Course Code				**23ZOMAP232**			
Title of the Course				**PRINCIPLES OF GENETICS**			
Offered to:				B.Sc. Hons Zoology			
L	0	**T**	**0**	**P**	**2**	**C**	**1**
Year of Introduction:		**2024-25**		**Semester:**			**3**
Course Category:		**MAJOR**		**Course Relates to:**		**GLOBAL**	
Year of Revision:		**NA**		**Percentage:**		**NA**	
Type of the Course:				**SKILL DEVELOMENT**			
Crosscutting Issues of the Course :				**GENDER**			
Pre-requisites, if any				**BASICS OF INHERITANCE**			

Course Description:

The logic of science has three essential components, such as observations, facts, and conceptualizations. The main processes of science are induction and deduction by reasoning and hypothesis testing. The father of Genetics, Gregor Mendel, even though applied all these principles by deducing the thumb rule of inheritance, took 35 years of gestation to accept it.

Genetics is a fundamental science which deals with the study of genes, characters, and their inheritance. The knowledge of genetics has become essential to unravel the mysteries of life processes that are extensively pursued in today's science. This course is designed to understand the major concepts and processes of genetics. It provides a foundation for understanding the rule of inheritance and genetics applications for undergraduate natural science students.

Course Objectives:

S. No	COURSE OBJECTIVES
1	Providing the background knowledge on the history of genetics and the importance of Mendelian principles
2	Providing the required knowledge on the gene interactions
3	Acquainting the students, distinguish between polygenic, sex-linked, and multiple allelic modes of inheritance and extra chromosomal inheritance.
4	Understanding the principles of sex determination in animals with a reference to human being, and sex-linked inheritance
5	Understanding the human karyotyping and the concept of pedigree analysis basics.

Course Outcomes

At the end of the course, the student will / will be…

NO	COURSE OUTCOME	BTL	PO	PSO
CO1	Gain knowledge on basic terminology of genetics	K1	2	1
CO2	Understand various types of inheritance patterns existing in animals with reference to non-Mendelian inheritance.	K2	2	1
CO3	Understand and gain knowledge on multiple alleles and Extra-chromosomal inheritance	K2	2	1
CO4	Applying several of aspects of genetics involved in sex determination.	K3	2	1
CO5	Analyzing human karyotyping, pedigree analysis and chromosomal disorders concepts of proteomics and genomics	K4	6	1

For BTL: K1: Remember; K2: Understand; K3: Apply; K4: Analyze; K5: Evaluate;K6: Create

CO-PO-PSO MATRIX									
CO NO	PO1	PO2	PO3	PO4	PO5	PO6	PO7	PSO1	PSO2
CO1		2						2	
CO2		2						2	
CO3		2						2	
CO4		3						3	
CO5						3		3	

Use the codes 3, 2, 1 for High, Moderate and Low correlation Between CO-PO-PSOrespectively

Course Structure:

Unit – I: Introduction to Genetics (6 Hrs)

Practical 1:

Study the Mendelian inheritance using suitable examples/Problems. Certainly! Here's a hands-on activity to help understand Mendelian genetics through a simple and engaging experiment. This activity is designed to illustrate basic concepts such as dominant and recessive traits, as well as the principles of inheritance using a simulated cross.

Activity: Pea Plant Genetics Simulation
Problem1: Monohybrid cross
Scenario:
In pea plants, flower color is determined by a single gene with two alleles: red (R) and white (r). Red is dominant over white.

Question:
If you cross two heterozygous red-flowered pea plants (Rr x Rr), what is the probability of their offspring having white flowers?

Solution: Determine the possible genotypes:

- Parent 1: Rr
- Parent 2: Rr

Use a Punnett square to find the genotypic ratio of the offspring:

	R	**r**
R	R R	R r
r	R r	r r

Genotypic Ratio:

- RR: 1
- Rr: 2
- rr: 1

Phenotypic Ratio:

- Red (RR + Rr): 3
- White (rr): 1

Probability of white flowers: 1/4 or 25%

Problem 2: Dihybrid Cross

Scenario:

In pea plants, seed shape (Round vs. Wrinkled) and seed color (Yellow vs. Green) are controlled by two different genes. Round (R) and Yellow (Y) are dominant over Wrinkled (r) and Green (y), respectively.

Question:

If you cross two pea plants that are both heterozygous for both traits (RrYy x RrYy), what is the probability that an offspring will have round, yellow seeds?

Solution:

Determine the gametes for each parent:

Each parent can produce gametes with the following combinations of alleles: RY, Ry, rY, ry.

Construct a Punnett square for each trait (for simplicity, use a 16-cell Punnettsquare for the dihybrid cross):

	RY	**Ry**	**rY**	**ry**
RY	RRYY	RRYy	RrYY	RrY y
Ry	RRYy	RR yy	RrYy	RrYy
r Y	RrYY	RrYy	rrYY	rrYy
ry	RrYy	Rryy	rrYy	rryy

1. **Count the phenotypes with round, yellow seeds:**
   Round and Yellow require at least one dominant allele for both traits.
2. **Genotypic Combinations:**
   - R-R-Y-Y
   - R-R-Y-y
   - R-r-Y-Y
   - R-r-Y-y

   Phenotypic Ratio (Round, Yellow):9/16
   Probability of round, yellow seeds: 9/16 or 56.25%

Problem 3: Test Cross

Scenario:

In a certain species, tall plants (T) are dominant over short plants (t). A tall plant is crossed with a short plant, and the offspring include both tall and short plants.

Question:

What is the genotype of the tall parent?

Solution:

1. **Analyze the cross:**
   Since the offspring include both tall and short plants, the tall parent must be heterozygous (Tt).
2. **Possible crosses:**
   - Tall (Tt) x Short (tt) results in offspring with a 1:1 ratio (Tall).
3. **Genotypic Ratio of the Offspring:**
   - Tt: Tall
   - tt: Short

   Conclusion: The genotype of the tall parent is Tt.

Unit – II: Non-Mendelian Genetics I

Practical 2

Problem1: Incomplete dominance

Scenario:

In snapdragons, flower color is controlled by a gene with incomplete dominance. Red (RR) and white (WW) flowers produce pink (RW) flowers when crossed.

Question:

What will be the phenotypic ratio of the offspring when two pink snapdragons (RW x RW) are crossed?

Solution:

1. **Determine the genotypes:**
   - Parent 1: RW
   - Parent 2: RW
2. **Use a Punnett square to determine the genotypic ratio:**

	R	**W**
R	R R	R W
W	R W	W W

3. **Genotypic Ratio:**
   - RR: 1
   - RW: 2
   - WW: 1

   Phenotypic Ratio:
   - Red (RR): 1
   - Pink (RW): 2
   - White (WW): 1

   Phenotypic Ratio: 1:2:1

Problem 2: Co-dominance

Codominance (Blood types) Human blood types are determined by genes that follow the Codominance pattern of inheritance.

There are two dominant alleles A and B and one recessive allele O.

Blood Type Phenotype	Blood Type Genotype	Blood Type Can donate blood to:	Blood Type Can receive blood from:
O	OO	A, B, AB and O (universal donor)	O
AB	AB	AB	A, B,AB and O (universal receiver)
A	AA or A O	AB, A	O, A
B	BB or BO	AB, B	O,B

Write the genotype for each person based on the description:

a. Homozygous for the "B" allele _____

b. Heterozygous for the "A" allele _____

c. Type O _____

d. Type "A" and had a type "O" parent _____

e. Type "AB" _____

f. Blood can be donated to anybody _____

g. Can only get blood from a type "O" donor _____

2. If a father is homozygous for the type B allele, and mother is type "O." What are all the possible blood types of their baby? (show your work)

3. Draw a Punnett square showing all the possible blood types for the offspring produced by a type "O" mother and a Type "AB" father

4. Mrs. Clink is type "A" and Mr. Clink is type "O." They have three children named Matthew, Mark, and Luke. Mark is type "O," Matthew is type "A," and Luke is type "AB." Based on this information:

a. Mr. Clink must have the genotype _____

b. Mrs. Clink must have the genotype_____because_____has blood type _____

c. Luke cannot be the child of these parents because neither parent has the allele_____.

5. Two parents think their baby's identity was mixed up at the hospital. The mother has blood type "O," the father has blood type "AB," and the baby has blood type "B."

a. Mother's genotype: _____

b. Father's genotype: _____

c. Baby's genotype:_____or _____

d. Punnett square showing all possible genotypes for children produced by this couple

e. Was the baby switched?

Note: More problems can be added

PRACTICAL 3

Problem 1: Simple Epistasis

Epistatic and non-epistatic gene interactions are important concepts in genetics that describe how different genes can influence one another. Here are some problems that illustrate these concepts:

Scenario:

In rabbits, coat color is determined by two genes: **B** for black or brown fur, and **E** for pigment expression. The B gene has two alleles: **B** (black) and **b** (brown). The E gene has two alleles: **E** (pigment expressed) and **e** (pigment not expressed). The presence of the **e** allele masks the effect of the **B** gene, resulting in a white coat regardless of the B gene's alleles.

Question:

If two rabbits heterozygous for both traits (BbEe) are crossed, what are the expected phenotypic ratios of the offspring?

Solution:

1. **Determine Genotypic Combinations:**
   - **Phenotypes to consider:**
     - Black fur (B- E-)
     - Brown fur (bb E-)
     - White fur (B- ee or bb ee)
2. **Set Up a Punnett Square for Each Gene:**
   - For the **B** gene and **E** gene interactions:

	BE	**Be**	**bE**	**be**
BE	BBEE	BBEe	BbEE	BbEe
Be	BBEe	BBee	BbEe	Bbee
bE	BbEE	BbEe	bbEE	bbEe
be	BeEe	BbEE	bbEe	bbee

3. **Count the Phenotypes:**
   - **White fur:** The presence of ee means the rabbit will be white regardless of the B gene's alleles.
     - **B- ee:** BbEe, BbEe, BbEe, Bbee, Bbee, BbEe, bbEe, bbEe, bbee, bbee
     - **Number of white fur offspring = 9 out of 16**
   - **Black fur (B- E-):** The rabbits must have at least one B and one E allele.
     - **BBEE, BBEe, BbEE, BbEe**
     - **Number of black fur offspring = 9 out of 16**
   - **Brown fur (bb E-):** The rabbits must be homozygous for b and have at least one E allele.
     - **bbEE, bbEe**
     - **Number of brown fur offspring = 3 out of 16**

Phenotypic Ratios:

- White: 9/16
- Black: 3/16
- Brown: 3/16

Problem 2: Duplicate Gene Action

Scenario:

In a certain flower species, flower color is influenced by two genes, **A** and **B**. The dominant alleles for each gene result in a red flower, while the recessive alleles produce a white flower. The presence of at least one dominant allele in either gene produces a red flower.

Question:

If two plants heterozygous for both traits (AaBb x AaBb) are crossed, what are the expected phenotypic ratios?

Solution:

1. **Set Up the Cross:**
   - Each gene pair can produce four types of gametes: AB, Ab, aB, ab.
2. **Construct a 16-Cell Punnett Square:**

	AB	**Ab**	**aB**	**ab**
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaB b	aaBB	aaBb
ab	AaBb	Aab b	aaBb	aabb

3. **Count the Phenotypes:**
   - **Red Flowers:** Requires at least one dominant allele from either gene (A- or B-).
     - **AABB, AABb, AaBB, AaBb, AABb, AaBb, AaBB, AaBb**
     - **Number of red flowers = 15 out of 16**
   - **White Flowers:** Requires both genes to be homozygous recessive (aabb).
     - **Number of white flowers = 1 out of 16**

Phenotypic Ratios:

- Red: 15/16
- White: 1/16

Problem 3: Recessive Epistasis

Scenario:

In Labrador retrievers, coat color is determined by two genes: **B** and **E**. The B gene controls black (B) or brown (b) fur color, while the E gene controls pigment deposition. The presence of a recessive **ee** genotype means no pigment is deposited, resulting in a yellow coat regardless of the B gene's alleles.

Question:

If two Labradors that are heterozygous for both traits (BbEe) are crossed, what are the expected phenotypic ratios?

Solution:

1. **Determine Possible Genotypes and Phenotypes:**
   - **Genotype combinations:**
     - BBEE, BBEe, BbEE, BbEe, BBee, Bbee, bbEE, bbEe, bbee
2. **Construct a Punnett Square:**

	BE	Be	bE	be
BE	BB EE	BBEe	BbEE	BbEe
Be	BBEe	BBEe	BbEe	Bbee
bE	BbEE	BbEe	bbEE	bbEe
be	BbEe	Bbee	bbEe	bbee

3. **Count the Phenotypes:**
   - **Yellow Fur:** The presence of **ee** will result in yellow fur regardless of the B gene.
     - **Number of yellow fur offspring = 9 out of 16**
   - **Black Fur:** Requires at least one dominant **B** and **E** allele.
     - **Number of black fur offspring = 3 out of 16**
   - **Brown Fur:** Requires homozygous recessive **bb** and at least one dominant **E** allele.
     - **Number of brown fur offspring = 3 out of 16**

Phenotypic Ratios:

- Yellow: 9/16
- Black: 3/16
- Brown: 3/16

Problem 4 : Dominant Epistasis

Scenario:

In summer squash, fruit color is determined by two genes. The **W** gene (dominant) inhibits the production of pigment regardless of the **Y** gene's alleles. The **Y** gene (dominant) produces yellow color if **W** is not present. The combination **ww** with **YY** or **Yy** produces a green color.

Question:

If two squash plants heterozygous for both traits (WwYy x WwYy) are crossed, what are the expected phenotypic ratios?

Solution:

1. **Determine Possible Genotypes and Phenotypes:**
   - **Genotype combinations:**
     - WWYY, WWYy, WWyy, WwYY, WwYy, Wwyy, wwYY, wwYy, wwyy
2. **Construct a Punnett Square:**

	WY	Wy	wY	wy
WY	WWYY	WWYy	WWYy	WWyy
Wy	WWYy	WwY y	WwY y	Wwy y

wY	WWYy	WwY y	wwYY	wwYy
wy	WWyy	Wwy y	wwYy	wwyy

Count the Phenotypes:

- **Yellow:** Any genotype with at least one **W** and one **Y** (W- Y-).
  - **Number of yellow fruits = 12 out of 16**
- **Green:** Requires both recessive **ww** and at least one dominant **Y**.
  - **Number of green fruits = 3 out of 16**
- **White:** Requires **W** and **yy** (W- yy).
  - **Number of white fruits = 1 out of 16**

Phenotypic Ratios:

- Yellow: 12/16
- Green: 3/16
- White: 1/16

These problems help illustrate how genes interact with one another in different genetic scenarios. Feel free to adjust these problems or add more complexity as needed!

Here's a practical activity designed to help understand the concepts of genetic linkage and crossing over. This activity simulates the processes of linkage and crossing over using a simple and engaging model.

PRACTICAL 4

Practical Activity: Simulating Genetic Linkage and Crossing Over

Problem 1:

Objective:

To understand how genetic linkage and crossing over affect the inheritance of traits and the resulting genetic variation.

Materials:

- Colored beads or small tokens (to represent different alleles)
- String or pipe cleaners (to represent chromosomes)
- Scissors
- Paper and pencils (for recording data)
- A chart or graph paper (for recording and analyzing results)

Preparation:

1. Create Chromosome Models:

oUse pipe cleaners or strings to represent chromosomes. Each chromosome will have loci (specific positions) for different genes. For simplicity, we will use two linked genes on each chromosome.

2. Choose Traits:

Gene 1 (Trait A): Dominant allele (A) and recessive allele (a)

Gene 2 (Trait B): Dominant allele (B) and recessive allele (b)

These genes are linked on the same chromosome.

3. Create Allele Sets:
Prepare sets of colored beads to represent different alleles:
- **Red bead:** Dominant allele A
- **Blue bead:** Recessive allele a
- **Green bead:** Dominant allele B
- **Yellow bead:** Recessive allele b

4. Model Chromosomes:
Create two chromosome models for each parent:
- **Chromosome 1:** AB or ab (linked genes)
- **Chromosome 2:** AB or ab (linked genes)

Activity Steps:
1. Simulate the Parental Genotypes:
Choose two parent models:
- **Parent 1:** AB/ab (heterozygous)
- **Parent 2:** AB/ab (heterozygous)

Create chromosome models by placing beads on the pipe cleaners or strings to represent these genotypes.

2. Set Up a Cross:
o Simulate the process of meiosis by separating the chromosome pairs into gametes. Each chromosome will be a combination of the alleles present on it.
Gametes for Parent 1:
AB
ab
Gametes for Parent 2:
AB
ab

3. Simulate Crossing Over:
oIntroduce crossing over by swapping segments between the chromosomes. For example, swap the segments containing **A** and **B** between the chromosomes.
Example of Crossing Over:
Original Chromosome: AB / ab
After Crossing Over: Ab / aB
Create new gametes from the crossed-over chromosomes:
AB (non-crossover)
ab (non-crossover)
Ab (crossover)
aB (crossover)

4. Create a Punnett Square:
Cross the gametes from Parent 1 and Parent 2:

	AB	ab	Ab	aB
AB	AABB	AAbb	AaBB	AaBb
ab	AAbb	aabb	AaBb	aabb
Ab	AaBB	AaBb	aaBB	aaBb
aB	AaBb	aabb	aaBb	aabb

Analyze the Results:

Count the Genotypes and Phenotypes:

Determine the ratios of the different phenotypes produced.

Compare the results with and without crossing over.

Phenotypic Ratios:

- Calculate the frequency of each phenotype and compare how linkage affects the inheritancepatterns.

Discussion:

Linkage: Discuss how genes that are close together on the same chromosome are more likely tobe inherited together.

Crossing Over: Discuss how crossing over during meiosis can create new allele combinations andincrease genetic diversity.

Extension Activity:

1.Multiple Genes: Extend the activity to more genes or different chromosomes to explore more complex interactions.

2. Real Data: Compare the simulated results with real genetic data from organisms that exhibit linkage and crossing over.

This practical activity allows you to visualize and simulate the effects of linkage and crossing over, helping to understand these important genetic concepts through hands-on learning.

UNIT 3: Multiple Alleles and Extra chromosomal inheritance

Practical 5

Problem 1: Blood Group Determination Using Blood Typing

Objective: To determine the blood group of individuals using blood typing techniques.

Materials:

- Blood typing reagents (Anti-A, Anti-B, Anti-D (for Rh factor))
- Blood samples (can use simulated blood samples if actual blood is not available)
- Microscope slides
- Pipettes
- Clean glassware (e.g., test tubes)
- Blood typing trays (if available)

Procedure:

1. **Prepare Blood Samples:**
    - Place a few drops of blood from the sample onto separate sections of a blood typingtray or microscope slide.

2. **Add Reagents:**
    - Add a drop of Anti-A reagent to one section, Anti-B reagent to another section, andAnti-D reagent to a third section of the tray or slide.
3. **Mix and Observe:**
    - Gently mix the blood with each reagent using a pipette or stick.
    - Observe for agglutination (clumping). Agglutination with Anti-A indicates the presence of A antigen, with Anti-B indicates the presence of B antigen, and withAnti-D indicates the Rh factor (positive or negative).
4. **Determine Blood Type:**
    - Based on the agglutination pattern, determine the blood group (e.g., A+, B-, AB+, O-,etc.).

Problem 2.: Genetic Inheritance of Blood Groups

Objective: To understand how blood groups are inherited according to Mendelian genetics.

Materials:

- Punnett squares
- Blood group charts for parents and offspring
- Simulation software (optional)

Procedure:

1. **Create Parental Genotypes:**
    - Use the ABO and Rh blood group systems. For example, create Punnett squares for various crosses, such as A (IAIA or IAi) x B (IBIB or IBi) or Rh+ (DD or Dd) x Rh-(dd).
2. **Analyze Offspring Genotypes:**
    - Calculate the possible genotypes and phenotypes of the offspring using the Punnettsquares.
3. **Compare with Real Data:**
    - If possible, use actual blood group data from family members to compare with theexpected results.

Problem: 3. Understanding Blood Group Distribution in Populations

Objective: To explore the distribution and frequency of different blood groups in various populations.

Materials:

- Blood group distribution data (can be found in literature or online resources)
- Graph paper or computer software for data analysis

Procedure:

1. **Collect Data:**
    - Obtain data on the distribution of blood groups in different populations or regions.
2. **Analyze Frequencies:**
    - Calculate the frequency of each blood group within the population. For example,determine the percentage of individuals with type A, B, AB, and O blood groups.
3. **Create Charts:**
    - Use graphs or charts to visualize the distribution of blood groups. Discuss factors thatmight influence these distributions, such as genetic drift, migration, or natural selection.

Problem 4. Simulating Blood Transfusions

Objective: To understand the principles of blood compatibility and the importance of matching blood types.

Materials:

- Simulated blood samples with known blood types
- Blood typing reagents
- Test tubes or mixing containers

Procedure:

1. **Prepare Blood Samples:**
   - Simulate different blood types (e.g., A, B, AB, O) and prepare samples for testing.
2. **Simulate Transfusions:**
   - Mix simulated blood samples from donors and recipients with different blood types.
3. **Observe Reactions:**
   - Look for agglutination or other reactions to demonstrate compatibility orincompatibility.
4. **Discuss Results:**
   - Discuss why certain blood types are compatible or incompatible and the importanceof blood typing in real-life blood transfusions.

Problem 6: Blood Typing in Forensic Science

Objective: To understand the application of blood typing in forensic science and paternity testing.

Materials:

- Blood typing reagents and samples (real or simulated)
- Forensic case studies or scenarios

Procedure:

1. **Examine Forensic Cases:**
   - Use hypothetical or real case studies where blood typing played a role in forensicinvestigations.
2. **Simulate Blood Typing:**
   - Perform blood typing tests on simulated crime scene samples and suspect samples.
3. **Analyze Results:**
   - Determine the likelihood of the suspect being the source of the crime scene bloodbased on the blood typing results.

PRACTICAL 6

Extranuclear inheritance, also known as cytoplasmic inheritance, refers to the transmission of genetic material that is not found within the nucleus. This genetic material is usually located in organelles such as mitochondria and chloroplasts. In humans, mitochondrial DNA (mtDNA) is the primary focus of extranuclear inheritance. Here are some common problems associated with extranuclear inheritance:

1. Mitochondrial Disorders

Mitochondrial disorders result from mutations in the mitochondrial DNA. These mutations can affect the function of the mitochondria, leading to a range of diseases. Some common mitochondrial disorders include:

Leber's Hereditary Optic Neuropathy (LHON):
Symptoms: Sudden vision loss in young adulthood, primarily affecting males.
Mitochondrial Myopathy:
Symptoms: Muscle weakness, exercise intolerance, and muscle pain.
Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-like episodes (MELAS):
Symptoms: Muscle weakness, seizures, strokes, and lactic acidosis.
Myoclonic Epilepsy with Ragged Red Fibers (MERRF):
Symptoms: Muscle jerks, seizures, ataxia, and muscle biopsy showing raggedred fibers.

Kearns-Sayre Syndrome (KSS):
Symptoms: Progressive external ophthalmoplegia, retinitis pigmentosa, andheart block.

2. Maternal Inheritance
Since mitochondria are typically inherited from the mother, mitochondrial disorders are passed down maternally. This means that all offspring of an affected mother can inherit the disorder, whereas fathers do not pass on mitochondrial DNA to their children.

3. Heteroplasmy
Heteroplasmy refers to the presence of both normal and mutated mtDNA within the same cell. The proportion of mutated to normal mtDNA can vary between cells and tissues, leadingto a wide range of phenotypic expressions of mitochondrial diseases. This variability complicates diagnosis and prognosis.

4. Threshold Effect
The threshold effect describes the phenomenon where a certain proportion of mutated mtDNA must be exceeded before a cell expresses symptoms of a mitochondrial disorder. Below this threshold, the cell can often function normally. Above it, the cell exhibits dysfunction, leading to disease. This threshold can differ among tissues and individuals, contributing to the complexity of mitochondrial diseases.

5. Complexity of Diagnosis
Diagnosing mitochondrial disorders can be challenging due to the variability in symptoms and the overlap with other diseases. Diagnosis often involves:

- **Genetic Testing:** To identify mutations in mtDNA.
- **Biochemical Tests:** To assess mitochondrial function.
- **Muscle Biopsy:** To detect abnormalities in muscle tissue.

6. Limited Treatment Options
Treatment for mitochondrial disorders is often limited to managing symptoms and slowing disease progression. Current approaches include:

- **Nutritional Supplements:** Such as Coenzyme Q10, L-carnitine, and vitamins.
- **Exercise Therapy:** To improve muscle function and endurance.
- **Symptomatic Treatment:** For specific symptoms like seizures or heart issues.

7. Research and Ethical Challenges

Research into mitochondrial disorders and potential therapies, such as mitochondrial replacement therapy (MRT), raises ethical and technical challenges. MRT involves replacing mutated mitochondria with healthy ones from a donor, creating a child with genetic material from three individuals. This procedure raises questions about long-term safety, genetic identity, and ethical considerations.

Examples of Research and Controversy:

Mitochondrial Replacement Therapy (MRT):

- **Benefits:** Potential to prevent the transmission of mitochondrial diseases.
- **Ethical Concerns:** Genetic modification of embryos, long-term effects on offspring, and implications for genetic identity.

Conclusion

Extranuclear inheritance, particularly involving mitochondrial DNA, presents unique challenges in understanding, diagnosing, and treating associated disorders. Continued research and advances in genetic and medical technology hold promise for improving the management and outcomes of these conditions

PRACTICAL7

Here are some problems related to sex-linked inheritance, along with their solutions:

Problem 1: Color Blindness

Question: A woman who is a carrier for color blindness (X*X) marries a man with normal vision (XY). What are the possible genotypes and phenotypes of their children?

Solution:

1. **Mother's Genotype:** X*X (Carrier)
2. **Father's Genotype:** XY (Normal)

The possible genotypes of their children are determined by the following Punnett square:

	X	**Y**
X	X*	X*
*	X	Y
X	X	X
	X	Y

Genotypes and Phenotypes:

- **X*X (Carrier Female):** 25% chance, female carrier but with normal vision.
- **XX (Normal Female):** 25% chance, female with normal vision.
- **X*Y (Color Blind Male):** 25% chance, male with color blindness.
- **XY (Normal Male):** 25% chance, male with normal vision.

Problem 2: Hemophilia

Question: A woman who is not a carrier for hemophilia (XX) marries a man with hemophilia (X*Y). What are the possible genotypes and phenotypes of their children?

Solution:

1. **Mother's Genotype:** XX (Normal)
2. **Father's Genotype:** X*Y (Hemophilia)

The possible genotypes of their children are determined by the following Punnett square:

	X	**X**
X*	X*	X*
Y	X	X
	Y	Y

Genotypes and Phenotypes:

- **X*X (Carrier Female):** 50% chance, female carrier of hemophilia.
- **XY (Normal Male):** 50% chance, male with normal blood clotting.

Problem 3: Duchenne Muscular Dystrophy
Question: A woman who is a carrier for Duchenne muscular dystrophy (X*X) marries a man who is unaffected (XY). What are the possible genotypes and phenotypes of their children?

Solution:

1. **Mother's Genotype:** X*X (Carrier)
2. **Father's Genotype:** XY (Normal)

The possible genotypes of their children are determined by the following Punnett square:

	X	**Y**
X*	X*	X*
	X	Y
X	X	X
	X	Y

Genotypes and Phenotypes:

- **X*X (Carrier Female):** 25% chance, female carrier of Duchenne muscular dystrophy.
- **XX (Normal Female):** 25% chance, female with normal muscle function.
- **X*Y (Affected Male):** 25% chance, male with Duchenne muscular dystrophy.
- **XY (Normal Male):** 25% chance, male with normal muscle function.

Problem 4: Rett Syndrome (X-Linked Dominant)
Question: A woman affected by Rett syndrome (X*X) marries a man who is unaffected (XY). What are the possible genotypes and phenotypes of their children?

Solution:

1. **Mother's Genotype:** X*X (Affected)

2. **Father's Genotype:** XY (Normal)

The possible genotypes of their children are determined by the following Punnett square:

	X	**Y**
X	X	X
*	*	*
	X	Y
X	X	X
	X	Y

Genotypes and Phenotypes:

- **X*X (Affected Female):** 50% chance, female with Rett syndrome.
- **XX (Normal Female):** 25% chance, female with normal development.
- **X*Y (Affected Male):** 25% chance, male with Rett syndrome (though males with Rettsyndrome often do not survive infancy).

Problem 5: Y-Linked Inheritance

Question: A man has a Y-linked trait (for example, a specific Y-linked genetic marker) and marries a woman without this trait. What are the possible genotypes and phenotypes of their children?

Solution:

1. **Mother's Genotype:** XX (Normal, no Y-linked trait)
2. **Father's Genotype:** XY* (Y-linked trait)

The possible genotypes of their children are determined by the following Punnett square:

	X	**X**
X	X	X
	X	X
Y	X	X
*	Y*	Y*

Genotypes and Phenotypes:

- **XX (Normal Female):** 50% chance, female without the Y-linked trait.
- **XY* (Affected Male):** 50% chance, male with the Y-linked trait.

These problems and solutions illustrate the basic principles of sex-linked inheritance and how to determine the potential outcomes for offspring based on the genotypes of the parents.

PRACTICAL 8

Hormonal sex determination in humans involves understanding how hormones influence thedevelopment of sexual characteristics. Here are some problems related to this concept, alongwith their solutions:

Problem 1: Androgen Insensitivity Syndrome (AIS)

Question: A person with an XY karyotype has Androgen Insensitivity Syndrome (AIS),where their body's cells are unable to respond to androgens. What are the expected phenotypic characteristics of this individual?

Solution: Individuals with complete AIS typically have:

External female genitalia.

Undescended testes.

Lack of uterus and fallopian tubes.

Female secondary sexual characteristics at puberty (e.g., breast development) due to theconversion of testosterone to estrogen.

Infertility.

Problem 2: Congenital Adrenal Hyperplasia (CAH)

Question: A person with an XX karyotype has Congenital Adrenal Hyperplasia (CAH), acondition causing excess production of androgens. What are the expected phenotypic characteristics of this individual?

Solution: Individuals with CAH typically have:

Ambiguous genitalia at birth (e.g., an enlarged clitoris or fused labia).

Normal internal female reproductive structures (e.g., ovaries, uterus).

Early onset of puberty.

Potential fertility issues if untreated.

Increased body hair and deepening of the voice in severe cases.

Problem 3: Turner Syndrome

Question: A person with an XO karyotype (Turner Syndrome) lacks a second sexchromosome. What are the expected phenotypic characteristics of this individual?

Solution: Individuals with Turner Syndrome typically have:

Short stature.

Webbed neck.

Broad chest with widely spaced nipples.

Underdeveloped ovaries leading to infertility.

Lack of secondary sexual development without hormone replacement therapy.

Possible congenital heart defects.

Problem 4: Klinefelter Syndrome

Question: A person with an XXY karyotype (Klinefelter Syndrome) has an extra Xchromosome. What are the expected phenotypic characteristics of this individual?

Solution: Individuals with Klinefelter Syndrome typically have:

Taller than average height.

Reduced muscle mass and body hair.

Gynecomastia (development of breast tissue).

Small testes and reduced fertility.

Learning difficulties and speech delay in some cases.

Possible need for testosterone replacement therapy.

Problem 5: 5-Alpha-Reductase Deficiency

Question: A person with an XY karyotype has 5-alpha-reductase deficiency, affecting the conversion of testosterone to dihydrotestosterone (DHT). What are the expected phenotypic characteristics of this individual?

Solution: Individuals with 5-alpha-reductase deficiency typically have:

Ambiguous genitalia at birth (e.g., a small phallus, hypospadias).

Normal male internal reproductive structures (e.g., testes).

At puberty, increased testosterone levels can lead to virilization (e.g., deepening of the voice,growth of facial and body hair).

Potential fertility issues.

Problem 6: SRY Gene Mutation

Question: A person with an XY karyotype has a mutation in the SRY gene, which isresponsible for initiating male sex determination. What are the expected phenotypic characteristics of this individual?

Solution: Individuals with an SRY gene mutation typically have:

Female external genitalia.

Lack of testes and presence of streak gonads (underdeveloped gonadal tissue).

Lack of male secondary sexual characteristics.

Female secondary sexual characteristics at puberty if estrogen is present.

Infertility.

Problem 7: Swyer Syndrome

Question: A person with an XY karyotype has Swyer Syndrome, a condition where the SRYgene is present but non-functional. What are the expected phenotypic characteristics of this individual?

Solution: Individuals with Swyer Syndrome typically have:

Female external genitalia.

Non-functional gonads (streak gonads).

Lack of spontaneous puberty; require hormone replacement therapy for the development ofsecondary sexual characteristics.

Presence of a uterus and fallopian tubes, enabling potential pregnancy with assisted reproductive technology.

Infertility.

Summary

These problems illustrate how various genetic and hormonal factors influence sexual development in humans. Understanding these conditions helps in diagnosing and managingdisorders of sex development (DSDs), and highlights the complexity of hormonal sex determination.

Unit – V: Human Genetics

PRACTICAL 9

Study of human karyotypes

PRACTICAL 10

Construction and analysis of PedigreeVirtual labs/demos

Demonstration of Ultrasonography (Virtual lab).

Demonstration of prenatal diagnosis (Virtual lab).

RFERENCE WEB LINKS:

https://www.iitg.ac.in/cseweb/vlab/anthropology/Experiments/Mendels%20law/index

.html

https://learn.genetics.utah.edu/content/labs/

https://virtuallabs.merlot.org/vl_biology.html

https://blog.praxilabs.com/2020/06/30/dna-extraction-virtual-lab/

https://jru.edu.in/studentcorner/lab- manual/agriculture/Fundamentals%20of%20Genetics.pdf

SEMESTER END LAB EXAMINATION

23ZOMAP232: Animal diversity – Biology of Non chordates Offered to B.Sc. Hons Zoology

Semester: III Max.Marks: 35M

Time: 3 Hrs.

I. **Answer the following.** **Max. Marks: 30 Marks**

Q1Create a Punnett Square: K3

Cross the gametes from Parent 1 and Parent 2:

	AB	**ab**	**Ab**	**aB**
AB				
ab				
Ab				
aB				

Analyze the Results:

Count the Genotypes and Phenotypes:

Determine the ratios of the different phenotypes produced.

Compare the results with and without crossing over.

Calculate the frequency of each phenotype and compare how linkage affects the inheritance patterns.

Q2

Use hypothetical or real case studies where blood typing played a role in forensicinvestigations. Perform blood typing tests on simulated crime scene samples and suspect samples. Determine the likelihood of the suspect being the source of the crime scene bloodbased on the blood typing results. K2

Q3

a. A man has a Y-linked trait (for example, a specific Y-linked genetic marker) andmarries a woman without this trait. What are the possible genotypes and phenotypes of theirchildren?. K3

b. A woman affected by Rett syndrome (X*X) marries a man who is unaffected(XY). What are the possible genotypes and phenotypes of their children?

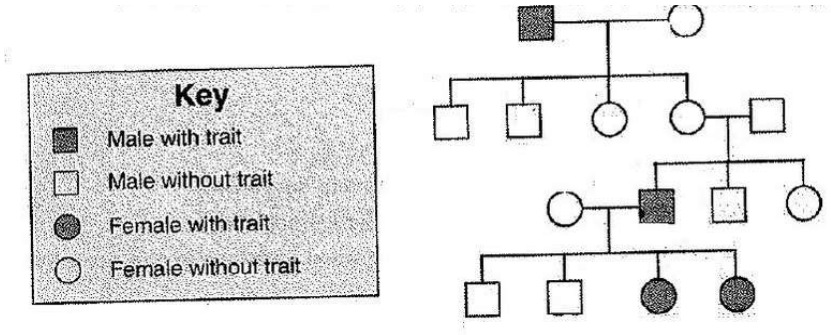
Q4

a. A person with an XXY karyotype has an extra Xchromosome. What is the syndrome with which he is suffering from? What are the expected phenotypic characteristics of this individual?K3

b. A person with an XY karyotype has Androgen Insensitivity Syndrome (AIS),where their body's cells are unable to respond to androgens. What are the expected phenotypic characteristics of this individual?K3

Q5

a. This pedigree shows the inheritance of colour blindness or sex-linked trait. Is this trait dominant or recessive? Is the mother of the colourblind girl in the F3 generation colourblind, a carrier or a person with normal colour vision? Explain.K3



b. What do you mean by Swyer Syndrome? How is it caused? List out the phenotypic characteristics.

II	**Viva**	**3 Marks**
III	**Record**	**2 Marks**

(B) CONTINUOUS ASSESMENT(Internal) 15 MARKS

15 marks for the continuous assessment (Day to day work in the laboratory shall be evaluated for 15 marks by the concerned laboratory teacher based on the regularity/ record/viva). Laboratory teachers are mandated to ensure that every student completes 80%-90% of the lab assessments.

TOTAL: (A)+(B) = 50 MARKS