



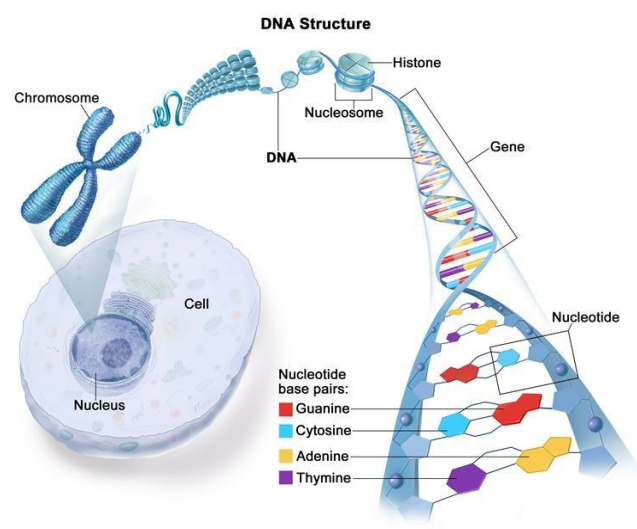
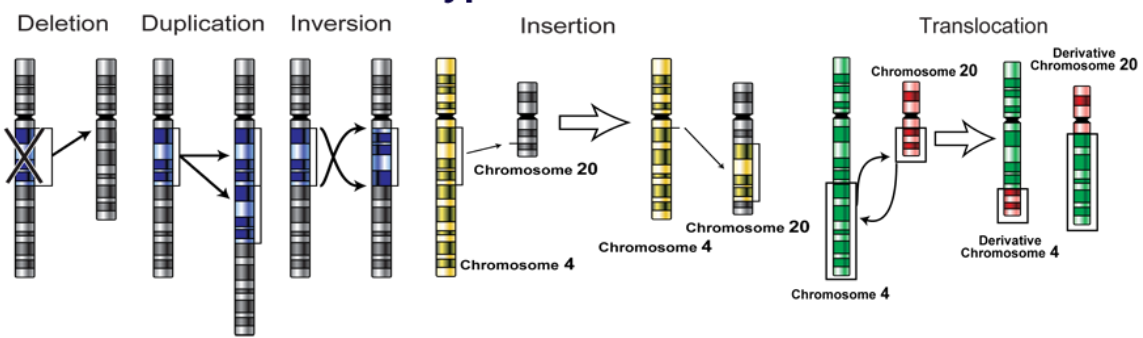
STUDY GUIDE

GENETICS-II MODULE

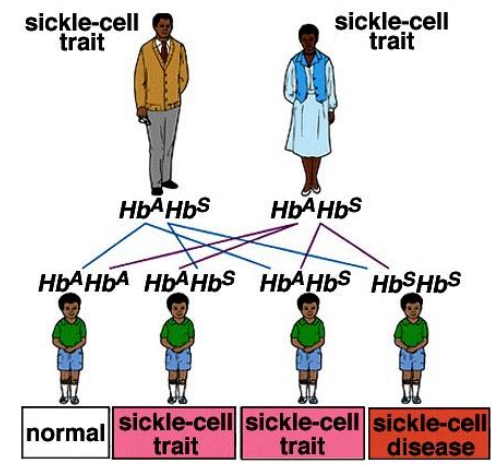
FOURTH YEAR MBBS

6th May - 11th May 2019
Duration: 1 week

Types of Mutations



Inheritance of sickle-cell disease



**LIAQUAT NATIONAL HOSPITAL
& MEDICAL COLLEGE**



STUDY GUIDE FOR GENETICS-II MODULE

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Module name: **Genetics-II**

Year: **Four**

Duration: **1 week (May 2019)**

Timetable hours: **Interactive Lectures, Case-Based Discussion (CBD), Clinical Rotations, Presentations, Demonstrations, Skills, Self-Study**

MODULE INTEGRATED COMMITTEE

MODULE COORDINATOR:	Dr. Shehnaz Sheikh (Biochemistry)
CO-COORDINATOR:	Dr. Sobia Ali (DHCE)

DEPARTMENTS' & RESOURCE PERSONS' FACILITATING LEARNING

BASIC HEALTH SCIENCES	CLINICAL AND ANCILLARY DEPARTMENTS
BIOCHEMISTRY <ul style="list-style-type: none"> Prof. Naheed Qadir Dr. Kashif Nisar 	MOLECULAR PATHOLOGY <ul style="list-style-type: none"> Dr. Muhammad Israr Nasir Dr. Sobia Rafiq
PATHOLOGY <ul style="list-style-type: none"> Prof. Naveen Faridi Dr. Rabia Ali 	
DEPARTMENT OF HEALTH PROFESSIONS EDUCATION	
<ul style="list-style-type: none"> Professor Nighat Huda Dr. M. Suleman Sadiq 	<ul style="list-style-type: none"> Dr. Sobia Ali Dr. Mehnaz Umair Dr. Afifa Tabassum
LNH&MC MANAGEMENT	
<ul style="list-style-type: none"> Professor Karimullah Makki, Principal, LNH&MC Dr. Shaheena Akbani, Director A.A & R.T LNH&MC 	
STUDY GUIDE COMPILED BY:	
<ul style="list-style-type: none"> Department of Health Professions Education 	

INTRODUCTION

WHAT IS A STUDY GUIDE?

It is an aid to:

- Inform students how student learning program of the module has been organized
- Help students organize and manage their studies throughout the module
- Guide students on assessment methods, rules and regulations

THE STUDY GUIDE:

- Communicates information on organization and management of the module.
This will help the student to contact the right person in case of any difficulty.
- Defines the objectives which are expected to be achieved at the end of the module.
- Identifies the learning strategies such as lectures, small group teachings, clinical skills, demonstration, tutorial and case based learning that will be implemented to achieve the module objectives.
- Provides a list of learning resources such as books, computer assisted learning programs, web- links, journals, for students to consult in order to maximize their learning.
- Highlights information on the contribution of continuous on the student's overall performance.
- Includes information on the assessment methods that will be held to determine every student's achievement of objectives.
- Focuses on information pertaining to examination policy, rules and regulations.

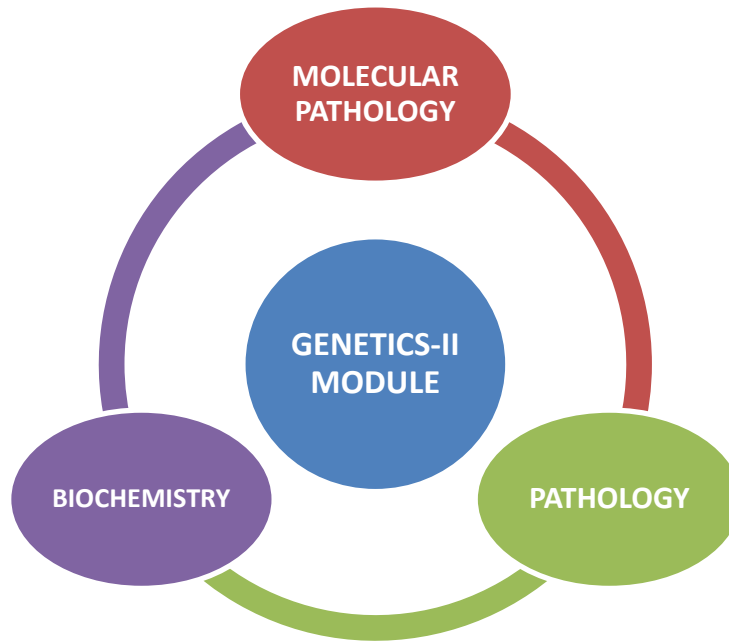
CURRICULUM FRAMEWORK

Students will experience *integrated curriculum* similar to previous modules.

INTEGRATED CURRICULUM comprises system-based modules such as Eye/ENT, dermatology, genetics, rehabilitation and neurosciences-II & psychiatry modules which link basic science knowledge to clinical problems. Integrated teaching means that subjects are presented as a meaningful whole. Students will be able to have better understanding of basic sciences when they repeatedly learn in relation to clinical examples.

LEARNING EXPERIENCES: Case based integrated discussions, Task oriented learning followed by task presentation, skills acquisition in skills lab, computer-based assignments, learning experiences in clinics, wards.

INTEGRATING DISCIPLINES OF GENETICS-II MODULE

**LEARNING METHODOLOGIES**

The following teaching / learning methods are used to promote better understanding:

- Interactive Lectures
- Small Group Discussion
- Case- Based Discussion (CBD)
- Clinical Experiences
 - Clinical Rotations
- Skills session

INTERACTIVE LECTURES: In large group, the lecturer introduces a topic or common clinical conditions and explains the underlying phenomena through questions, pictures, videos of patients' interviews, exercises, etc. Students are actively involved in the learning process.

SMALL GROUP SESSION: This format helps students to clarify concepts, acquire skills or desired attitudes. Sessions are structured with the help of specific exercises such as patient case, interviews or discussion topics. Students exchange opinions and apply knowledge gained from lectures, tutorials and self study. The facilitator role is to ask probing questions, summarize, or rephrase to help clarify concepts.

CASE-BASED DISCUSSION (CBD): A small group discussion format where learning is focused around a series of questions based on a clinical scenario. Students' discuss and answer the questions applying relevant knowledge gained previously in clinical and basic health sciences during the module and construct new knowledge. The CBD will be provided by the concern department.

CLINICAL LEARNING EXPERIENCES: In small groups, students observe patients with signs and symptoms in hospital wards, clinics and outreach centers. This helps students to relate knowledge of basic and clinical sciences of the module and prepare for future practice.

- **CLINICAL ROTATIONS:** In small groups, students rotate in different wards like Medicine, Pediatrics, Surgery, Obs & Gyne, ENT, Eye, Family Medicine clinics, outreach centers & Community Medicine experiences. Here students observe patients, take histories and perform supervised clinical examinations in outpatient and inpatient settings. They also get an opportunity to observe medical personnel working as a team. These rotations help students relate basic medical and clinical knowledge in diverse clinical areas.

SKILLS SESSION: Skills relevant to respective module are observed and practiced where applicable in skills laboratory.

SELF-DIRECTED STUDY: Students' assume responsibilities of their own learning through individual study, sharing and discussing with peers, seeking information from Learning Resource Center, teachers and resource persons within and outside the college. Students can utilize the time within the college scheduled hours of self-study.

MODULE 3: GENETICS II

INTRODUCTION

A genetic disorder is a disease that is caused by a change, or mutation, in an individual's DNA sequence. These mutations can be due to an error in DNA replication or due to environmental factors, such as cigarette smoke and exposure to radiation, which cause changes in the DNA sequence.

Internationally, 3-5% of all births result in congenital malformations; 20-30% of all infant deaths are due to genetic disorders; 30-50% of post-neonatal deaths are due to congenital malformations. 11.1% of pediatric hospital admissions are for children with genetic disorders and 18.5% are children with other congenital malformations; 12% of adult hospital admissions are for genetic causes. 50% of mental retardation, a common global occurrence, has a genetic basis. Cancers are one of the most dreaded conditions; 15% of all cancers have an inherited susceptibility whereas 10% of chronic diseases (heart, diabetes arthritis) which occur in the adult population have a significant genetic component.

Pakistan has a high frequency of marriages among close cousins, i.e. consanguineous marriages. This percentage is 62.70 — by far the highest among countries in the 'consanguinity belt' which includes countries of the Middle East and the Sub continent. It is estimated that about 29 million people out of Pakistan's 200 million population suffer from genetic defects attributable to close or first-cousin marriages.

Hence, it becomes imperative for medical graduates in Pakistan to understand how such conditions occur and how they can be managed and prevented. Since genetics, and the pathophysiology of genetic disorders, is a complex process, this topic was initially dealt with in the first spiral of the curriculum at a simpler level and is now being re-visited at a more advanced level. In the first year, normal genetic processes were described so that learners get a clear understanding of how chromosomes function.

In this 2nd spiral of Genetics, students will learn about the process of mutations, single gene defects and techniques used to diagnose such disorders. You may experience various genetic disorders during the clinical rotation and clerkship.

Reference: JSMU Study guide dated May 10th, 2018



COURSE OBJECTIVES AND STRATEGIES

By the end of Genetics-II module students should be able to:

OBJECTIVES	TEACHING STRATEGY
BIOCHEMISTRY	
<ul style="list-style-type: none"> Describe the process of DNA Replication and repair 	Interactive Lectures/Small Group Discussion
<ul style="list-style-type: none"> Explain the mechanism of Transcription and Post Transcriptional Modification 	
<ul style="list-style-type: none"> Discuss the process of Translation and Post Translational Modification 	
MOLECULAR PATHOLOGY	
<ul style="list-style-type: none"> Discuss the basic concepts of genetics including DNA and RNA structure, Mendel's Laws of inheritance and Pedigree Chart 	Interactive Lectures/Small Group Discussion
<p>Mutations</p> <ul style="list-style-type: none"> List the different types of mutations in the coding and non-coding regions of genes Distinguish between the different types of mutations in the coding and non-coding regions of genes that result in phenotypic change Differentiate between spontaneous and induced mutations Explain how a point mutations or frame shift mutation in a gene may alter the activity of the protein it encodes 	Interactive Lectures
<p>Single gene disorders</p> <ul style="list-style-type: none"> Define single gene disorders List different types of single gene disorders List characteristics of single gene which gives variation in expression of diseases Describe genetic changes which occur in these disorders 	
<p>Genetic techniques</p> <ul style="list-style-type: none"> Discuss the basic principles of recombinant genetic techniques and their applications in the detection of genetic diseases which includes PCR, FISH, RFLP, BLOTTING 	Small Group Discussion/Interactive Lectures

Prenatal Diagnosis <ul style="list-style-type: none"> Define prenatal diagnosis List different techniques used for prenatal diagnosis List the advantages and disadvantages of prenatal diagnosis Justify indications for and utility of prenatal diagnostic tests 	Interactive Lectures
Gene Therapy & Counseling <ul style="list-style-type: none"> Describe how gene therapy works Explain the tools and techniques used to deliver gene therapies, to the disease candidates for gene therapy and associated risks and challenges 	
PATHOLOGY	
Pathophysiology of Inheritance <ul style="list-style-type: none"> Explain the pathophysiology of classical and non-classical mode of inheritance of genetic diseases 	Interactive Lecture
<ul style="list-style-type: none"> Discuss the clinical features of important genetic disorders which includes Down's syndrome, Turner's syndrome, Cystic Fibrosis, Sickle Cell Anemia, Thalassemia 	Small Group Discussion

Apart from attending daily scheduled sessions, students too should engage in self-study to ensure that all the objectives are covered



LEARNING RESOURCES

SUBJECT	RESOURCES
BIOCHEMISTRY	<u>TEXTBOOKS</u> <ol style="list-style-type: none"> 1. Harper's Illustrated Biochemistry 2. Lehninger Principle of Biochemistry 3. Biochemistry by Devlin
PATHOLOGY	<u>TEXT BOOKS</u> <ol style="list-style-type: none"> 1. Robbins & Cotran, Pathologic Basis of Disease, 9th edition. 2. Rapid Review Pathology, 4th edition by Edward F. Goljan MD <u>WEBSITES:</u> <ol style="list-style-type: none"> 1. http://library.med.utah.edu/WebPath/webpath.html 2. http://www.pathologyatlas.ro/
MOLECULAR PATHOLOGY	<u>REFERENCE BOOK:</u> <ol style="list-style-type: none"> 1. Thompson & Thompson Genetics in Medicine 8th Edition

ADDITIONAL LEARNING RESOURCES

<u>Hands-on Activities/ Practical</u>	Students will be involved in Practical sessions and hands-on activities that link with the Genetics-II Module to enhance learning.
<u>Museum</u>	Models available in the museum are a rich learning resource for quick review of anatomy and related educational activities
<u>Skills Lab</u>	Skills acquisition in a simulated environment in the skills lab involving experiential learning will ensure patient safety and will also help to build confidence in approaching the patients
<u>Videos/Podcasts</u>	Videos and podcasts will familiarize the student with the procedures and protocol which they can watch and listen to at any time and wherever they are, as part of task oriented learning
<u>Internet Resources</u>	Students will use easily accessible internet resources with added time flexibility to enrich and update their knowledge and its application

ASSESSMENT METHODS:**Theory:**

- **Best Choice Questions (BCQs)** also known as MCQs (Multiple Choice Questions) are used to assess objectives covered in each module.
 - A BCQ has a statement or clinical scenario followed by four options (likely answer).
 - Students after reading the statement/scenario select ONE, the most appropriate response from the given list of options.
 - **Correct answer carries one mark, and incorrect 'zero mark'. There is no negative marking.**
 - Students mark their responses on specified computer-based/OMR sheet designed for LNHMC.

OSPE/OSCE: Objective Structured Practical/Clinical Examination:

- Each student will be assessed on the same content and have same time to complete the task.
- Comprise of 12-25 stations.
- Each station may assess a variety of clinical tasks, these tasks may include history taking, physical examination, skills and application of skills and knowledge
- Stations are observed, unobserved, interactive and rest stations.
- Observed and interactive stations will be assessed by internal or external examiners.
- Unobserved will be static stations in which there may be an X-ray, Labs reports, pictures, clinical scenarios with related questions for students to answer.
- Rest station is a station where there is no task given and in this time student can organize his/her thoughts.

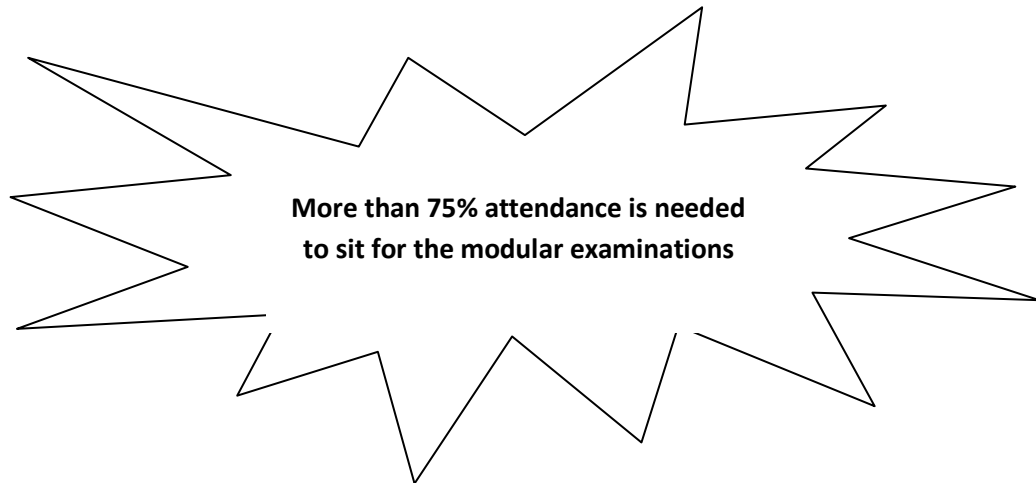
Internal Evaluation

- Students will be assessed to determine achievement of module objectives through the following:
 - **Module Examination:** will be scheduled on completion of each module. The method of examination comprises theory exam which includes BCQs and OSPE (Objective Structured Practical Examination).
 - **Graded Assessment of students by Individual Department:** Quiz, viva, practical, assignment, small group activities such as CBL, TBL, TOL, online assessment, ward activities, examination, and log book.
- Marks of both modular examination and graded assessment will constitute 20% weightage.
- As per JSMU policy, this 20% will be added by JSMU to Final Theory Examination.

Example : Number of Marks allocated for Final Theory and Internal Evaluation			
Semester	Final Examination Theory Marks	Internal Evaluation (Class test + Assignments + Modular Exam)	Total (Theory)
	80%	20%	100%

Formative Assessment

- Individual department may hold quiz or short answer questions to help students assess their own learning. The marks obtained are not included in the internal evaluation



MODULAR EXAMINATION RULES & REGULATIONS (LNH&MC)

- Student must report to examination hall/venue, 30 minutes before the exam.
- **Exam will begin sharp at the given time.**
- No student will be allowed to enter the examination hall after 15 minutes of scheduled examination time.
- Students must sit according to their roll numbers mentioned on the seats.
- **Cell phones are strictly not allowed in examination hall.**
- If any student is found with cell phone in any mode (silent, switched off or on) he/she will be not be allowed to continue their exam.
- No students will be allowed to sit in exam without University Admit Card, LNMC College ID Card and Lab Coat
- Student must bring the following stationary items for the exam: Pen, Pencil, Eraser, and Sharpener.
- Indiscipline in the exam hall/venue is not acceptable. Students must not possess any written material or communicate with their fellow students.

JSMU Grading System

- It will be based on GPA – 4 system

Marks obtained in Percentage range	Numerical Grade	Alphabetical Grade
80-100	4.0	A+
75-79	4.0	A
70-74	3.7	A-
67-69	3.3	B+
63-66	3.0	B
60-62	2.7	B-
56-59	2.3	C+
50-55	2.0	C
<50 Un-grade-able	0	U

- A candidate obtaining GPA less than 2.00 (50%) is declared un-graded (fail).
- Cumulative transcript is issued at the end of clearance of **all** modules.

SCHEDULE:

WEEKS	4 th Year	MONTH
WEEK 1	OPHTHALMOLOGY/ ENT	1 st April 2019
WEEK 2		
WEEK 3		20 th April 2019
	MODULAR EXAM	22 nd April 2019
WEEK 1	REHABILITATION	23 rd April 2019
WEEK 2		4 th May 2019
WEEK 1	GENETICS	6 th May –11 th May 2019
WEEK 1	DERMATOLOGY	13 th May 2019*
WEEK 2		24 th May 2019*
	DERMATOLOGY, GENETICS & REHABILITATION MODULAR EXAM**	29 th May & 30 th May 2019*
WEEK 1	NEUROSCIENCES-II & PSYCHIATRY	June 2019*
WEEK 2		
WEEK 3		
WEEK 4		
WEEK 5		
WEEK 6		
WEEK 7		
WEEK 8		Aug 2019*
	MODULAR EXAM	Aug 2019*

*Final dates will be announced later

** There will be combined module exam for Dermatology, Genetics and Rehabilitation modules