Kingdom of Saudi Arabia
Taibah University – Madinah
College of Applied Medical Sciences
Department of Clinical Laboratory Sciences

A- COURSE TITLE, CODE, ACADEMIC YEAR:
Molecular & Medical Genetics (MLT 331) 1438-1439H

B- COURSE INFORMATION:

Course Code	Course Title	Credit Units			Study Level	Pre-requisites
		Total	Theory	Practical		
MLT 331	Molecular & Medical Genetics	2	2	0	5th level	NA

Course Coordinator	Extension	Email Address
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C- COURSE DESCRIPTION:

The course is introducing the basic concepts, terminology and principles in medical and molecular genetics. Different medical diseases caused by inherited defect in the genetic code are well illustrated. Mendel's laws of heredity governing mode of inheritance of medical genetic disease are explained and determined through constructing a family pedigree. The structure and function of molecular unit of hereditary from gene, chromosomes to genome are described and their role in transmission the medical genetic diseases from generation to generation are defined. Permanent changes in molecular genotype and their influence in developing mutant phenotype is clearly correlated and stated and different types of gene mutations and chromosomal aberrations and their effects in causing genetic disorder are illustrated. Several different medical and molecular genetic diagnostic techniques in current practice in clinical molecular genetics labs including cytogenetic, DNA sequencing, prenatal testing and screening to detect and identify different gene and chromosomal mutations causing genetic diseases are described.

D- COURSE OBJECTIVES:

1. To clearly distinguish between medical genetics and molecular genetics and their interrelations.
2. To introduce and define the molecular unit of hereditary and medical and molecular genetic related terms including genes, chromosomes, genomics and heredity.
3. To ascertain the role of defective molecular unit of hereditary in transmitting genetic disease from generation to the next.
4. To correlate between changes in molecular genotype and their influence in developing mutant phenotype .
5. To identify the molecular structure of genes, chromosomes and genome.
6. To recognize the role of Mendel's laws of heredity in controlling genetic disease mode of inheritance.
7. To describe cell division process and differentiate between steps in cell meiosis and mitosis .
8. To describe the structure and function of DNA and RNA molecules.
9. To outline and recognize the different stages of the three cellular molecular mechanisms; DNA replication, transcription and translation.
10. To explain the different modes of transmission of single gene disorder.
11. To differentiate between chromosomal structural and number aberrations.
12. To list different molecular genetic diagnostic techniques to detect chromosomal abnormalities.

13. To learn different cytogenetic and molecular cytogenetic techniques and their application in clinical diagnosis.
14. To record different types of mutations and their phenotypic effects on protein structure and function.
15. To name the different mutational bases of Sickle cell disease.
16. To define the different steps in developing cancer and their cancer causing genes.
17. To construct a family pedigree and interpret and decipher the mode of inheritance of disrober.
18. To evaluate the applications of different DNA sequencing methods in discovering different gene mutations causing genetic diseases.
19. To summarize the different medical and molecular genetic prenatal diagnostic techniques.

E- THEORY TOPICS:

Week	Theory Topic	Contact Hours
1	Introduction to Genetics	2
2	DNA & RNA	2
3	Genes & Chromosomes	2
4	The Genetic Code	2
5	Modes of Inheritance	2
6	Genetic Diseases: Chromosomal Abnormalities	2
7	Case Study I: Down Syndrome	2
8	Genetic Diseases: Mutations	2
9	Case Study II: Sickle Cell Disease	2
10	Cancer Genetics	2
11	Diagnostic Techniques: Pedigrees	2
12	Diagnostic Techniques: Prenatal Testing	2
13	Diagnostic Techniques: Cytogenetics & FISH	2
14	Diagnostic Techniques: Sequencing	2

G- ASSESSMENT TASKS:

#	Type of assessment task	Week	Total Grades
1	MCQs on scratch card	Weeks 1-14	20%
2	Midterm examination (written)	Week 7	20%
3	Assignment submission	Week 9	20%
4	Final written examination	Week 15-16	40%

H- LEARNING RESOURCES:

1- Required textbook:

- Turnpenny, Peter D. and Ellard, Sian. Emery's Elements of Medical Genetics. 14th ed. Philadelphia: Elsevier Churchill Livingstone, 2012.
- Nussbaum, Robert L., McInnes, Roderick R.,Willard, Huntington F. and Hamosh, Ada. Thompson & Thompson Genetics in Medicine. 8th ed. Philadelphia: Elsevier, 2016.

2- Essential references:

- Bennett, Robin L. , French, Kathryn Steinhaus, Resta , Robert G. and Doyle , Debra Lochner. Standardized Human Pedigree Nomenclature: Update and Assessment of the Recommendations of the National Society of Genetic Counselors. Journal of Genetic Counseling 17, (2008):424-433. DOI 10.1007/s10897-008-9169-9
- John R. Bradley, David Johnson, Barbara Pober. Lecture Notes: Medical Genetics, Wiley, 2006
- Journal of Medical Genetics: http://jmg.bmj.com/

Notes:

- Assignments topics and requirements shall be announced by the end of Week-1, the deadline for submission is 12pm Thursday of Week-9.
- Assignments and written assessment tasks must be verified against plagiarism, the maximum acceptable percentage is determined by the department (according to each level).
- MCQs on scratch card: 10 quizzes will be given during the semester. Each quiz will contain 5 MCQs and will be given at the end of the lecture.