

# AJK Medical College, Muzaffarabad

Genetics, Neoplasia & Pediatric Diseases  
(GN&PD-0204)



**Pre-requisite modules:** Foundation, CPGAP, IHRI.

**Duration:** 2 Weeks

**Starting on:**

**DEPARTMENT OF MEDICAL EDUCATION**

## MODULE TEAM

Dr. Sarosh Majid Salaria	(Module Planner)
Dr. Muhammad Arif	(Module Coordinator)
Dr. Javed Rathore	(Medicine )
Dr. Ziyad Afzal Kayani	(DME)
Dr. Shaukat Ahmad Dar	(Radiology)
Dr. Khurshid Ahmad Lone	(Oncology)
Dr. Naheem Ahmed	(Pediatrics)
Dr. Naheed Akhter	(Surgery)

## CAUTION

**Eighty percent (80%) is mandatory attendance to appear in Modular/ Professional/University Examination as per PMDC regulations!**



## INTRODUCTION

This module combines two essential components of modern medicine! The art and science of genetics blasted only about 60 years back and then travelled to skies with missile speed! With sudden and miraculous discovery of "go" and "stop" whistles we for the first time ever were able to direct cells into cell cycle and to stop them just the right moment to take their snap with smile! Discoveries of Barr bodies and then Lyon's hypothesis made significant inroads! Postulation and construction of DNA molecule model was a leap forward! Since then the most fascinating journey of space of genetics goes on. I always regard genetics the language of life! For language you need alphabets which form words and words then forms meaningful sentences! It of course needs a medium to write upon! For spoken speech it's air and for written speech it's paper, computer discs etc. For genetic language it's DNA and RNA! Our language of life not only make our cells to communicate with other cells but also to transfer to the new generation! This magnificent communication is with superb marvellous articulation and regulation. Needless to say that the unity of this language of life in all earthly creatures is an open testimony to the oneness of the creator! Although we not be able to talk to each other but all cells whether human or virus understand each other very well.

Disorder of chromosomes produce genetic disorders on one hand and neoplastic disorders on the other hand! Both genetic and neoplastic disorders constitute significant percentage of patients of variable age groups. Additionally almost all "non genetic" disorders have some genetic component! It's obvious that the future doctors must have deep understanding of genetics and neoplastic process! In this module our faculty will bring to you various aspects from basic biochemistry to pathogenesis, therapeutics and diagnostic modalities at basic and clinical levels.

### Organization of Module

Genetics & Neoplasia module consists of six themes, and two PBL search based on a real life situation. Each theme has its explicit learning objectives. In order to enhance the quality of integration of basic sciences concepts with clinical sciences and to facilitate problem solving skills, this module has been designed and will be delivered by a committed integrated team comprising of Pathologists, Pharmacologists, Forensic experts, Radiologists, Physicians and Surgeons.

This module has been structured in a way that you will attain your objectives by pursuing different clinical themes while following a presented schedule/time table. Based on these themes will be clinical cases; that you will discuss in different sessions (SGD, PBL, SDL, LGIS, Skill Lab) during the program. The time table/ schedule with clinical themes are included in this study guide.

### Teaching Strategy

The content of this module will be delivered by a combination of different teaching strategies. These include large group interactive sessions (LGIS), small group interactive discussions (SGID), demonstrations in pathology laboratories, practicals and clinical skill sessions at skill lab/clinical sessions. In addition there will be a group project which will be assessed at the end of the block.

### Content Delivery

Entire curriculum will be delivered by clinical case scenarios each covering a theme. Read cases and the objectives of the theme which you are supposed to encounter next day, understand and explain the case to yourself and read the relevant information. Following learning/teaching strategies will be employed to discuss the cases:

#### Small Group Discussion (SGD):

Main bulk of the course content will be delivered in small group sessions. Each theme has an associated case. The case will be the core around which learning will take place. Depending on the case you might be required to deduce objectives and learning issues or only learning issues. Every group will have a facilitator assigned to it. The facilitator will be there to keep you on track, giving you maximum liberty to discuss and achieve the objectives as a group. Small groups in some cases may be followed by a wrap up session. Rest of the information will be there in the schedule.

#### Large group Interactive Sessions (LGIS):

Large group instruction will be employed at times sparingly. Attend large group sessions with the following focus

- Identify important points
- Ask questions of points not well understood in the text
- Measure your learning comprehension

## Hands-on Activities/ Practical:

Practical activities, linked with the case, will take place.

## Skill Lab/ Laboratory:

Attend your scheduled skill lab/ laboratory sessions and take advantage of free time for study. Use your labs to correlate text with actual specimens in lab practice.

## Assessment:

In this module, you will have formative and summative assessment. This will give you an idea about the format of the examination that you will go through at the end of the year. This will be followed by feedback on your performance in the exam. Marks obtained in the module examination will contribute 30% (internal assessment) towards end of year Professional University Examination. **There is no re-sit exam for module written assessment and block IPE** under any circumstances. If you miss them, your internal assessment will be recorded as zero. No excuse of any kind is permissible for absence in module or IPE assessment.



## RECOMMENDED LIST OF ICONS



**Introduction to case**



**For objectives**



**Critical questions**



**Assessment**



**Laboratory sessions**



**Resource material**

**Keywords**

## Table of Specifications

THEME	WEIGHT%
Essence of being—a miracle	10%
Corrupted Files	20%
Special child	25%
Lichen –Growth	30%
Human Acquiesce—to concur	15%

## LEARNING OBJECTIVES

### ESSENCE OF BEING—A MIRACLE (THEME-1)

At the end of the module the students will *In Sha Allah* be able to

1. Define the following terminologies:

RNA	nonsense mutation	pedigree
DNA	frame shift mutation	para bound
Gene	penetrance	non disjunction
Nitrogen bases	genotype	translocation
Codon	Phenotype	balanced translocation
Gene Translation	Autosomes	reciprocal translocation
Transcription	Sex chromosomes	isochromosome
Post translational modifications	Lyon's hypothesis	ring chromosome
Locus	Barr body, sex chromatin	reverse transcription
Allele	Karyotype	gene heterogeneity
Mutation	chorionic villus sampling (CVS)	pleomorphism
Point mutation	amniotic fluid sampling	expressivity
		Mosaicism

2. Draw and label DNA and a chromosome
3. Enumerate different types and groups of chromosomes Explain gene- gene interaction and gene environment interaction
4. Define and explain "Principles" of autosomal dominant, autosomal recessive, X linked recessive disorders

## CORRUPTED FILES (THEME 2)

1. Categorize genetic disorders
2. Classify chromosomal numerical and structural disorders
3. Define and classify Mendelian and Multifactorial disorders
4. Define mitochondrial genes and enlist important mitochondrial disorders and their characteristics.
5. Discuss molecular diagnosis of genetic disorders.
6. Define polymorphic markers and molecular diagnosis.

## SPECIAL CHILD (THEME 3)

**The students will *In Sha Allah* be able to give;**

1. Definition, clinical features, types and diagnosis of following genetic disorders:
  - a. Down Syndrome
  - b. Turner
  - c. Klinefelter
  - d. Thalassemia
  - e. Hemophilia
  - f. Glycogen storage
  - g. Gaucher's disease
  - h. Neiman Pick
  - i. Tay-Sach's disease
  - j. Sickle cell disease
  - k. Trisomy 18
  - l. Trisomy 13
  - m. Familial Hypercholesterolemia
  - n. Familial Adenomatous polyposis
  - o. Phakomatosis
2. Genetic counseling
3. List drug targets in normal hematopoietic system.
4. List various iron preparations. Describe their pharmacokinetic properties.
5. Discuss clinical uses and adverse effects of various iron preparations.
6. Describe complications and treatment of iron overload.
7. Describe precautions for the use of different iron preparations.
8. Enumerate various iron chelators with their important characteristics.
9. List various folic acid and Vit. B<sub>12</sub> preparations and describe their pharmacokinetic and Pharmacodynamic properties.
10. Discuss therapeutic uses and adverse effects of folic acid & vitamin B 12 preparations.
11. Enumerate different hematopoietic growth factors.
12. Describe the pharmacokinetics and pharmacodynamics of erythropoietin.
13. Discuss clinical pharmacology and toxicity of erythropoietin.
14. Describe pharmacokinetics and pharmacodynamics of myeloid growth factors and megakaryocyte growth factors.
15. Explain clinical pharmacology and toxicity of myeloid growth factors and megakaryocyte growth factors.
16. Explain Pharmacogenetics, Pharmacogenomics & Gene therapy

## LICHEN –GROWTH (THEME 4)

**At the end of this theme the students will *In Sha Allah* be able to**

1. Define Protooncogene and oncogene
2. List common oncogenes and their associated neoplasms
3. Define tumor suppressor gene and enlist important tumor suppressor genes
4. Explain the process of carcinogenesis ,tumor growth and factors affecting it
5. Explain host tumor relationship the Describe various disturbances of growth in terms of classification, causes, natural history, and significance paying particular attention to malignant neoplasms.
6. List the defining features and one typical example for each of the following:

Neoplasm	Haematoma	Carcinoma	Polyp
Malignant neoplasm	Hetrotopia	Sarcoma	Scirrhou
Benign neoplasm	Ectopia	Mixed tumor	Desmoplasia
Metaplasia	Differentiation	Teratoma	Metastases
Choriostoma	Anaplasia	Dermoid	Grade
Carcinoma <i>in situ</i>	Cancer	Desmoid	Stage
7. Compare and contrast the characteristics of benign and malignant neoplasms
8. Grading and staging of malignant tumors
9. Discuss different cancer treatment modalities and write down their role on cell cycle kinetics and anticancer effect.
10. List major classes of anticancer drugs.
11. Describe the pharmacokinetic properties of Alkylating agents.
12. Describe the mechanism of action, resistance, antibacterial activity and clinical Alkylating agents.
13. List adverse effects, contraindications/ precautions and drug interactions of Alkylating agents.
14. Describe the pharmacokinetic properties of Antifolates.
15. Describe the mechanism of action, resistance, antibacterial activity and clinical Antifolates.
16. List adverse effects, contraindications/ precautions and drug interactions of Antifolates.

17. Describe the pharmacokinetic properties of Fluoropyrimidines.
18. Describe the mechanism of action, resistance, antibacterial activity and clinical uses of Fluoropyrimidines.
19. Describe adverse effects, contraindications/ precautions and drug interactions of Fluoropyrimidines.
20. Describe the pharmacokinetic properties of Deoxycytidine analogs.
21. Describe the mechanism of action, resistance, antibacterial activity and clinical uses of Deoxycytidine analogs.
22. Describe adverse effects, contraindications/ precautions and drug interactions of Deoxycytidine analogs.
23. Describe the pharmacokinetic properties of Purine analogs.
24. Describe the mechanism of action, resistance, antibacterial activity and clinical uses of Purine analogs.
25. List adverse effects, contraindications/ precautions and drug interactions of Purine analogs.
26. Explain pharmacokinetic properties of Vinca Alkaloids & Taxanes.
27. Explain the mechanism of action, resistance, antibacterial activity and clinical uses of Vinca Alkaloids & Taxanes.
28. Describe adverse effects, contraindications/ precautions and drug interactions of Vinca Alkaloids & Taxanes.
29. Explain pharmacology of other natural product anti-cancer drugs.
30. Explain pharmacokinetic properties of antitumor antibiotics.
31. Explain the mechanism of action, resistance, antibacterial activity and clinical uses of antitumor antibiotics.
32. Explain adverse effects, contraindications/ precautions and drug interactions of antitumor antibiotics.
33. Describe the clinical pharmacology of Imatinib, Dasatinib, Nilotinib & Asparaginase.
34. Describe the pharmacokinetic properties of Growth factor receptor inhibitors.
35. Describe the mechanism of action, resistance, antibacterial activity and clinical uses of Growth factor receptor inhibitors.
36. List adverse effects, contraindications/ precautions and drug interactions of Growth factor receptor inhibitors.

### **ACQUIESCE –TO CONCUR (THEME 5)**

The students will insha Allah be able to:

1. List imaging modalities for antenatal genetic abnormalities
2. List imaging modalities used in diagnosis of neoplasia
3. Explain Characteristics present in imaging in neoplasia
4. Discuss Role of Laboratory in diagnosis of genetic and neoplastic disorders
5. Discuss Role of tumor markers in neoplasia
6. Discuss the proper choice, procedure and limits of histo-pathological and cytological evaluation
7. Explain the importance of history, physical examination findings in interpretation of tissue biopsies and cytology
8. Classification of anemia secondary to neoplastic disorder

## **PROBLEM BASED LEARNING**

### **PBL**

A boy was born to a young couple. He was little sloppy and fluffy. His eyes were slanted! Physical examination revealed an umbilical hernia. The boy had cardiac problems. He had constipation. He was admitted to neonatal intensive care! His reflexes were poor and his abdominal muscles tone was weak!

His karyotype showed 46 chromosomes while his father's karyotype contained 45 chromosomes! Further investigations are requested and required counseling was provided to his parents.

### **PBL**

42 years old female presented with H/O lump outer, lower quadrant of left breast. Patient noted the lump while taking bath one and half month back which gradually increased to present size. On local examination lump was fixed and hard. Mammography revealed calcified spots. Fine needle aspiration cytology (FNAC) was performed that revealed large cells with high N/C ratio. Various investigations and management plans were discussed.



## **RESOURCE FOR LEARNING**

### **Reference Books**

1. Basic Pathology by Robbins 9<sup>th</sup> edition
2. Pathologic Basis of Disease 8<sup>th</sup> edition
3. Pathology illustrated 25<sup>th</sup> edition
4. Walter & Israel General pathology 7<sup>th</sup> edition
5. Basic and Clinical Pharmacology by Katzung BG, Masters, SB, Trevor AJ, 13<sup>th</sup> Edition, 2015

6. Katzung & Trevor's Pharmacology by Trevor AJ, Katzung BG, Kruidering-Hall M, Masters SB, 10<sup>th</sup> Edition, 2013
7. Lippincott's Illustrated Reviews: Pharmacology, Whalen K, 6<sup>th</sup> Edition, 2015
8. Goodman & Gilman The Pharmacological Basis of Therapeutics, Brunton LL, 12<sup>th</sup> Edition, 2012
9. Pretest Pharmacology. MCQs with explanatory answer.
10. Epidemiology and biostatics by Richard Hebel 7<sup>th</sup> edition.
11. Tietz Fundamentals of Clinical Chemistry
12. Kaplan Book of Clinical Chemistry
13. Nelson text book of pediatrics
14. Manual of clinical Oncology

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## AJK Medical College, Muzaffarabad

### Genetics, Neoplasia & Pediatric Disease Module – (3<sup>rd</sup> Year)

#### Week - 1

DATE→						
TIME↓	MONDAY	TUESDAY	WEDNESDAY	THURSDAY	FRIDAY	
8:00am-09:00am	<b>LGIS</b> Introduction to Module <b>Dr. Sarosh &amp; team</b>	<b>LGIS</b> Mendelian Disorders <b>Dr. Naheem Awan</b>	<b>LGIS</b> Inherited Syndromes Associated with Cancer <b>Dr. Naheed Akhter</b>	<b>LGIS</b> Imaging for Diagnosis of Genetic Disorders in Perinatal Life <b>Dr. Shaukat Dar</b>	<b>LGIS</b> Prenatal Diagnosis & Counseling <b>Dr. Maryam Zubair</b>	
9:00am - 10:00am	<b>CLINICAL ROTATION</b>	<b>LGIS</b> Oncogenes / tumor suppressor genes <b>Dr. Zahid Azeem</b>	<b>LGIS</b> Multifactorial Disorders <b>Dr. Mateen Khan</b>	<b>CLINICAL ROTATION</b>	<b>LGIS</b> Congenital Heart Diseases <b>Dr. Manzoor Ali Khan</b>	
10:00-10:30 AM		<b>BREAK</b>			<b>BREAK</b>	
10:30-12:30am		<b>SGD</b> Genetic Disorders <b>Team-3</b> <b>Wrap up</b> <b>Dr. Muhammad Munir</b>	<b>SGD</b> Agents used in Anemias-II <b>Dr. Arif &amp; Team</b> <b>Wrap-Up: Dr. Arif</b>		<b>SGD</b> Pharmaco-genetics <b>Dr. Arif &amp; Team</b>  <b>WRAP-UP</b> <b>Dr. Arif</b>	
12:30–1:30 pm		<b>LGIS</b> Agents used in Anemias-I <b>Prof. Arif</b>	<b>LGIS</b> Chromosomal Defects-I <b>Dr. Anwar Ul Haque</b>			
1:30–2:00 PM	<b>LUNCH BREAK 1:30 – 2:00 PM</b>					
2:00pm-4:00pm	<b>LGIS</b> Genetic Language of Life <b>Dr. Anwar Ul Haque</b>	<b>PBL-1A</b> <b>Prof. Sarosh &amp; Team-3</b>	<b>SGD</b> Anemias <b>Team-3</b> <b>Wrap-up</b> <b>Dr. Malik Mahmood</b>	<b>LGIS</b> Chromosomal Defects-II <b>Dr. Anwar Ul Haque</b>	<b>SDL</b>	
	<b>SDL</b>	<b>SDL</b>		<b>LGIS</b> Agents used in Anemias-II <b>Dr. M. Arif</b>		



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### Genetics, Neoplasia & Pediatric Disease Module – (3<sup>rd</sup> Year)

#### Week - 2

DATE→					
TIME↓	MONDAY	TUESDAY	WEDNESDAY	THURSDAY	FRIDAY
8:00am-09:00am	<u><b>LGIS</b></u> Nomenclature, Grading & staging of tumors <b>Dr. Sarosh Majid</b>	<u><b>LGIS</b></u> Role of Interventional Radiology in Tumor Management <b>Dr. Shaukat Dar</b>	<u><b>LGIS</b></u> Tumors Markers & Ectopic Hormones (Paraneoplastic Syndromes) <b>Dr. Wafa</b>	<u><b>LGIS</b></u> Psychiatric help to cancer patients <b>Dr. Ayesha</b>	<u><b>LGIS</b></u> Treatment modalities in malignancies <b>Dr. Khurshid Lone</b>
9:00am-10:00am	<b>CLINICAL ROTATION</b>	<u><b>LGIS</b></u> Role of viruses in neoplasia <b>Dr. Muhammad Munir</b>	<u><b>LGIS</b></u> Molecular Diagnosis of Genetic Diseases <b>Dr. Zahid Azeem</b>	<b>CLINICAL ROTATION</b>	<u><b>LGIS</b></u> Cancer chemotherapy-III <b>Dr. M. Arif/Dr. Inayat</b>
10:00-10:30 AM		<b>TEA BREAK 10:00 – 10:30 AM</b>			
10:30am-12:30am		<u><b>SGD</b></u> Cancer chemotherapy-I <b>Dr. Arif &amp; Team</b> <u><b>Wrap-up</b></u> <b>Dr. M. Arif</b>	<u><b>SGD</b></u> Cancer chemotherapy-II <b>Dr. Arif &amp; Team</b> <u><b>Wrap-up</b></u> <b>Dr. M. Arif</b>		<u><b>SGD</b></u> Cancer chemotherapy-IV <b>Dr. Arif &amp; Team</b>  <u><b>Wrap-Up</b></u> <b>Dr. M. Arif/Dr. Inayat</b>
12:30-1:3pm		<u><b>LGIS</b></u> Signs & symptoms of Neoplasia <b>Dr. Khurshid Lone</b>	<u><b>LGIS</b></u> Blood malignancies <b>Dr. Malik Mahmood</b>		<b>Revision</b>
1:30 – 2:00 PM	<b>LUNCH BREAK 1:30 – 2:00 PM</b>				
2:00pm-4:00pm	<u><b>LGIS</b></u> Carcinogenesis <b>Dr. Anwar-ul-Haque</b>	<u><b>PBL-1B</b></u>  <b>Dr. Sarosh &amp; Team-3</b>	<u><b>SGD</b></u> Carcinogenic Microorganisms (non viral) <b>Dr. Mumtaz &amp; Team-3</b>  <u><b>Wrap-up</b></u> <b>Dr. Mumtaz</b>	<u><b>Practical</b></u>  Pathology Team-3	<b>SDL</b>
	<b>SDL</b>				

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**AJK Medical College, Muzaffarabad**  
**Genetics, Neoplasia & Pediatric Disease Module – (3<sup>rd</sup> Year)**  
**Week - 3**

DATE→					
TIME↓	MONDAY	TUESDAY	WEDNESDAY	THURSDAY	FRIDAY
8:00am-09:00am	<b>LGIS</b> Nomenclature, Grading & staging of tumors <b>Dr. Sarosh Majid</b>	<b>LGIS</b> Role of Interventional Radiology in Tumor Management <b>Dr. Shaukat Dar</b>	<b>LGIS</b> Tumors Markers & Ectopic Hormones (Paraneoplastic Syndromes) <b>Dr. Wafa</b>	<b>LGIS</b> Role of viruses in neoplasia <b>Dr. Muhammad Munir</b>	<b>SDL</b>
9:00am-10:00am	<b>CLINICAL ROTATION</b>	<p style="text-align: center;"><b>Annual Sport Week 2017</b>   <b>28<sup>th</sup> – 31<sup>st</sup> March 2017</b></p>			
10:00-10:30 AM					
10:30am-12:30am					
12:30-1:3pm					
1:30 – 2:00 PM					
2:00pm-4:00pm	<b>LGIS</b> Carcinogenesis <b>Dr. Anwar-ul-Haque</b>				
	<b>LGIS</b> Treatment of Anemia <b>Dr. M. Arif/Dr. Inayat</b>				

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**Week - 4**

DATE→						
TIME↓	MONDAY	TUESDAY	WEDNESDAY	THURSDAY	FRIDAY	
8:00am-09:00am	<u>College Holiday</u>	<u>College Holiday</u>	<u>LGIS</u> Molecular Diagnosis of Genetic Diseases <b>Prof. Anwar ul Haque</b>	<u>LGIS</u> Oncogene & Proto-oncodenes <b>Prof. Anwar</b>	<u>LGIS</u> Radiological modalities in malignancies <b>Dr. Shaukat Dar</b>	
9:00am-10:00am	College Holiday		<u>LGIS</u> Stem Cell & Homological malignancies overview <b>Dr. Malik Mehmood</b>	CLINICAL ROTATION	<u>LGIS</u> Cancer chemotherapy-I <b>Dr. M. Arif/Dr. Inayat</b>	
10:00-10:30 AM		TEA BREAK 10:00 – 10:30 AM				
10:30am-11:30pm		<u>College Holiday</u>	<u>LGIS</u> Signs, Symptoms and management strategies of Cancer <b>Dr. Khursheed Lone</b>		<u>SGD</u> Cancer chemotherapy-II <b>Dr. Arif &amp; Team</b>	
11:30am-12:30am			<u>LGIS</u> Treatment of Anemia <b>Dr. M. Arif/Dr. Inayat</b>			<u>Wrap-Up</u> <b>Dr. M. Arif/Dr. Inayat</b>
12:30-1:3pm			<u>LGIS</u> Blood malignancies <b>Dr. Malik Mahmood</b>			
1:30 – 2:00 PM	LUNCH BREAK 1:30 – 2:00 PM					
2:00pm-4:00pm	College Holiday	<u>College Holiday</u>	<u>SGD</u> Diagnostic Modalities in Genetics & Cancer <b>Prof. Anwar &amp; Team-3</b> <u>Wrap-up</u> <b>Prof. Anwar</b>	<u>Practical</u> Pathology <b>Team-3</b>	SDL	



Inquires & trouble shooting

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