# Subject No. 4 PHARMACOLOGY, PATHOLOGY AND GENETICS

Total Hours: 115 Lab Hours: 10

## Theory Hours: 105

# SECTION B - PATHOLOGY AND GENETICS (PART II – GENETICS)

Total Hours: 15

### AIM:

• This course is designed to help the student to develop an understanding of Genetics, its role in causation and management of defects and diseases.

## **OBJECTIVES:**

At the end of course students will be able to develop:

- Understanding of basic concepts of genetics.
- Understanding of maternal, prenatal, and genetic influences on development of defects and diseases.
- Understanding of genetic testing.
- Understanding of genetic disorders in various age groups.
- Understanding of services related to genetics.

### **COURSE CONTENTS:**

### **Unit I - Introduction**

- Practical application of genetics in nursing. Impact of genetic condition on family. Review of cellular division: mitosis and meiosis. Characteristics and structure of genes. Chromosomes sex determination.
- Chromosomal aberrations, patterns of inheritance: Mandolin theory of inheritance Multiple allots and blood groups. Sex linked inheritance. Mechanism of inheritance. Errors in transmission.

## Unit II - Maternal, prenatal and genetic influences on development of defects and diseases:

• Conditions affecting the mother: genetic and infections. Consanguinity atopy. Prenatal nutrition and food allergies. Maternal age. Maternal drug therapy. Prenatal testing and diagnosis. Effects of radiation, drugs and chemicals. Infertility. Spontaneous abortion. Neural tube defects and the role of folic acid in lowering the risks. Down syndrome (Trisomy 21)

## Unit III - Genetic testing in neonates and children:

• Screening for: Congenital abnormalities, Developmental delay, Dysmorphism.

## Unit IV - Genetic conditions of adolescents and adults:

• Cancer genetics – Familial cancer. Inborn errors of metabolism. Blood group alleles and haemochromatosis. Huntington's disease. Mental illness.

#### Unit V - Services related to Genetics:

• Genetic testing. Human genome project. Gene therapy. The eugenics movement. Genetic counseling. Legal and ethical issues. Role of Nurse.

Theory Hours: 15

# **GENETICS**

Total hours: 15

Unit No.	Ohiectives			Contents with distributed hours						
& Hrs.	Ubje		Must Know			Know	Nice to Know			
	At the end of unit studer	nts are able to	Introdu	action:						
Ι	<b>Knowledge:</b> Understand and describe the cellular division, chromosomes and sex determination.		• Prac • Imp • Rev • Chr	<ul> <li>Practical application of genetics in nursing.</li> <li>Impact of genetic condition on family.</li> <li>Review of cellular division: mitosis and meiosis.</li> <li>Chromosomes – sex determination.</li> </ul>			ics and genes. heory of f	Ι		
(3 Hrs)	disease conditions in cli	nical practice.	• Chr	omosomal aberratio	ons, patterns of	inheritance Errors in		(3 Hrs)		
	Attitude: Incorporate the chromosomes in identify various disease condition	e knowledge of ying genetic impac ns.	• Mul • Sex	<ul> <li>Multiple allots and blood groups.</li> <li>Sex linked inheritance. (2 Hrs)</li> </ul>			n. (1 Hr)			
Unit I	l					I				
(	Course outcome			Pro	gramme outcome					
Students	s should be able to-	Nurse/Clinician	Professional	Communicator	Leader & Member	Lifelong learner	Critical thinker	Resear cher		
•		PO1	PO2	PO3	PO4	PO5	PO6	PO7		
CO-1: I application	Describe practical on of genetics in nursing	3	3	2	3	3	2	0		
CO-2: E genetic c	Explain Impact of ondition on family.	3	3	2	3	3	2	0		
<b>CO-3:</b> I mitosis a	Differentiate between nd meiosis	3	3	2	3	3	2	0		
<b>CO-4:</b> D	efine sex determination.	3	3	2	3	3	2	0		
CO-5: E aberratio	Explain Chromosomal ns and patterns of	3	3	2	3	3	2	0		

inheritan	ce.							
CO-6: D and blood	Describe multiple allots d groups	3	3	2	3	3	2	0
<b>CO-7</b> : Exinheritan	xplain Sex linked ce	3	3	2	3	3	2	0
CO-8: E and draw	nlist the characteristics the structure of genes	3	3	2	3	3	2	0
CO-9: E theory of	xplain Mendalian inheritance.	3	3	2	3	3	2	0
<b>CO-10</b> : Mechanis	Describe the sm of inheritance.	3	3	2	3	3	2	0
<b>CO11:</b> E transmiss	numerate the errors in sion	3	3	2	3	3	2	0
IIAt the end of unit students are able toII <b>Knowledge:</b> Describe the mode of transmission of genetic diseases.II <b>Skill:</b> Counsels regarding role of consanguineous marriages in inheritance of diseases.(3 Hrs) <b>Attitude:</b> Motivates individuals for genetic testing and thereby contribute in preventing hereditary diseases.			Mate devel sion • C i • C • F • N • N • I • I • I • I	rnal, prenatal and g opment of defects an Conditions affecting the fections. Consanguinity atrophy renatal nutrition and Maternal age. Maternal drug therapy infertility Prenatal testing and of Ciffects of radiation, of Down syndrome (Tri 2 hours)	<ul> <li>Spontaneous abortion.</li> <li>Neural tube and the role acid in lower risks. (1 Hr)</li> </ul>	defects of folic ing the	II (3 Hrs)	
Unit II								
Course outcome     Programme outcome								

Students should be able to-	Nurse/Clinician	Professional	Communicator	Leader & Member	Lifelong learner	Critical thinker	Resear cher
	PO1	PO2	PO3	PO4	PO5	PO6	PO7
<b>CO-1:</b> List out the genetic and infections conditions affecting the mother at the time f pregnancy.	3	3	2	3	3	2	1
<b>CO-2:</b> Explain Consanguinity atrophy.	3	3	2	3	3	2	2
<b>CO-3:</b> Explain prenatal nutrition and food allergies.	3	3	2	3	3	2	1
<b>CO-4:</b> Explain the maternal age and maternal drug therapy that influence on development of defects and disease in maternal.	3	3	2	3	3	2	1
CO-5: Explain Infertility.	3	3	2	3	3	2	1
<b>CO-6</b> : Describe prenatal testing and its diagnostic evaluation.	3	3	2	3	3	2	1
<b>CO-7:</b> Explain effects of radiation, drugs and chemicals on development of defects and disease in maternal.	3	3	2	3	3	2	1
<b>CO-8</b> : Describe Down syndrome (Trisomy 21)	3	3	2	3	3	2	1
<b>CO-9</b> : Explain Spontaneous abortion.	3	3	2	3	3	2	2

CO-10: tube defe folic ac risks.	Describe the Neural ects and enlist role of id in lowering the	3	3	2	3	3	2	2
III (2 Hrs)	At the end of unit stud <b>Knowledge:</b> Understa abnormalities. <b>Skill:</b> Identify congent <b>Attitude:</b> Provide con to client having congent	Image: Interpretendent structure       Genetic testing in neonates and children:         Ind and explain congenital       Screening for: Congenital abnormalities, Developmental delay, (1 Hr)         Ital abnormalities.       Ital abnormalities.			es and children: tal abnormalities, (1 Hr)		• (1	Dysmor phism hour)
Unit III C	ourse outcome			Prog	ramme outcome			
Students	should be able to-	Nurse/Clinician PO1	Professional PO2	Communicator PO3	Leader & Member PO4	Lifelong learner PO5	Critical thinker PO6	Resear cher PO7
<b>CO-1:</b> I abnorma screening detection abnorma children.	Define Congenital lities and list out the g test used for of congenital lities in neonates and	3	3	2	3	3	2	1
CO-2: E procedur developn	Explain screening e for detection of nental delay.	3	3	2	3	3	2	1
<b>CO-3</b> : E	Explain dysmorphism	3	3	2	3	3	2	0
IV (2Hrs)	IVAt the end of unit students are able to Hrs)Genetic conditions of adolescents and adults:• Blo and ad haeHrs)Knowledge: Understand and explain the genetic abnormalities, their causes and signs &• Cancer genetics – Familial cancer. • Inborn errors of metabolism.• Hut					<ul> <li>Blood group and haemochrom</li> <li>Huntington'</li> </ul>	alleles natosis s	•

sympto Skill: Attitu clients	oms. Identify the clier <b>de:</b> Provide effe	nt with genetic disc	• Me (1) to such	ental illness. Hr)		disease. (1 Hr)			
Unit IV									
Course	outcome			Programme outcome					
Students should be able to-		Nurse/Clinician	Professional	Communicator	Leader & Member	Lifelong learner	Critical thinker	Resear cher	
•		PO1	PO2	PO3	PO4	PO5	PO6	<b>PO7</b>	
<b>CO-1:</b> Define ( and Familial car causes.	Cancer genetics ncer. Enlist its	3	3	2	3	3	2	0	

causes.							
<b>CO-2:</b> Explain Inborn errors of metabolism with its cause transmission.	3	3	2	3	3	2	0
<b>CO-3:</b> Define mental illness and list out the causes, sign and symptoms, complication and prevention.	3	3	2	3	3	2	0
<b>CO-4:</b> Define haemochromatosis; explain its causes and types.	3	3	2	3	3	2	0
<b>CO5:</b> Explain blood group alleles.	3	3	2	3	3	2	0
<b>CO6:</b> Define Huntington's disease. Enlist its symptoms	3	3	2	3	3	2	0

V (5 Hrs)	At the end of unit stude <b>Knowledge:</b> Understar <b>Skill:</b> Provide genetic of testing and assist in gen <b>Attitude:</b> Perform nurs	ents are able to nd the Gene therapy counseling for gene ne therapy. ses' role effectively	7. Services • Gen • Gen • Role (3 H	<ul> <li>Services related to Genetics:</li> <li>Genetic testing.</li> <li>Genetic counseling.</li> <li>Role of Nurse (3 Hrs)</li> </ul>			<ul> <li>Legal and ethical issues</li> <li>Gene therapy</li> <li>(1 Hr)</li> </ul>	
Unit V	I							
(	Course outcome			Prog	gramme outcome			
Students	s should be able to-	Nurse/Clinician	Professional	Communicator	Leader & Member	Lifelong learner	Critic think	cal Resear er cher
		PO1	PO2	PO3	PO4	PO5	PO	5 PO7
CO-1: I and gene	Define Genetic testing tics counseling.	3	3	2	3	3	2	1
CO-2: E related to	Explain the services genetic counseling.	3	3	2	3	3	2	1
<b>CO-3:</b> I ethical is	Describe the legal and sues in genetic testing.	3	3	2	3	3	2	1
<b>CO-4:</b> E	xplain gene therapy.	3	3	2	3	3	2	0
<b>CO-5</b> : E	Explain eugenics.	3	3	2	3	3	2	0
<b>CO-6</b> : D project an importan	Define Human genome nd list out its ce.	3	3	2	3	3	2	0

#### **TEACHING STRATEGY:**

Total Teaching Hours: 15

#### **ASSIGNMENTS:**

A. Section 'C': Genetics

Theory

Sr. No	Assignments	No./Quantity	Marks Per Assignment	Total Marks
1	Home assignment	One	20	20

While calculating Internal Assessment –Marks obtained in the assignments of Pharmacology and Pathology and Genetics shall be amalgamated as one subject, 'Pharmacology, Pathology and Genetics'.

#### **TEACHING METHODS:**

• Lectures, Laboratory Demonstration, Group Discussion, Clinical Observation and Tutorial

#### A.V. AIDS:

• Over head projector, L.C.D. Computer Assisted Instruction, Black Board.

#### LIST OF RECOMMENDED BOOKS:

- S. Mandal : Fundamentals of Human Genetics
- S. D. Gangane : Human Genetics
- Jordey carey Roberts : An Introduction to Medical Genetics
- Elizabeth F. Lanzl : Medical Genetics
- J. Ben Hill, Helen Hill : Medical Genetics and Human Heredity
- Edmund W. Sinnott : Principles of Genetics
- P. C. Winter, G. I. Hickey : Instant Notes in Genetics
- Ching Chun L : Human Genetics Principles and methods
- Mary B. Mahowald, et al : Genetics In Clinic
- Robert F. Muller et al :Emery's Elements of Medical Genetics
- Moore Keith L : Developing Human clinically Oriented Embryology
- Pansky Ban : Review of Medical Embryology
- Smell Richard S : Clinical Embryology for Medical Students
- Lnagman Jan : Medical Embryology