

# GENETICS MODULE

## STUDY GUIDE

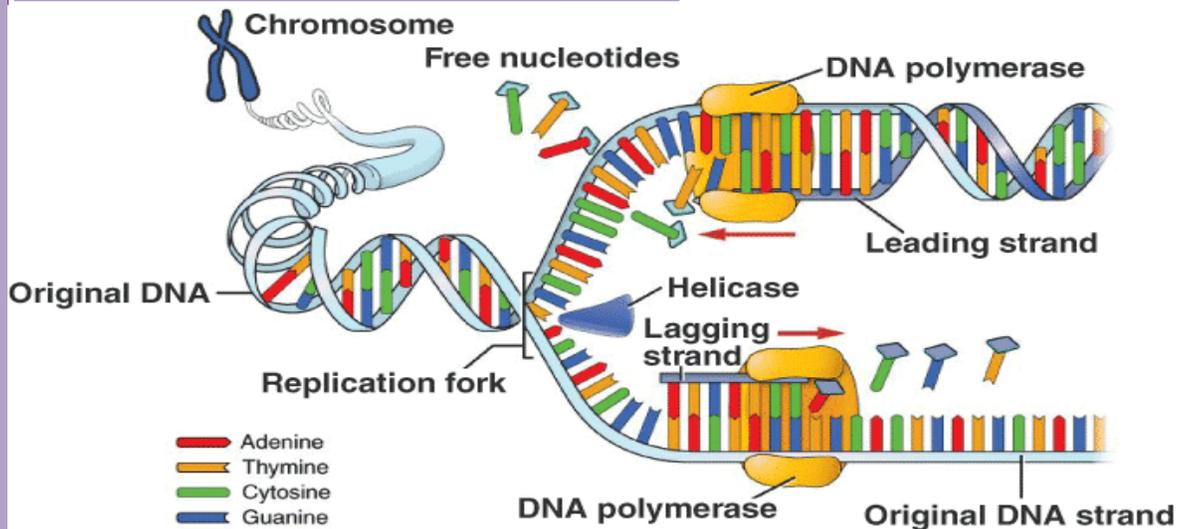
FOURTH YEAR MBBS

07<sup>TH</sup> JUNE- 19<sup>TH</sup> JUNE 2021

DURATION: 2 WEEKS



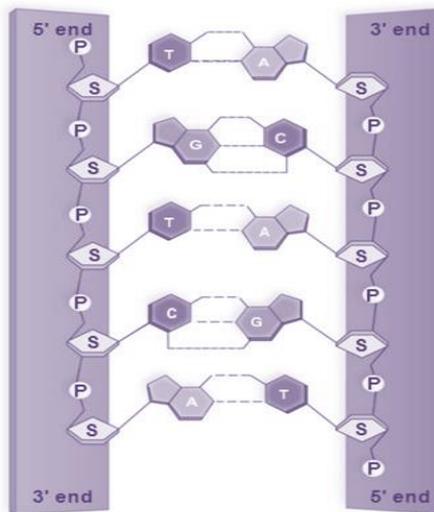
### DNA REPLICATION



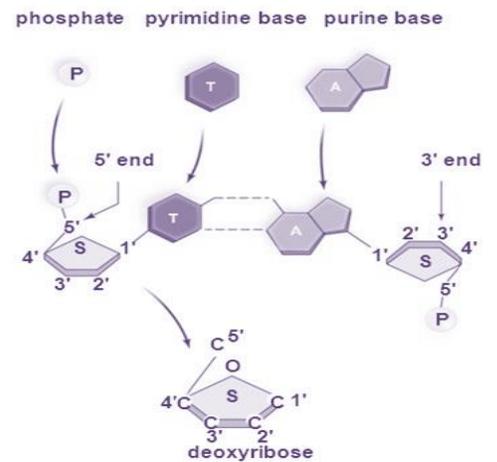
### OVERVIEW OF DNA STRUCTURE



a. Double helix



b. Ladder structure



c. One pair of bases



**STUDY GUIDE FOR GENETICS MODULE**

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Module name: **Genetics**Year: **Four**Duration: **2 weeks (June 2021)**

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

### MODULE INTEGRATED COMMITTEE

<b>MODULE COORDINATOR:</b>	<ul style="list-style-type: none"> <li>• Prof. Kashif Nisar (<b>Biochemistry</b>)</li> </ul>
<b>CO-COORDINATOR:</b>	<ul style="list-style-type: none"> <li>• Dr. S. Zulfiqar Ali Naqvi (<b>Molecular Pathology</b>)</li> <li>• Dr. M. Suleman Sadiq (<b>DHPE</b>)</li> </ul>

### DEPARTMENTS' & RESOURCE PERSONS' FACILITATING LEARNING

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

## **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.

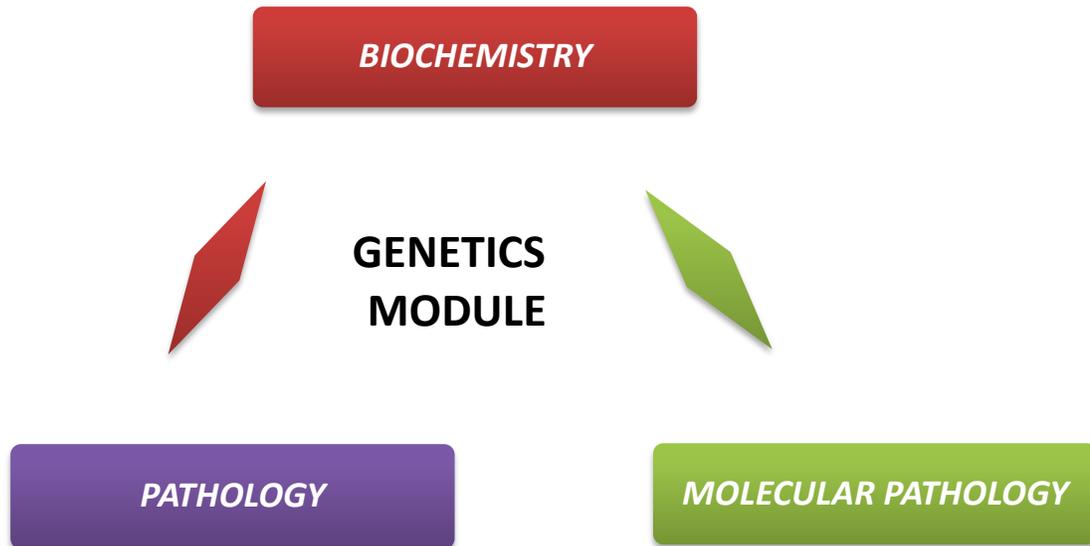
### **CURRICULUM FRAMEWORK**

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 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

### 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 10<sup>th</sup>, 2018



**COURSE OBJECTIVES AND STRATEGIES**

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

**BIOCHEMISRY**

OBJECTIVES	TEACHING STRATEGIES
<b>DNA Replication and repair</b>	Interactive Lectures & Small Group Discussion
1. Describe the process of DNA Replication and repair	
<b>Transcription and post transcription modifications</b>	
2. Explain the mechanism of Transcription and Post Transcriptional Modification	
<b>Post-Translational modifications</b>	
3. Discuss the process of Translation and Post Translational Modification	

**PATHOLOGY**

OBJECTIVES	TEACHING STRATEGIES
<b>Pathophysiology of Inheritance</b>	
1. Explain the pathophysiology of classical and non-classical mode of inheritance of genetic diseases	Interactive Lectures
2. 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

**MOLECULAR PATHOLOGY**

OBJECTIVES	TEACHING STRATEGIES
<b>Introduction to genetics</b>	
1. Discuss the basic concepts of genetics including DNA and RNA structure, Mendel's Laws of inheritance and Pedigree Chart	Interactive Lectures
<b>Mutations</b>	
2. List the different types of mutations in the coding and non-coding regions of genes	Interactive Lectures
3. Distinguish between the different types of mutations in the coding and non-coding regions of genes that result in phenotypic change	
4. Differentiate between spontaneous and induced mutations	
5. Explain how a point mutations or frameshift mutation in a gene may alter the activity of the protein it encodes	
<b>Single gene disorders</b>	
6. Define single gene disorders	Interactive Lectures
7. List different types of single gene disorders	
8. List characteristics of single gene which gives variation in expression of diseases	
9. Describe genetic changes which occur in these disorders	
<b>Genetic techniques</b>	
10. Discuss the basic principles of recombinant genetic techniques and their applications in the detection of genetic diseases which includes PCR, FISH, RFLP & BLOTTING	Interactive Lectures & Small Group Discussion
<b>Prenatal Diagnosis</b>	
11. Define prenatal diagnosis	Interactive Lectures
12. List different techniques used for prenatal diagnosis	
13. List the advantages and disadvantages of prenatal diagnosis	
14. Justify indications for utility of prenatal diagnostic tests	
<b>Gene Therapy &amp; Counseling</b>	
15. Describe how gene therapy works	Interactive Lectures
16. Explain the tools and techniques used to deliver gene therapies, disease candidates for gene therapy and associated risks and challenges	
17. Discuss & Interpretation of Karyogram	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**

<b>SUBJECT</b>	<b>RESOURCES</b>
<b>BIOCHEMISTRY</b>	<b><u>TEXTBOOKS</u></b> 1. Harper's Illustrated Biochemistry 2. Lehninger Principle of Biochemistry 3. Biochemistry by Devlin
<b>PATHOLOGY</b>	<b><u>TEXT BOOKS</u></b> 1. Robbins & Cotran, Pathologic Basis of Disease, 9th edition. 2. Rapid Review Pathology, 4th edition by Edward F. Goljan MD  <b><u>WEBSITES:</u></b> 1. <a href="http://library.med.utah.edu/WebPath/webpath.html">http://library.med.utah.edu/WebPath/webpath.html</a> 2. <a href="http://www.pathologyatlas.ro/">http://www.pathologyatlas.ro/</a>
<b>MOLECULAR PATHOLOGY</b>	<b><u>REFERENCE BOOK:</u></b> 1. Thompson & Thompson Genetics in Medicine 8th Edition

**ADDITIONAL LEARNING RESOURCES**

<b><u>Hands-on Activities/ Practical</u></b>	Students will be involved in Practical sessions and hands-on activities that link with the Genetics Module to enhance learning.
<b><u>Museum</u></b>	Models available in the museum are a rich learning resource for quick review of anatomy and related educational activities
<b><u>Skills Lab</u></b>	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
<b><u>Videos/Podcasts</u></b>	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
<b><u>Internet Resources</u></b>	Students will use easily accessible internet resources with added time flexibility to enrich and update their knowledge and its application

**ASSESSMENT METHODS:**

- **Best Choice Questions(BCQs)** also known as MCQs (Multiple Choice Questions)
- **Objective Structured Practical/Clinical Examination (OSPE or OSCE)**

**BCQs:**

- A BCQ has a statement or clinical scenario of four options (likely answers).
- **Correct answer carries one mark, and incorrect 'zero mark'. There is NO negative marking.**
- Students mark their responses on specified computer-based sheet designed for LNHMC.

**OSCE:**

- All students rotate through the same series of stations in the same allocated time.
- At each station, a brief written statement includes the task. Student completes the given task at one given station in a specified time.
- Stations are observed, unobserved, interactive or rest stations.
- In unobserved stations, flowcharts, models, slide identification, lab reports, case scenarios may be used to cover knowledge component of the content.
- Observed station: Performance of skills /procedures is observed by assessor
- Interactive: Examiner/s ask questions related to the task within the time allocated.
- In Rest station, students in the given time not given any specific task but wait to move to the following station.

**Internal Evaluation**

- Students will be assessed comprehensively through multiple methods.
- 20% marks of internal evaluation will be added to JSMU final exam. That 20% may include class tests, assignment, practicals and the internal exam which will all have specific marks allocation.

**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

**For JSMU Examination Policy, please consult JSMU website!**

**More than 75% attendance is needed to sit for the internal and final examinations**

**LNH&MC EXAMINATION RULES & REGULATIONS**

- 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.

**SCHEDULE:**

WEEKS	4 <sup>th</sup> Year	MONTH
WEEK 1	ORTHOPEDICS & REHABILITATION MODULE	29 <sup>th</sup> March 2021
WEEK 2		
WEEK 3		
WEEK 4		
WEEK 5		
WEEK 6		
WEEK 7		
WEEK 8		
WEEK 1	DERMATOLOGY MODULE	24 <sup>th</sup> May 2021
WEEK 2		
WEEK 1	GENETICS MODULE	5 <sup>th</sup> June 2021
WEEK 2		7 <sup>th</sup> June 2021
WEEK 1		
WEEK 2		19 <sup>th</sup> June 2021
<b>Mid Term Examination*</b>		

\*Final dates will be announced later