	Evaluate **%10**	analyse **%10**	Apply %20	Understand **%20**	Remember %30					
1	1	1	4	2	1	10	100	100	10%	1

Final exam specifications table

No. of questions/ cognitive level						No. of questions per CLO	Total exam mark	Total no. of questions	CLO Weight	CLO no.
Create **%10**	Evaluate **%10**	analyse **%10**	Apply %20	Understand **%20**	Remember %30					
										1
										2
										3
										4
										5

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26. Course Requirements:

- ✓ Internet connection
- ✓ Computer

27. Course Policies:

A- Attendance policies:

Attendance will be monitored by the course coordinator. Attendance policies will be announced at the beginning of the course.

B- Absences from exams and handing in assignments on time:

Will be managed according to the University of Jordan regulations. Refer to http://registration.ju.edu.jo/Documents/daleel.pdf

C- Health and safety procedures:

Faculty Members and students must always, conform to Health and Safety rules and procedures.

D- Honesty policy regarding cheating, plagiarism, misbehavior:

As a student in this course (and at this university) you are expected to maintain high degrees of professionalism, commitment to active learning and participation in this course and also integrity in your behavior in and out of the classroom. Students violate this policy would be subjected to disciplinary action according to University of Jordan disciplinary policies

E- Grading policy:

Grade-point average, Rules are preset by the Faculty and Department Councils

F- **Available university services that support achievement in the course:**

Availability of comfortable lecture halls, data show, internet service and E learning website https://elearning.ju.edu.jo/ .

28. References:

A- Required book(s), assigned reading and audio-visuals:

New Clinical Genetics, fourth edition Paperback – December 15, 2020
by Andrew Read (Author), Dian Donnai (Author)
ISBN-10 : 1911510703 ISBN-13: 978-1911510703

Name of the Instructor or the Course Coordinator: Dr. Zaid Aburubaiha	Signature: Aburubaiha	Date: 15.07.2025
Name of the Head of Quality Assurance Committee/ Department Dr Enas Al-Zayadneh	Signature:	Date: .....
Name of the Head of Quality Assurance Committee/ School or Center Professor Ayman Wahbeh	Signature:	Date: 16/7/2025
Name of the Dean or the Director Professor Ayman Wahbeh	Signature:	Date: 16/7/2025