Indira Gandhi National
Open University
School of Sciences

BZYCL-138 GENETICS AND EVOLUTIONARY BIOLOGY: LABORATORY

List of Exercises

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GENETICS AND EVOLUTIONARY BIOLOGY: LABORATORY

The lab course on Genetics and Evolutionary Biology is based on the theory course "Genetics and Evolutionary Biology" which is of 4 credits. This course is worth 2 credits. This lab component deals with genetics as well as evolution.

The course comprises one block. In this block there are 13 exercises that are intended to develop your skill to work with hands as well as skill of observation, analysis, statistical measurement and interpretation.

Exercises 1 to 6 are oriented towards Genetics. By simple simulation exercises (Exercises 1 and 2) you will verify Mendel's laws of inheritance i.e., Law of Dominance, Law of segregation and Law of Independent Assortment. You will perform Exercises 3 by constructing linkage maps and test crossovers. In Exercise 4 you will learn to identity the different groups of human chromosomes (♂ and ♀, normal and abnormal) and prepare a karyotype of them. Exercise 5 has been included to help you learn to construct and interpret human pedigree charts. In Exercise 6, you will solve problems in Genetics by application of Probability. Exercises 7 to 13 are related to component of evolution. In Exercise 7, you will appreciate the occurrence of major events during the course of evolution by observing the plaster cast models of fossils of arthropods/reptilians. In Exercise 8 you will observe pictures/charts of homologous and analogous organs and relate them with the evolution. Exercises 9 and 10 will apprise you with phylogeny of horse and correlation between Darwin's finches, beaks and food habits respectively. In Exercises 11, and 12 and 13 you will perform simple simulation exercises to understand the role of natural selection in evolving adaptations and interrelationship of genetic drift and natural selection and their significance in evolution.

We have written the exercises in an interesting and easy way to understand so that you can enjoy your process of learning by doing in the laboratory.

Like all other IGNOU laboratory courses this is an intensive residential exercise requiring one week to complete it. Everyday there will be two laboratory sessions of 4 hours each. So there will be a total of 14 sessions. The first session will be introductory and the remaining 2nd to 13th session will be based on the exercises given in the course. Sessions 1 to 12 will have guided exercises under the supervision of the academic counsellor. The last two sessions i.e., 13 and 14 will be unguided sessions that is the term end examination. In each session you will perform exercises for 3 hours and for the remaining 1 hour you will complete your practical note book. A schedule for laboratory exercises will be given to you in the first session.

You are aware that there is a time constraint as you will have limited access to laboratory work, therefore, you are required not to miss any of the laboratory sessions.

You will be assessed for your performance each day (Guided Experiments) and on the last day you will have the term end examination (Unguided Experiments). This examination will be compulsory for you to pass.

Study Guide

1. Before you enter your laboratory for performing laboratory exercises you should read the theory components of Genetics and Evolutionary Biology course.

2. You should also go through the laboratory manual and underline the important steps given in it.

3. Do not forget to carry laboratory manual and a practical record book for making and recording your observation.

Objectives

After completing this course, you should be able to:

- demonstrate Mendel's law of segregation in a monohybrid cross,
- demonstrate Mendel's law of independent assortment in a dihybrid cross,
- describe crossover and recombination processes by solving problems,
- diagnose the chromosomal disorders from the abnormal karyotypes provided,
- analyse and interpret pedigree chart and assign the trait to autosome or sex chromosome and say whether the concerned allele is dominant or recessive,
- apply the formula for the binomial expansion to determine the probability of any combination of events,
- link the geographical distribution of various species of contemporary plants and animals to the course of evolution,
- explain analogous and homologous structures,
- piece together events in development that took place during the evolution of horse,
- analyse the large collection of museum specimens/cut outs of beaks of different birds species,
- make use of simple devices to illustrate the concept of natural selection,
- describe that the alleles for maladaptations continue to occur at very low frequencies in the population, and
- illustrate the concept of genetic drift.

EXERCISE 1

STUDY OF MENDELIAN INHERITANCE AND GENE INTERACTION (NON-MENDELIAN INHERITANCE) USING SUITABLE EXAMPLES AND VERIFYING THE RESULTS USING CHI-SQUARE ANALYSIS (MONOHYBRID MENDELIAN RATIO-I)

Structure

1.1 INTRODUCTION

In Unit 1 of core course-II on Genetics and Evolutionary Biology (BZYCT-137) you have studied Mendel's laws of Inheritance i.e., the law of segregation, and the law of independent assortment. This and the following laboratory exercise pertain to these two laws respectively. Before, you begin this exercise, you try to, recapitulate briefly the law of segregation. You may recall that Mendel used varieties or lines of garden pea (*Pisum sativum*), that exhibit pairs of contrasting characters, e.g., the long stem lines (or the tall plants) and the short stem lines (or the short plants) as parents (P generation) in his crosses. Each line was pure breeding, so that plants from that line always bred true for the character being studied, i.e., tall × tall produced only tall, progeny. When

two lines with contrasting characters are crossed (e.g., tall × short) to produce a first filial generation, all the progeny are of one phenotype, which is the same as one of the parents (e.g., tall). This character is said to be *dominant,* and the character that does not appear in the F_1 (e.g., short) is called *recessive*. There has been no blending of the two characters. When the F_1 is allowed to self, the progeny in the next generation (F_2) will be in the ratio of approximately 3 dominant to 1 recessive phenotypes (e.g., 3 tall : 1 short). What were his conclusions from the above experiments?

(i) Each parent contains two unit factors (genes) of which one is contributed to each member of the F_1 progeny.

(ii) Each gene could exist in two alternate forms or *alleles,* one of which is for the dominant character (i.e., T) and determines the phenotype of the F_1 while the other being for the recessive character (i.e., t).

(iii) Each member of the F_1 contains one of each allele (Tt) and is heterozygous, whereas each parent contains two identical alleles (**TT** in the tall parent, **tt** in the short parent) and is homozygous.

(iv) Thus, the dominant phenotype (tall) results from two different genotypes, the homozygous one (TT) or the heterozygous one (Tt), whereas the recessive phenotype is only determined by one homozygous genotype (tt).

(v) When the F_1 plants produce pollen and eggs, they are clearly of two types, occurring with equal frequency and containing either one allele for the dominant character (T) or one allele for the recessive character (t), i.e., the two alleles in the F_1 get segregated clearly from each other when gametes are formed.

(vi) The pollen and egg nuclei of the two genotypes T and t fuse at random to produce the F_2 zygotes. Thus, the F_2 genotypes would be of three types in the ratio of 1 homozygous for alleles for the dominant character : 2 heterozygous : 1 homozygous for alleles for the recessive character (i.e., 1TT : 2Tt : 1tt). This gives the F_2 phenotypic ratio of 3 dominant : 1 recessive (3 tall : 1 short). Let us now proceed to conduct this exercise.

Objectives

After doing this laboratory exercise you should be able to:

- demonstrate Mendel's law of segregation in a monohybrid cross,
- correlate the experimental steps with the natural processes occurring; and
- analyse the ratio obtained with regard to its goodness of fit, by using Chi-Square test.

1.2 MATERIAL REQUIRED

1) 3 containers/250 ml plastic beakers
2) 50 red beads
3) 50 yellow beads

1.3 PROCEDURE

You will have to work in pairs for this exercise.

Step 1 : Place 50 red beads in one container to represent the gametes of a tall parent (T). Place 50 yellow beads in the other container to represent the gametes of a dwarf parent (t). We assume that both the gametes are from parents that breed true for the characteristic, namely, stem height, whose inheritance is being studied in this experiment.

Step 2 : Withdraw a bead from each container. Each bead withdrawn represents a gamete containing a single pair of allele. Place the beads together, this represents the process of fertilization, by which the paired alleles of gene in the offspring are re-established.

Step 3 : Just as in Step 2, continue to withdraw pairs of beads as above, and arrange on the table.

What would be the genotype of the individuals of the F_1 generation?

.....

Step 4 : To simulate the gametes of this F_1 generation, place 50 beads (25 red and 25 yellow) in each container. One container represents the female gametes, and the other represents the male gametes produced by the F_1 generation (Fig. 1.1 a, b, c).

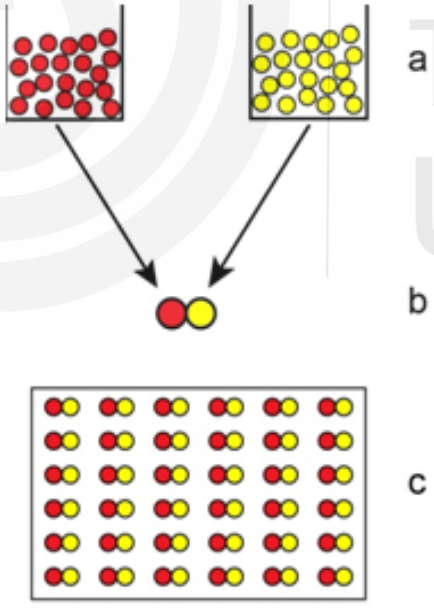


Fig. 1.1: a, b and c.

Step 5 : Shake each container vigourously for 30 seconds, taking care that beads do not fall off.

Step 6 : To produce the F_2 generation, withdraw one bead from each container with your eyes closed and place them together. Your partner should note the combination of genes obtained.

This represents the genotype of F_2 individuals.

Step 7 : After noting the genotype of each pair, discard the pair of beads into a spare container.

Step 8 : Repeat steps 6 and 7 until all beads have been paired and their combinations noted.

Step 9 : Calculate the ratio of the **phenotypes** of the F_2 individuals.

Step 10 : Record the ratios obtained by other groups in your batch and calculate the average ratio (Fig. 1.1d, e, f).

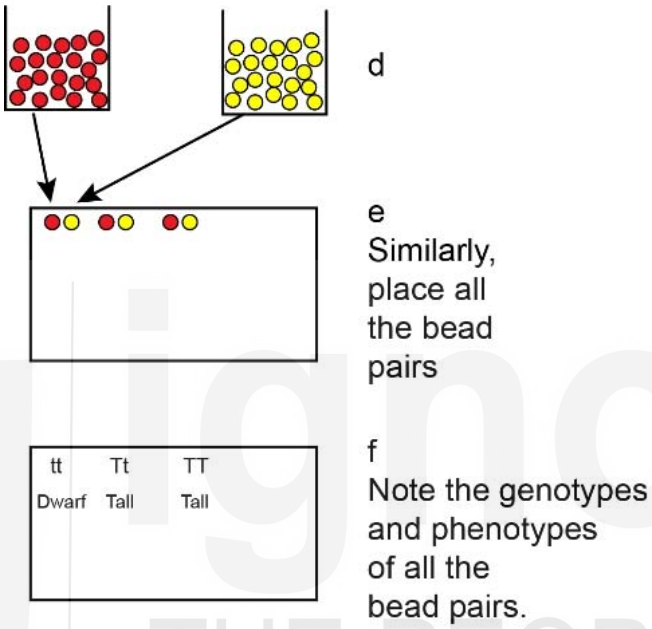


Fig. 1.1: d, e and f.

1.4 OBSERVATIONS AND RESULTS

A. F_1 Generation

a) Total number of F_1 individuals ……

b) Phenotypes of F_1 individuals ……

c) Genotypes of F_1 individuals ……

B. F_2 Generation

a) Total number of F_2 individuals ……

b) Phenotypes of F_2 individuals ……

c) Number of individuals in each phenotypic class ……

d) The phenotypic ratio ……

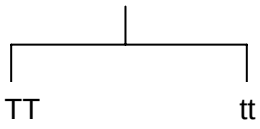
e) Genotypes of F_2 individuals ……

f) Number of individuals in each genotypic class ……

g) The genotypic ratio ……

Group Average

Phenotypic classes



Group 1	……..	……..
Group 2	……..	……..
Group 3	……..	……..
……..	……..	……..
……..	……..	……..
Group n	……..	……..
Total		
Average	……..	……..

1.5 DISCUSSION OF RESULTS

1) In the above exercise which character was dominant?

……
……

2) Was the F_1 homozygous or heterozygous?

……
……

3) When F_1 plants were self-pollinated how many different types of pollen and egg nuclei were produced? What were their genotypes and what were the frequencies of the genotypes?

……
……

4) Why were the beads shaken in step 5, and withdrawn with closed eyes in step 6?

……
……

5) How does your ratio, and the group average ratio compare with Mendel's prediction? Explain any differences.

……
……

6) Make a Punnett square to show the outcome of $F_1 \times F_1$ cross.

……
……

7) What would be the monohybrid test cross of the example taken in this experiment?

……
……

8) What would be the back cross for the example taken in this experiment?

……
……

9) Can you record and present the results in different manner?

……
……

10) Explain how this laboratory exercise acts as a model for breeding and inheritance in pea?

……
……

11) Supposing a random sample of F_2 seeds obtained were sown and their mature plants were allowed to self pollinate. What proportion of plants would produce (i) only tall plants (homozygous), (ii) only dwarf plants (homozygous) and (iii) a mixture of tall and dwarf (heterozygous) plants?

……
……

1.6 χ^2 ANALYSIS

By using the χ^2 (Greek letter, chi pronounced as 'kye squared') test, you can evaluate your results of this experiment. This is a statistical test that is frequently used to determine whether the data obtained experimentally provides a 'good fit', or approximation to the expected or theoretical data. Basically this test can be used to determine whether any deviations from the expected values are due to chance. Chance alone can cause the actual observed ratio to vary from the calculated ratio for a genetic cross. For example, for a monohybrid cross you would get very rarely an exact ratio of 3 : 1. The observed results differ, but there comes a point when the difference is so great that the observed data are not in conformity with the expected one. The chi-square test indicates this point. In this exercise, for example, the hypothesis is that the F_2 data do not differ significantly from a 3 : 1 ratio. This type of formulation is called **null hypothesis,** because we will test that nothing has happened to disturb the expected ratio significantly. Consider an alternative hypothesis, such as the F_2 data do differ significantly from a 3 : 1 ratio because gametes with different genotypes are produced in unequal numbers. Such a hypothesis would be imprecise and would not lead to an exact numerical prediction. From this example it should be clear that the null hypothesis being tested should always be stated for the test.

The formula for this test is:

$$\chi^2 = \sum \frac{(o - e)^2}{e} \text{ or } \sum \frac{d^2}{e}$$

χ^2 = chi-square

$\sum$ = sum of

d = difference between the expected and observed results, often termed as deviation (o – e)

e = expected results

o = observed results

Let us see how we can apply this formula. As you know, in a monohybrid cross a 3 : 1 ratio is expected. Supposing you count a total of 160 plants, out of which 120 are tall and 40 are dwarf. But another student counts 116 tall plants and 44 dwarfs. Then the value for the chi-square test would be calculated as shown in Table 1.1.

Table 1.1: Calculation of the Chi-square Value.

Phenotype	**Observed number (o)**	**Expected number (e)**	**difference (d = o – e)**	**d^2**	**Partial Chi-square d^2/e**
Tall	116	120	4	16	16/120 = .133
Dwarf	44	40	4	16	16/40 = .400
					$\sum \frac{d^2}{e} = .533$ $\chi^2 = .533$

The next step is to look up this Chi-square value (χ^2) in the Table 1.2, that indicates whether the probability (P) is that the differences noted are only due to chance in the form of random sampling error or whether it would be better to explain the results on the basis of a different prediction or hypothesis.

Table 1.2: Critical Values of Chi-square for 1-30 degree of freedom that are equalled with or exceeded with particular probabilities (P). (Figures at the top of the table indicate levels of significance)

	.99	**.98**	**.95**	**.90**	**.80**	**.70**	**.50**	**.30**	**.20**	**.10**	**.05**	**.02**	**.01**	**.001**
1	.00016	.00063	.0039	.016	.046	.15	.46	1.07	1.64	2.71	3.84	5.41	6.64	10.83
2	.02	.04	.10	.21	.45	.71	1.39	2.41	3.22	4.60	5.99	7.82	9.21	13.82
3	.12	.18	.35	.58	1.00	1.42	2.37	3.66	4.64	6.25	7.82	9.84	11.34	16.27

4	.30	.43	.71	1.06	1.65	2.20	3.36	4.88	5.99	7.78	9.49	11.67	13.28	18.46
5	.55	.75	1.14	1.61	2.34	3.00	4.35	6.06	7.29	9.24	11.07	13.39	15.09	20.52
6	.87	1.13	1.64	2.20	3.07	3.83	5.35	7.23	8.56	10.64	12.59	15.03	16.81	22.46
7	1.24	1.56	2.17	2.83	3.82	4.67	6.35	8.38	9.80	12.02	14.07	16.62	18.48	24.32
8	1.65	2.03	2.73	3.49	4.59	5.53	7.34	9.52	11.03	13.36	15.51	18.17	20.09	26.12
9	2.09	2.53	3.32	4.17	5.38	6.39	8.34	10.66	12.24	14.68	16.92	19.68	21.67	29.59
10	2.56	3.06	3.94	4.86	6.18	7.27	9.34	11.78	13.44	15.99	18.31	21.16	23.21	29.59
11	3.05	3.61	4.58	5.58	6.99	8.15	10.34	12.90	14.63	17.28	19.68	22.62	24.72	31.26
12	3.57	4.18	5.23	6.30	7.81	9.03	11.34	14.01	15.81	18.55	21.03	24.05	26.22	32.91
13	4.11	4.76	5.89	7.04	8.63	9.93	12.34	15.12	16.89	19.81	22.36	25.47	29.69	34.53
14	4.66	5.37	6.57	7.79	9.47	10.82	13.34	14.22	18.15	21.06	23.68	26.87	29.14	36.12
15	5.23	5.98	7.26	8.55	10.31	11.72	1434	17.32	19.31	22.31	25.00	28.26	30.58	37.70
16	5.81	6.61	7.96	9.31	11.15	12.62	15.34	18.42	20.42	23.54	26.30	29.63	32.00	39.29
17	6.41	7.26	8.67	10.08	12.00	13.63	16.34	19.51	21.62	24.37	27.59	31.00	33.41	40.75
18	7.02	7.91	9.39	10.86	12.86	14.4	17.34	20.60	22.76	25.99	28.87	32.35	34.80	42.31
19	7.63	8.57	10.12	11.65	13.72	15.35	18.34	21.69	23.90	27.20	30.14	33.69	36.19	43.82
20	8.26	9.24	10.85	12.44	14.58	16.27	19.34	22.78	25.04	28.41	31.41	35.02	37.57	45.32
21	8.90	9.92	11.59	13.24	15.44	17.18	20.34	23.86	26.17	29.62	32.67	36.34	38.93	46.80
22	9.54	10.60	12.34	10.04	16.31	18.10	21.34	34.04	27.30	31.81	33.92	37.66	40.29	48.27
23	10.20	11.29	13.09	14.85	17.19	19.02	22.34	26.02	28.43	32.01	35.17	38.97	41.64	49.73
24	10.86	11.90	13.85	15.66	18.06	19.94	23.34	27.10	29.55	33.20	36.42	40.27	42.38	51.18
25	11.52	12.70	14.61	16.47	18.94	20.87	24.34	28.17	30.68	34.38	37.65	41.57	44.21	52.62
26	12.20	13.41	15.38	17.29	19.82	21.79	25.34	29.25	31.80	35.56	38.88	42.86	45.64	54.06
27	12.88	14.12	16.15	18.11	20.70	22.72	26.34	30.32	32.91	36.74	40.11	44.14	46.96	55.48
28	13.56	14.85	16.93	18.94	21.59	23.65	27.34	31.39	34.03	37.93	41.34	45.42	48.28	56.89
29	14.26	15.57	17.71	19.77	22.48	24.58	28.34	32.46	35.14	39.09	42.56	46.69	49.53	58.30
30	14.95	16.31	18.49	20.60	23.36	25.51	29.34	33.53	36.25	40.26	43.77	47.96	50.89	59.70

In Table 1.2, the notation df refers to the degree of freedom, which in this experiment would be determined by the number of phenotypic traits studied. In our example, we have two classes, tall and dwarf plants. As indicated in the table for the value of df, i.e., we need to know the value of C-1. You might have followed that the degree of freedom (df) is calculated by using the

formula C-1, where C is the total number of classes. In this case C is 2 X-X therefore, df is equal to 1 (i.e., 2 – 1 = 1). Therefore, you should look for χ^2 value in the first row (i.e., in 1), of the Table 1.2. The value .533 lies between 0.50 and .30 probability values. This means that by random chance, this difference between the actual count and the expected count would occur between 30 and 50% of the time. In biology, it is generally accepted that a P value greater than 0.05 is acceptable, while a P value lower than 0.05 would indicate that the results cannot be due to random sampling and, therefore, do not fit the original prediction (hypothesis).

A) Do a Chi-square analysis of your results by filling in Table 1.3.

Table 1.3: Chi-Square Analysis of Results.

Phenotype	**Observed number (o)**	**Expected number (e)**	**difference (d = o – e)**	**d^2**	**Partial Chi-square d^2/e**
					$\sum \frac{(d)^2}{e} =$

χ^2 = ……..

C-1 = ……..

P (from Table 1.2) = …….

Do your results support Mendel's prediction? ……

If no, can you account for this? ……

B) Do a Chi-square analysis for your entire batch.

Phenotype	**Observed Numbers**					
	Group 1	**Group 2**	**Group 3**	**----------**	**Group n**	**Total**
Tall						
Dwarf						

Total no. of Individuals …….

Chi-square analysis

Phenotype	**Observed number (o)**	**Expected number (e)**	**difference (d = o – e)**	**d^2**	**Partial Chi-square d^2/e**
Tall					
Dwarf					
					$\sum \frac{(d)^2}{e} =$

χ^2 = …………..

C-1 = …………..

P (from Table 1.2) = ……………

Do these results support Mendel's prediction?

………………………………………………………………………………………………
………………………………………………………………………………………………

Comment on the above results.

………………………………………………………………………………………………
………………………………………………………………………………………………

What are the advantages of taking a bigger sample?

………………………………………………………………………………………………
………………………………………………………………………………………………

EXERCISE 2

STUDY OF MENDELIAN INHERITANCE AND GENE INTERACTION (NON-MENDELIAN INHERITANCE) USING SUITABLE EXAMPLES AND VERIFYING THE RESULTS USING CHI-SQUARE ANALYSIS (DIHYBRID MENDELIAN RATIO-II)

Structure

2.1 INTRODUCTION

This exercise pertains to Mendel's second law of inheritance, that is, the law of independent assortment. Mendel, by choosing plants with two pairs of contrasting characters provided evidence that the alternate forms of factors assort independently and recombine to give a dihybrid ratio 9 : 3 : 3 : 1. Alternatively, segregation of one gene pair is independent of the segregation of the other gene pair, and will give a similar ratio (3 : 1 × 3 : 1). In this experiment, you will learn to demonstrate a Mendelian dihybrid ratio using coloured beads and test the significance with the statistical procedure – the Chi-square test.

Unit 1 of course on Genetics and Evolutionary Biology is to be studied thoroughly prior to undertaking this exercise.

Objectives

After doing this exercise you should be able to:

- demonstrate Mendel's law of independent assortment in a dihybrid cross,
- correlate the experimental steps with the natural processes occurring, and
- analyse the ratio obtained for its goodness of fit, using Chi-square test.

2.2 MATERIAL REQUIRED

1) 5 Containers
2) 48 Green Beads
3) 48 Yellow Beads
4) 48 Black Beads
5) 48 White Beads
6) 1 Packet Modelling Clay

2.3 PROCEDURE

You will have to work in pairs for doing this exercise.

Step 1 : Place 48 beads of red, yellow, black and white colours separately in four containers.

Black beads represent the dominant trait, smooth seed coat (S); and

White beads represent the recessive trait, wrinkled seed coat (s).

Yellow beads denote the dominant character, yellow colour of seeds (Y); and

Green beads denote the recessive character, green colour of seeds (y).

Step 2 : Make a cross between parents with phenotypes smooth, yellow and wrinkled, green.

Step 3 : What kind of gametes would be produced by each parent?

.....

.....

Step 4 : Place one bead each of white, green, black and yellow colour together. This process represents fertilisation. Note the phenotype and genotype of F_1 individual.

.....

.....

Step 5 : Next step is to make a cross between the F_1 individuals. From the F_1 individuals obtained above what kind of gametes can be formed?

.....
.....

To simulate the gametes of this F_1 generation, place 24 beads (24 white, 24 black, 24 green and 24 yellow) in each of two containers. One container represents the female gametes and the other represents male gametes produced by the F_1 parents.

Step 6 : In order to make the identification of the four kinds of gametes clear, use a small ball of modelling clay to join the respective beads. For example, take one white and one green bead and join them firmly with a small amount of modelling clay. Similarly make pairs of one white, one yellow bead; a black and a green bead and a black and a yellow bead. Thus, make pair of all the beads of both the containers (Fig. 2.1 a, b, c, d).

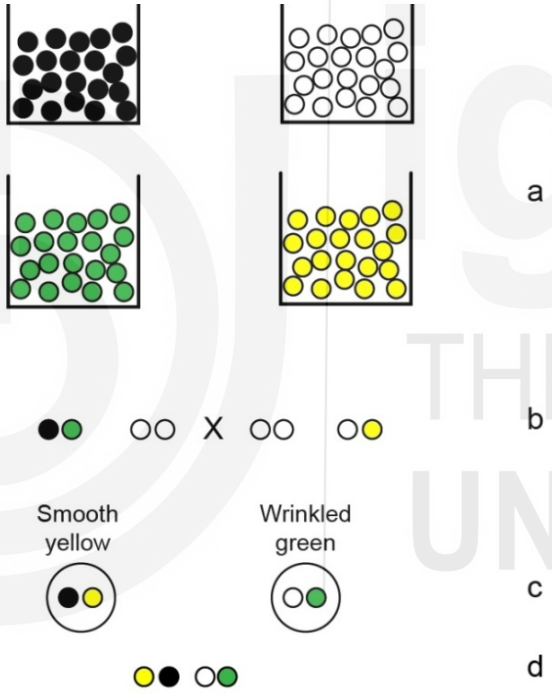


Fig. 2.1: a, b, c and d.

Step 7 : Shake both the containers (each having four kinds of gametes) for 30 seconds.

Step 8 : To produce the F_2 generation, withdraw a bead pair from each container **with your eyes closed,** and place them together. Your partner should note the combination of genes obtained. This represents the phenotypes and genotypes of an F_2 individual.

Step 9 : After noting the phenotype and genotype of each pair, discard the pair of beads into a spare container.

Step 10 : Repeat steps 8 and 9 till all the pairs of beads of both the containers have been utilised and their combinations noted.

Step 11 : Calculate the ratio of the **phenotypes** of the F_2 individuals.

Step 12 : Record the ratios obtained by other groups in your batch and calculate the average ratio.

Step 13 : Analyse your results as well as the group average ratio separately for their goodness of fit by using Chi-square method that you have used in the previous experiment (Fig. 2.1 e, f, g, h).

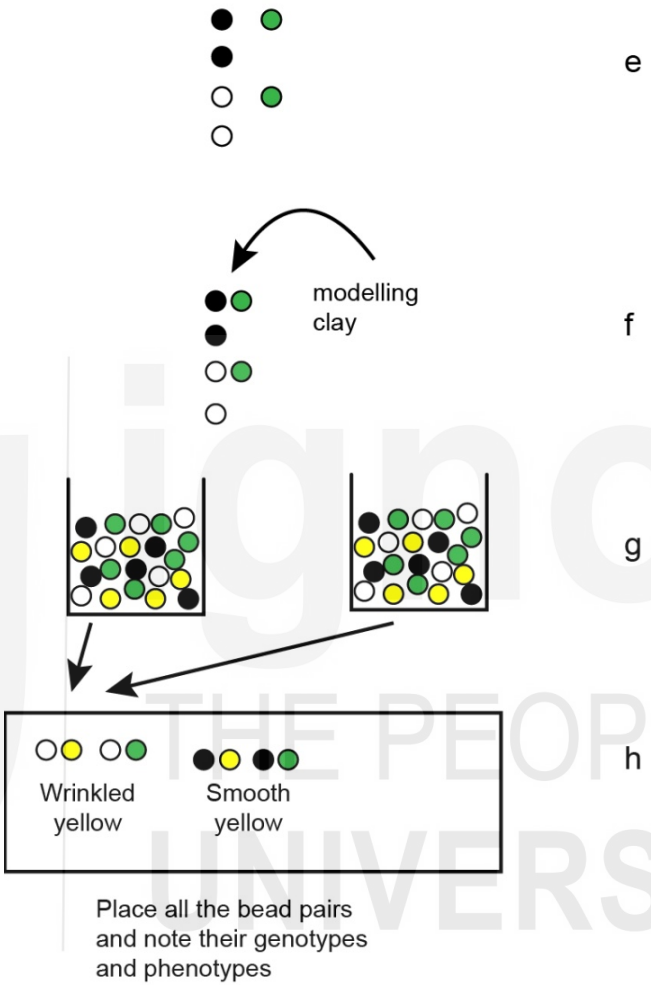


Fig. 2.1: e, f, g and h.

2.4 OBSERVATIONS AND RESULTS

I. Individual Observations

F_1 Generation

a) Total number of individuals ……

b) Phenotypes ……

c) Genotypes ……

F_2 Generation

a) Total number of individuals ……

b) Phenotypes ……

c) Number of individuals in each phenotypic class ……

d) The phenotypic ratio ……

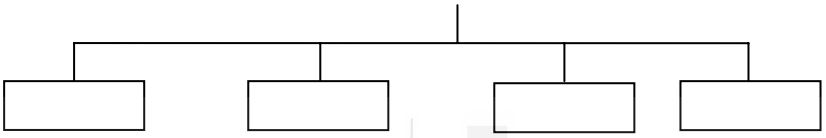
e) Genotypes ……

f) Number of individuals in each genotypic class ……

g) The genotypic ratio ……

II. Group Average

Number of individuals in the phenotypic classes



Group 1	……..	……..	……..	……..
Group 2	……..	……..	……..	……..
Group 3	……..	……..	……..	……..
Group 4	……..	……..	……..	……..
Total				
Average	……..	……..	……..	……..

2.5 DISCUSSION OF RESULTS

1) Compare your ratio and the group average ratio with Mendel's prediction. Explain differences.

……

2) Make Punnett square or Branch diagram to show the outcome of $F_1 \times F_2$ cross.

……

3) What would be the dihybrid test cross of the example taken in this exercise?

……

4) Can you record and present your results in another manner?

……

5) Do Chi-square analysis of your results.

Phenotype	Observed number (o)	Expected number (e)	difference (d = o – e)	d^2	Partial Chi-square d^2/e
					$\sum \frac{(d)^2}{e} = \chi^2$

EXERCISE 3

LINKAGE, RECOMBINATION AND GENE MAPPING

Structure

3.1 INTRODUCTION

Before you undertake this exercise you are supposed to study Units 4 and 5 of Genetics and Evolutionary Biology course (BZYCT-137). When two or more genes reside on the same chromosome, they are said to be linked. They may be linked together on one of the autosomes or connected together on the sex chromosome. Genes on different chromosomes are distributed into gametes independently of one another (Mendel's Law of Independent Assortment). Genes on the same chromosome, however, tend to stay together during the formation of gametes. Thus, the results of test crossing dihybrid individuals will yield different results, depending upon whether the genes are linked on same or on different chromosomes.

Objectives

After doing this exercise, you should be able to:

- explain the concept of linkage by solving problems,
- describe crossover and recombination processes by solving problems, and
- construct linkage maps and test crossovers.

3.2 LINKAGE

When two or more genes reside in the same chromosome, they are said to be linked and their transmission pattern is called linkage. They may be linked together on one of the autosomes as connected together on the sex chromosomes.

EXAMPLE 1

Linkage between two genes which leads to zero percent recombination is said to be completely linked.

Genes on different chromosomes assort independently during meiosis, giving a 1:1:1:1 testcross ratio.

Parents: *AaBb* × *aabb*

Gametes: *AB* *Ab* *aB* *ab* *ab*

F_1: 1/4 *AaBb* : 1/4 *Aabb* : 1/4 *aaBb* : 1/4 *aabb*

EXAMPLE 2

Synaptonemal complex (SC) is a protein structure that is formed between homologous chromosomes (two pairs of sister chromatids) during meiosis and is thought to mediate synapsis and recombination during meiosis I in eucaryotes.

Linked genes do not assort independently, but tend to stay together in the same combinations as they were in the parents. Genes to the left of the slash line (/) are on one chromosome and those to the right are on the homologous chromosome. Very closely linked genes may not recombine in the formation of gametes.

Parents: *Aa/B* × *aa/bb*

Gametes: *AB* *ab* *ab*

F_1: 1/2 *AB/ab* : 1/2 *ab/ab*

Large deviations from a 1 : 1 : 1 : 1 ratio on the testcross progeny of a dihybrid could be used as evidence for linkage. Linked genes do not always stay together, however, because homologous nonsister chromatids may exchange segments of varying length with one another during meiotic prophase. You may recall that homologous chromosomes pair with one another in a process called “synapsis” and that the points of genetic exchange, called “chiasmata,” produce recombinant gametes through crossing over.

If in a single tetrad, only exchange occurs between non-sister chromatids as a result of crossover it is **single crossover** and if more than one then it is **double crossover**. The tendency of one crossing over to interfere in the occurrence of another crossing over in its nearest vicinity is called **chiasma interference**. This phenomenon was first observed by M.J. Muller.

3.3 CROSSING OVER

In preparation for meiosis, the DNA of each chromosome replicates, producing two genetically identical (barring mutation) sister chromatids. During prophase I, homologous chromosomes form pairs called **synapses** [Fig. 3.1(*a*)] with the aid of proteins in the synaptonemal complex. Very large protein complexes, called **recombination modules** [about 90 nanometers (nm) in diameter], occur at intervals along the synaptonemal complex; each of these recombination modules is thought to function as a multienzyme “**recombination machine**” that affects synapsis and recombination. A **nick** is the removal of a phosphodiester bond between adjacent nucleotides in a DNA strand. Endonucleases in the recombination modules nick a single strand of each chromatid, allowing non-sister strands to be exchanged [Fig. 3.1 (*b*)], and thus affecting the recombination of linked genes. A DNA polymerase may extend the exchanged strands, and an enzyme called DNA ligase repairs the nicks [Fig. 3.1(*c*)]. If the top chromatid strand is rotated by 180°, a cross-shaped structure called a **chi (χ) form** can be seen under the microscope.

This structure is also referred to as the **Holliday model** after R. Holliday who proposed it in 1964 [Fig. 3.1 (*d*)]. An endonuclease-nicks the two previously uncut strands at tetranucleotide sequences 5′-(A/T) TT(G/C)-3′. Gaps and nicks are then repaired, creating four recombinant chromatids [Fig. 3.1 (*e*)] which will segregate during the second meiotic division to be incorporated into different gametes. Note that if only *A* and *B* loci are being studied in the progeny of dihybrid parents (*AB*/*ab*), two of the four possible gametes will retain the linkage relationships of the dihybrid parents (*AB* and *ab*)and are thus referred to as **parental** or **non-crossover types;** the two other gametes will be **recombinant** or **crossover types** (*AB* and *aB*)]. Thus, each crossover or chiasma event is expected to produce four gametes (*AB, Ab, aB, ab*) with equal frequencies. However, if a crossover between the two genes under study does not occur in every meiosis, then among all of the gametes (both those with and without crossovers in this region) produced by a dihybrid individual, the frequency of non-crossover-type gametes will exceed that of crossover-type gametes.

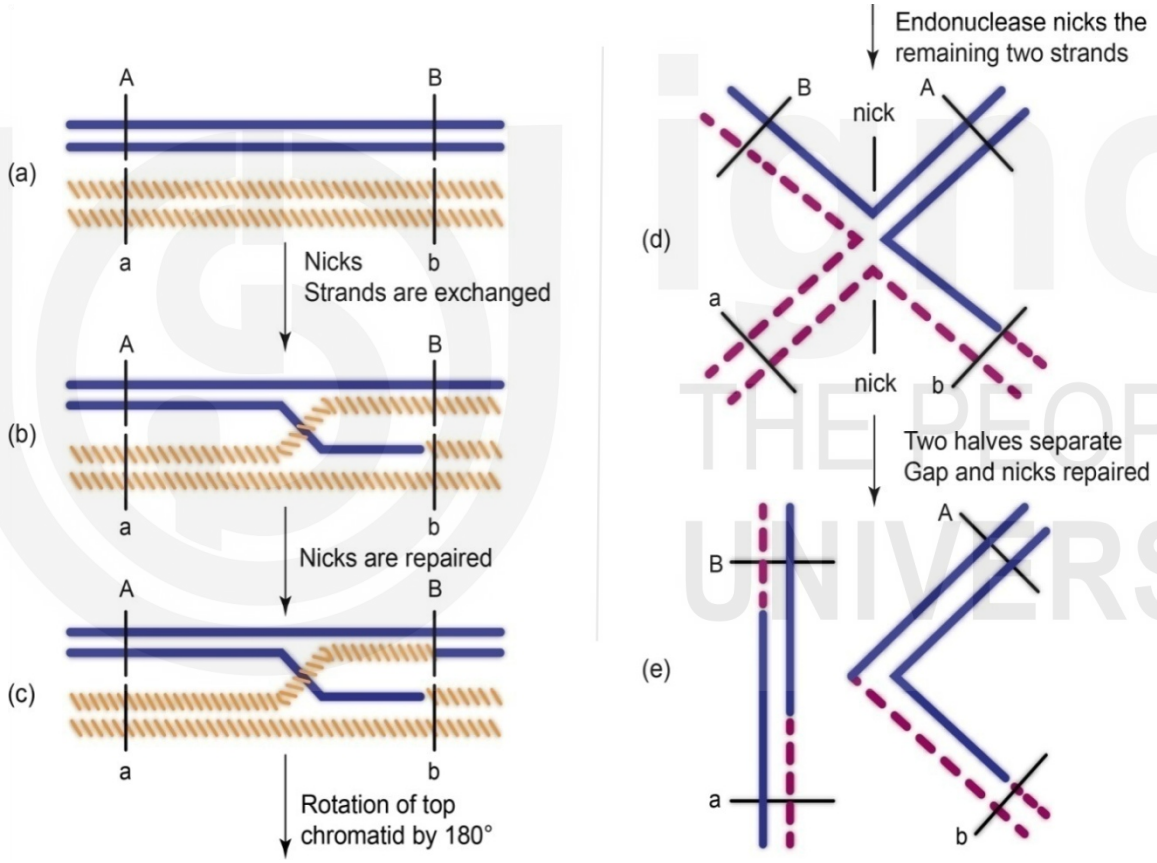


Fig. 3.1: General recombination and the formation of a Holliday intermediate. (*a*) Two homologous chromatids synapse. (*b*) A strand of each homologous chromatid is nicked and exchanged. (*c*) DNA synthesis may extend the exchanged strands subsequent to DNA nick repair. (*d*) Branch migration and formation of the Holliday intermediated. (*e*) The nicking of the uncut strands of the chromatids creates separate recombined chromatids. Nicks in the recombined chromatids are repaired by a ligase enzyme.

The alleles of double heterozygotes (dihybrids) at two linked loci may appear in either of two positions relative to one another. If the two dominant (or wild-type) alleles are on one chromosome and the two recessives (or mutants) on the other (*AB*/*ab*), the linkage relationship is called **coupling phase.** When the dominant alleles of one locus and the recessive allele of the other occupy the

same chromosome (*Ab*/*aB*) the relationship is termed **repulsion phase.** Parental and recombinant gametes will be of different types, depending upon how these genes are linked in the parent.

EXAMPLE 3

Coupling Parent: *Aa/Bb*

Gametes: Parental:

Recombinant: *AB* *ab* *Ab* *aB*

EXAMPLE 4

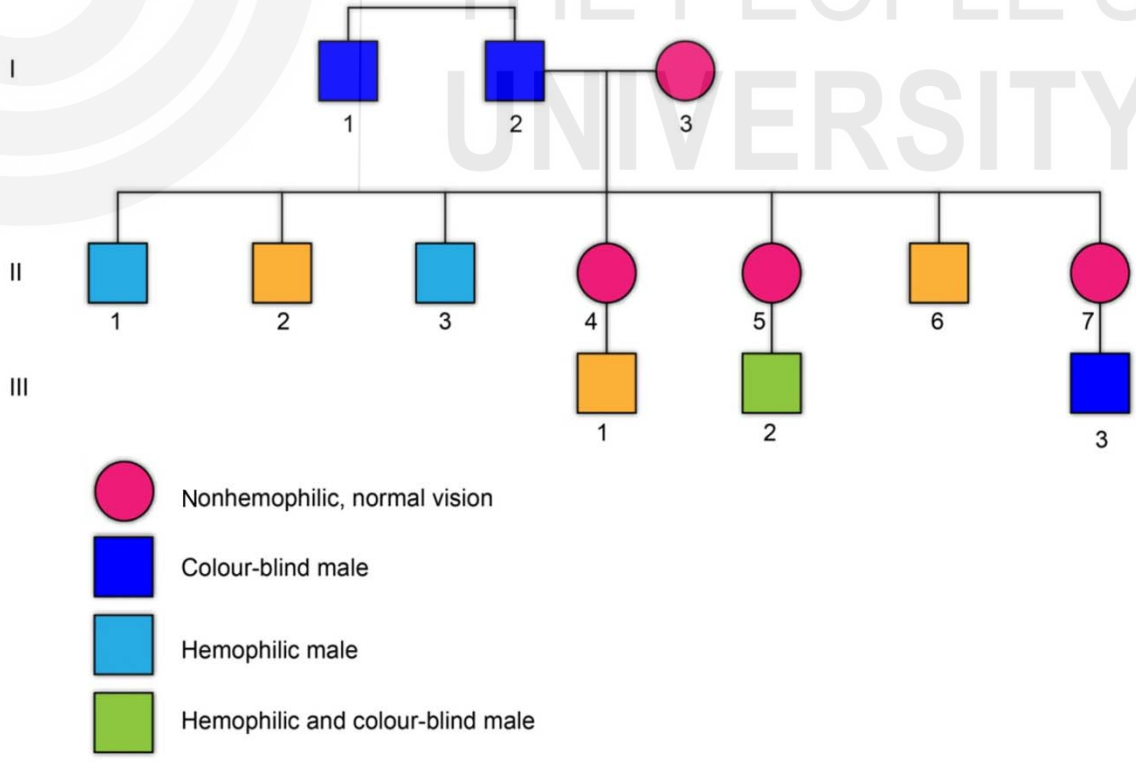
Repulsion Parent: *Ab/aB*

Gametes: Noncrossover: *Ab* *aB*

Croossover: *AB* *ab*

Problem 3.1

In the human pedigree below where the male parent does not appear, it is assumed that he is phenotypically normal. Both hemophilia (*h*) and colour blindness (*c*) are sex-linked recessive traits. Determine the genotypes for each individual in the pedigree.



The diagram shows the inheritiance of colorblindness in a family colorblindness is a recessive and X-linked trait. The allele for normal vision is dominant.

In generation I, neither parent has the trait, but one of their children (II-3) is colorblind. Because there are unaffected parents that have an affected offspring, the trait may be assumed to be recessive. In addition, the trait appears to affect males more than females suggesting that the trait may be X-linked.

Solution

The linkage relationship of the males' genes on their single X chromosome is obvious from their phenotype. Thus, I1, I2, and III3 are all hemophilic with normal colour vision and According to legend, these males are color-blind and not hemophilic therefore must be *hC/Y*. Nonhemophilic, colour-blind males II1 and II3 must be *Hc*/Y. Normal males II2, II6, III1 must possess both dominant *HC*/Y. III2 is both hemophilic and colour blind and therefore must possess both recessives *hc/Y*. Now let us determine the female genotypes. I3 is normal produces sons, half of which are colour blind and half normal. The X chromosome contributed by I3 to her colour-blind sons II1 and II3 must have been *HC*; the X chromosome she contributed to her normal sons II2 and II6 must have been *HC*. Therefore, the genotype for I3 is *HC/HC*.

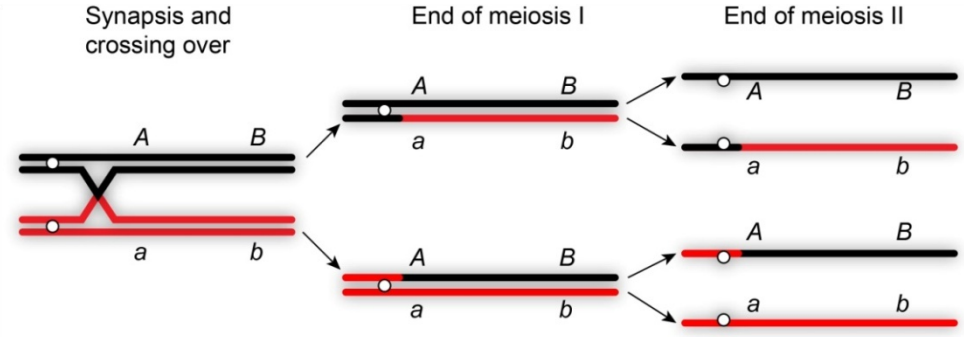
Normal females II2, II5, and II7 each receive *hC* from their father (I2), but could have received either *Hc* or *HC* on the X chromosomes they received from their mother (I3). II4 has a normal son (III1) to which she gives HC; therefore, II2 is probable *hC/HC,* although it is possible for II4 to be *hC/Hc* and produced an *HC* gamete by crossing over. II5, however, could not be hC/HC and produce a son with both hemophilia and colour blindness (III2); therefore, II5 must be *hC/Hc* in order to give the crossover gamete *hc* to her son.

3.4 CHIASMA FREQUENCY

A pair of synapsed chromosomes (bivalent) consists of four chromatids called a tetrad. Every tetrad usually experiences at least one chiasma somewhere along its length. Generally speaking, the longer the chromosome, the greater the number of chiasmata. Each type of chromosome within a species has characteristic (or average) number of chiasmata. The frequency with which a chiasma occurs between any two genetic loci also has a characteristic or average probability. **The further apart two genes are located on chromosome, the greater the opportunity for a chiasma to occur between them. The closer two genes are linked, the smaller the chance for a chiasma occurring between them.** These chiasmata probabilities are useful in predicting the proportions of parental and recombinant gametes expected to be formed from a given genotype. The percentage of cross over (recombinant) gametes formed by a given genotype is a direct reflection of the frequency with which a chiasma forms between the genes in question. Only when a crossover forms between the gene loci under consideration will recombination be detected.

EXAMPLE 5

Crossing over outside the *A-B* region fails to recombine these markers.



When a chiasma forms between two gene loci, only half of the meiotic products will be of crossover type. Therefore, chiasma frequency is twice the frequency of crossover products.

Chiasma % = 2X (crossover %) or Crossover % = I/2 (chiasma %)

EXAMPLE 6

If a chiasma forms between the loci of genes *A* and *B* in 30% of the tetrads of an individual of genotypes *AB/ab*, then 15% of the gametes will be recombinant (*Ab* or *aB*) and 85% will be parental (*AB* or *ab*).

EXAMPLE 7

Suppose progeny from the testcross $\mathrm{Ab/ab} \times \mathrm{ab/ab}$ found in the proportions 40% *Ab*/*ab*, 40% *Ab*/*ab,* 10% *Ab*/*ab,* and 10% *ab*/*ab*. The genotypes *Ab*/*ab* and *ab*/*ab* were produced from crossover gametes. Thus, 20% of all gametes formed by the dihybrid parent were crossover types. This means that a chiasma occurs between these two loci in 40% of all tetrads.

3.5 MULTIPLE CROSSOVERS

When two-strand double crossovers occur between two genetic markers, the products, as detected through the progeny phenotypes, are only parental types [Fig. 3.2 (a)]. However, a third gene locus c between the outside markers allows detection of double crossovers [Fig. 3.2 (b)]

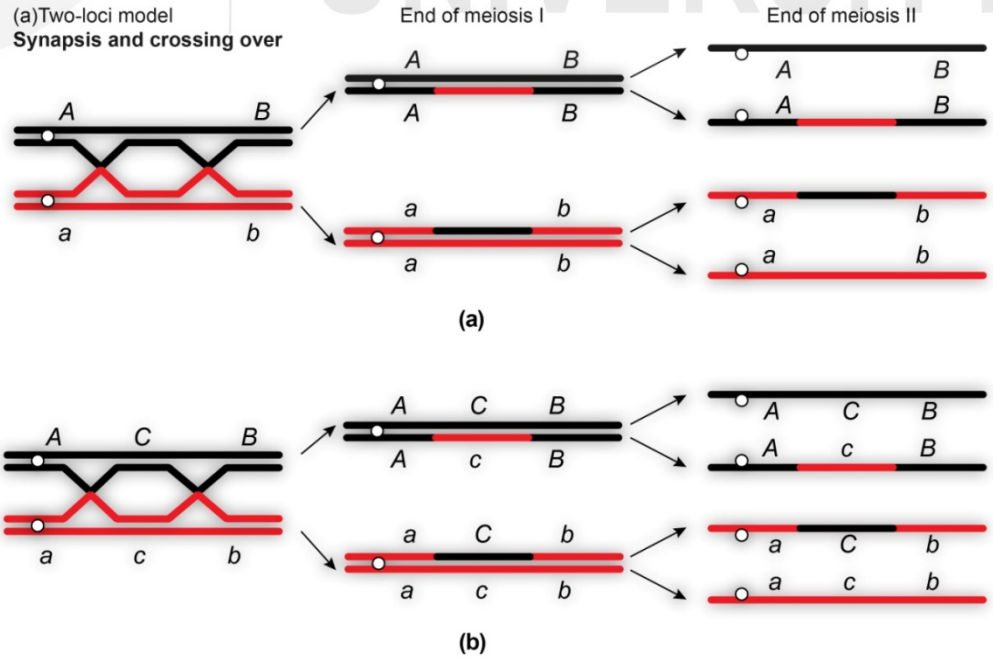


Fig. 3.2: Multiple crossovers with two loci segregating (a) and three loci segregating (b).

If there is a certain probability that a crossover will form between the *A* and *C* loci and another independent probability of a crossover forming between the *C* and *B* loci, then the probability of a double crossover is the product of the two independent probabilities.

EXAMPLE 8

If a crossover between the *A* and *C* loci occurs in 20% of the tetrads and between *C* and *B* loci in 10% of the tetrads in an individual of genotype *ACB*/*acb*, then 2% (0.2 × 0.1) of the gametes are expected to be of double-crossover types *AcB* and *aCb*.

Odd numbers of two-strand crossovers (one, three, five, etc.) between two gene loci produce detectable recombinations between the outer markers, but even numbers of two-strand crossovers (two, four, six, etc.) do not.

3.6 LIMITS OF RECOMBINATION

If two gene loci are so far apart in the chromosome that the probability of a chiasma forming between them is 100%, then 50% of the gametes will be parental type (non-crossover) and 50% recombination (crossover) type. When such dihybrid individuals are test-crossed, they are expected to produce progeny in a 1 : 1 : 1 : 1 ratio as would be expected for genes on different chromosomes. Recombination between two linked genes cannot exceed 50% even when multiple crossovers occur between them.

GENETIC MAPPING

1. Map Distance

The places where genes reside in the chromosome (loci) are positioned in linear order analogous to beads on a string. There are two major aspects to genetic mapping: (1) the determination of the linear order with which the genetic units are arranged with respect to one another (gene order) and (2) the determination of the relative distances between the genetic units (gene distance). The unit of distance that has the greatest utility in predicting the outcome of certain types of matings is an expression of the probability that crossing over will occur between the two genes under consideration. **One unit of map distance, is therefore, equivalent to 1% crossing over. Map Units are often referred to as Centi Morgans (cM)** in honour of the work of Thomas Hunt Morgan, a famous *Drosophila* geneticist.

EXAMPLE 9

If the genotype *Ab*/*aB* produce 8% each of the crossover gametes *AB,* and *ab,* then the distance between *A* and *B* is estimated to be 16 map units.

EXAMPLE 10

If the map distance between the loci *B and* C is 12 units, then 12% of the gametes of genotype *BC*/*bc* should be crossover types; i.e., 6% *Bc* and 6% *bC*.

Each chiasma produces 50% crossover products. Fifty percent crossing over is equivalent to 50 map units. If the average (mean) number of chiasmata is known for a chromosome pair, the total length of the map for that linkage group may be predicted:

Total length = mean number of chiasmata × 50

2. Two-Point Testcross

The easiest way to detect crossover gametes in a dihybrid is through the testcross progeny. Suppose we testcross dihybrid individuals in coupling phase (AC/ac) and find in the progeny phenotypes 37% dominant at both loci, 37% recessive at both loci, 13% dominant at the first locus and recessive at the second, and 13% dominant at the second locus and recessive at the first. Obviously, the last two groups (genotypically *Ac*/*ac* and *aC*/*ac*) were produced by crossover gametes from the dihybrid parent. Thus, 26% of all gametes (13+13) were of crossover types and the distance between the loci *A* and *C* is estimated to be 26 map units, or 26 *cM*.

3. Three-Point Testcross

Double crossovers usually do not occur between genes less than 5 map units apart. For genes further apart, it is advisable to use a third marker between the other two in order to detect any double crossovers.

Suppose that we testcross trihybrid individuals of genotype *ABC*/*abc* and find in the progeny the following:

36% *ABC*/*abc*	9% *AbC*/*abc*	4% *ABc*/*abc*	1% *Abc*/*abc*
36%*abc*/abc	9%a*BC*/*abc*	4%*abC*/*abc*	1%*aBc*/*abc*
72% Parental type:	18% Single crossovers between *A* and *B* (region I)	8% Single crossovers: between *B* and *C* (region II)	2% Double crossover

To find the distance *A*-*B* we must count all crossovers (both singles and doubles) that occurred in region I = 18% + 2% = 20% or 20 map units between the loci *A* and *B*. To find the distance *B*-*C* we must count all crossovers (both singles and doubles) that occurred in region II = 8% + 2% = 10% or 10 map units when double crossovers are detected in the two-point linkage experiment above.

Without the middle marker *B,* double crossovers would appear as parental types and hence we underestimate the true map distance (crossover percentage). In this case the 2% double crossovers would appear with the 72% parental types, making a total of 74% parental types and 26% recombination types. Therefore, for any three linked genes whose distances are known, the amount of detectable crossovers (recombinants) between the two outer markers *A* and *C* when the middle marker *B* is missing is (*A*-*B* crossover percentage) plus (*B*-*C* crossover percentage) minus (2 × double-crossover percentage). This procedure is appropriate only if a crossover in the *A*-*B* region occurs independently of that in the *B*-*C* region.

Problem 3.2

A kidney-bean-shaped eye is produced by a recessive gene *k* on the third chromosome of *Drosophila*. Orange eye colour, called “cardinal”, is produced by the recessive gene *cd* on the same chromosome. Homozygous “kidney”, cardinal females are mated to homozygous ebony males. The trihybrid F_1 females are then test crossed to produce the F_2. Among 4000 F_2 progeny are the following:

1761 kidney, cardinal	97 kidney
1773 ebony	89 ebony, cardinal
128 kidney, ebony	6 kidney, ebony, cardinal
138 cardinal	8 wild type

(a) Determine the linkage relationships in the parents and F_1trihybrids.

(b) Estimate the map distances.

Solution

(a) The parents are homozygous lines:

$k\ e^+cd/k\ e^+cd$ × $ke^+cd^+/k^+\ ecd^+$

kidney, cardinal females ebony males

The F_1 is then trihybrid

$k\ e^+cd/k^+e\ cd^+$

wild type

The linkage relationships in the trihybrid F_1 can also be determined directly from the F_2. By far the most frequent F_2 phenotypes are kidney, cardinal (1761) and ebony (1773)] indicating that kidney and cardinal were on the chromosome in the F_1 and ebony on the other.

(b) Crossing over between the loci *k* and *e* produces the kidney, ebony (128) and cardinal (138) offspring. Double crossovers are the triple mutants (6) and wild type (8). Altogether there are 128 + 138 + 6 + 8 = 280 crossovers between *k* and *e*:

280/4000 = 0.07 or 7% crossing over = 7 map units

Crossovers between *a* and *cd* produced the single-crossover types kidney (97) and ebony, cardinal (89). Double crossovers again must be counted in this region.

97 + 89 + 6 + 8 = 200 crossovers between *a* and *cd*

200/4000 = 0.05 or 5% crossing = 5 map units

Note: Students are advised to solve the other unknown problems as stated above.

EXERCISE 4

STUDY OF HUMAN KARYOTYPES (NORMAL AND ABNORMAL)

Structure

4.1 INTRODUCTION

In this exercise, you will learn and identify the different groups of human chromosomes and prepare a karyotype of them from the figures provided. You will also learn about the abnormal chromosomal numbers that result in specific syndromes commonly occurring in the human males and females.

Individual chromosomes are most easily studied during metaphase. At that time, each chromosome clearly shows the two chromatids connected by a centromere. It is possible to stop the process of mitosis in metaphase by chemical means and to photograph the chromosomes. These photographs of metaphase smears are arranged in a prescribed manner to obtain the karyotype of that individual.

Objectives

After doing this laboratory exercise you should be able to:

- prepare karyotypes from the xerox photographs of chromosomes provided,

- ❖ identify karyotype of normal male and female, and
- ❖ diagnose the chromosomal disorders from the abnormal karyotypes provided.

4.2 MATERIAL REQUIRED

1. Human chromosome photographs
2. Human karyotype forms
3. Scissors
4. Pencil
5. Tape or Glue

4.3 KARYOTYPING

Biologists have developed a system for identifying each of the 46 chromosomes. The 22 pairs of autosomes are numbered from 1 to 22 according to their length. The sex chromosomes constitute the pair 23. It is very difficult to arrange chromosomes exactly according to number. However, the 23 pairs have been arranged into 7 groups according to the size and location of centromere. Table 4.1 gives this information. This table would be your key guide in the preparation and study of karyotypes in this exercise.

Table 4.1: The Seven Groups of Chromosomes.

Group	Chromosome	Characteristic
A	1, 2, 3	Very long; centromere in the centre of chromosome
B	4 and 5	long; centromere away from centre of chromosomes
C	6, 7, 8, 9, 10, 11, 12, X	Medium length; centromeres in the centre or slightly away from center of chromosomes
D	13, 14, 15	Medium length; centromeres at or very near to the end of chromosomes
E	16, 17, 18	Somewhat short; centromeres in the centre or away from the centre of chromosomes
F	19 and 20	Short; centromeres in the centre of chromosomes
G	21, 22, Y	Very short; centromeres at or very near to the end of chromosomes

The chromosomes of the first 22 pairs (autosomal chromosomes) are similar in all human karyotypes. But the chromosomes of the twenty-third pair, the sex chromosomes are dissimilar in male, i.e., there is a larger X chromosome and a smaller Y chromosome. Females have two X chromosomes in their karyotype.

In this investigation you would study three abnormal karyotypes.

i) Down's syndrome – an extra chromosome 21 is associated with this disorder.

ii) A missing X chromosome in females causes Turner's syndrome; and

iii) The occurrence of an extra X chromosome (XXY) in males leads to Klinefelter's syndrome.

4.4 PROCEDURE

Step 1 : Cut each individual chromosome from sheet I and II. Be careful not to cut any part of the chromosomes. Count how many you have.

Note: The chromosome cut-outs are very light and are easily lost. Should a chromosome be lost from the sheet set, the set would become incomplete and the whole exercise will be useless. So be very careful.

Step 2 : Arrange the chromosomes in the karyotype forms **a** and **b** respectively according to the chromosomes characteristics provided in Table 4.1 placing the short arm of each chromosome towards the top.

Step 3 : Pair the chromosomes referring to their banding pattern (if provided) and other features mentioned above.

Step 4 : After the chromosomes are arranged in order, glue or tape them in place.

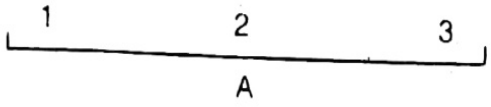
Step 5 : Answer the questions based on sheets I and II about the karyotypes you have prepared.

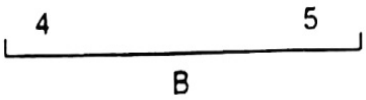
Step 6 : Study the karyotype sheets III, IV and V critically, and answer the questions based on them.

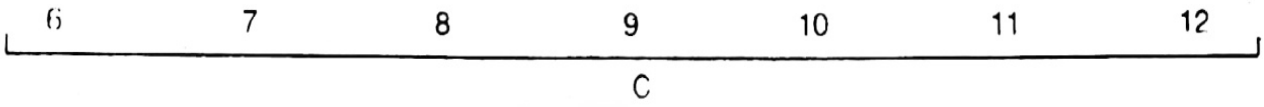
SHEET – I

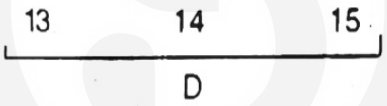
Hint: This is chromosome spread from a normal human.

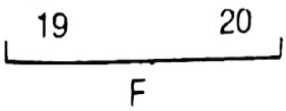
Karyotype Form a

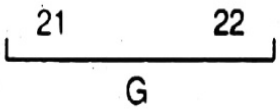










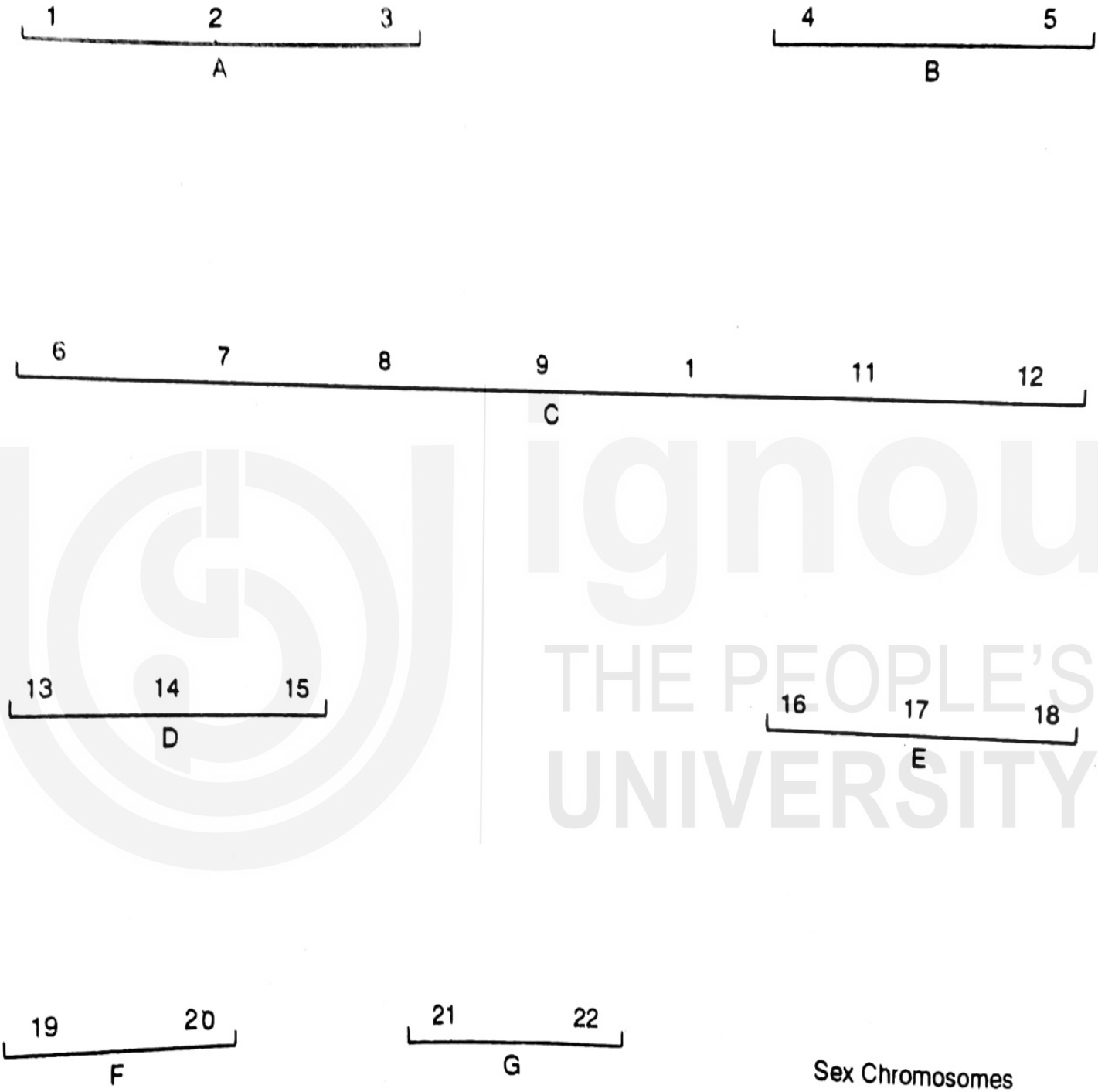


Sex Chromosomes

SHEET – II

Hint: This is a chromosome spread from a normal human.

Karyotype Form b



4.5 QUESTIONS BASED ON SHEETS I and II

1) Why must the photographs of individual chromosome be cut separately from sheet I and II, and pasted in order to karyotype it?

……
……
……

2) Why metaphase smears are considered the best for karyotype studies?

……
……
……

3) Identify the sex of the individuals whose chromosomes appear in Sheet I and Sheet II respectively?

……
……
……

4) Compare the two karyotypes you have made. What specific difference can you find?

……
……
……

5) How important is this difference? Explain.

……
……
……

6) For what kind of an individual would a genetic counsellor, most likely recommend karyotyping done? Why?

……
……
……

7) a) What kind of difficulties did you have during sorting out the chromosomes into pairs? B) Into groups? C) Would someone be preparing a karyotype in a genetics lab, have the same difficulties? Comment.

……
……
……

4.6 STUDY OF AN UNKNOWN KARYOTYPE FROM SHEET-III

This is a karyotype of an individual. Observe it and answer the questions given in Section 4.7.

SHEET – II

4.7 QUESTIONS BASED ON SHEET-III

1) Is this a karyotype of a normal individual or does it show any abnormality?

……

2) What is the feature that makes it different from the earlier two karyotypes?

……

3) Based on (2), name the genetic condition that such persons have?

……

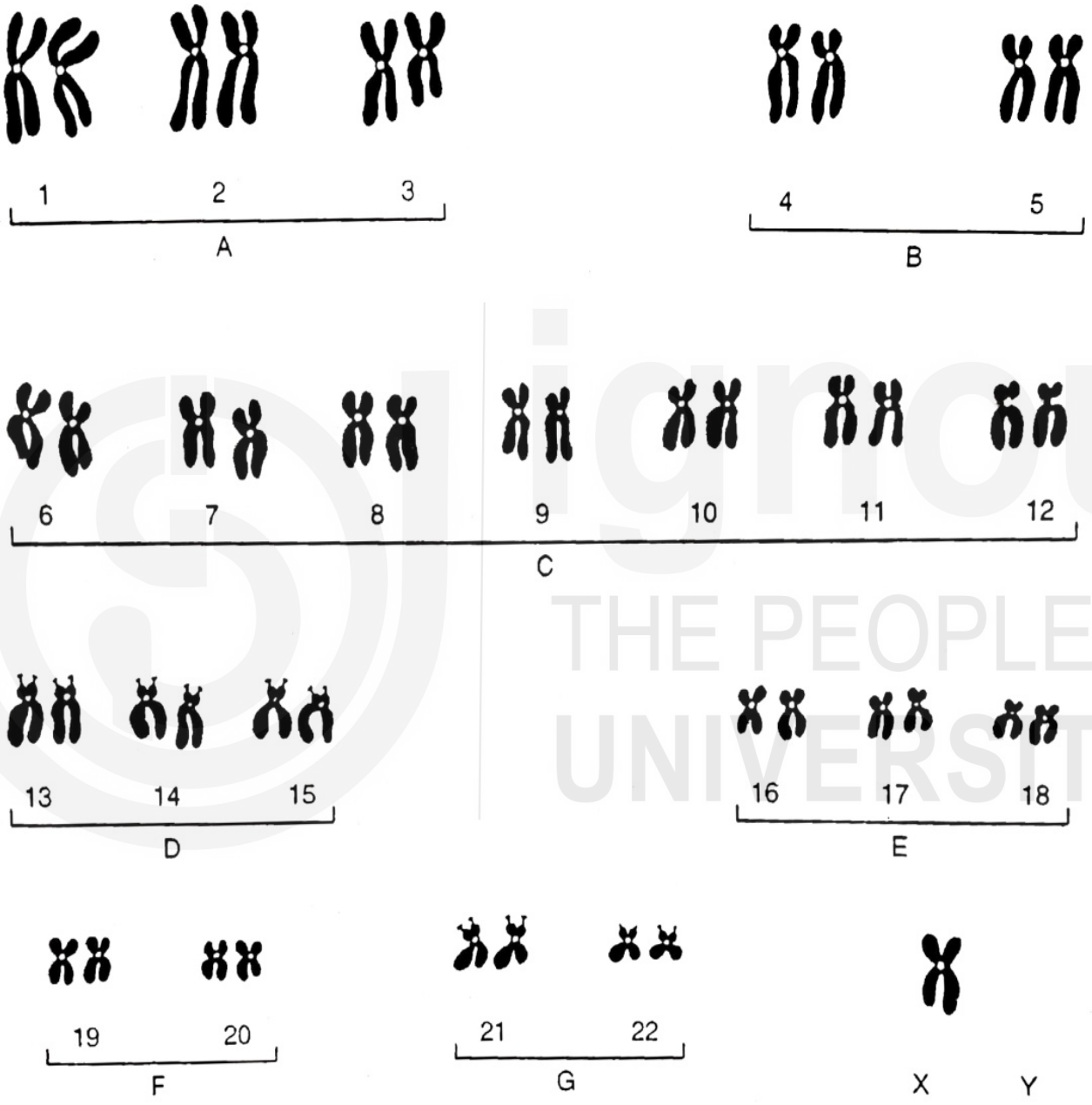
4) What are the prominent physical features of such individuals?

……

4.8 STUDY OF AN UNKNOWN KARYOTYPE FROM SHEET-IV

This is a karyotype of an individual. Observe it and answer the questions given in Section 4.9.

SHEET – IV



4.9 QUESTIONS BASED ON SHEET-IV

1) Does sheet IV show a normal karyotype or any abnormality?

……

2) If found abnormal, then what is the difference?

……

3) Such karyotype belongs to a person with which kind of genetic aberration?

……

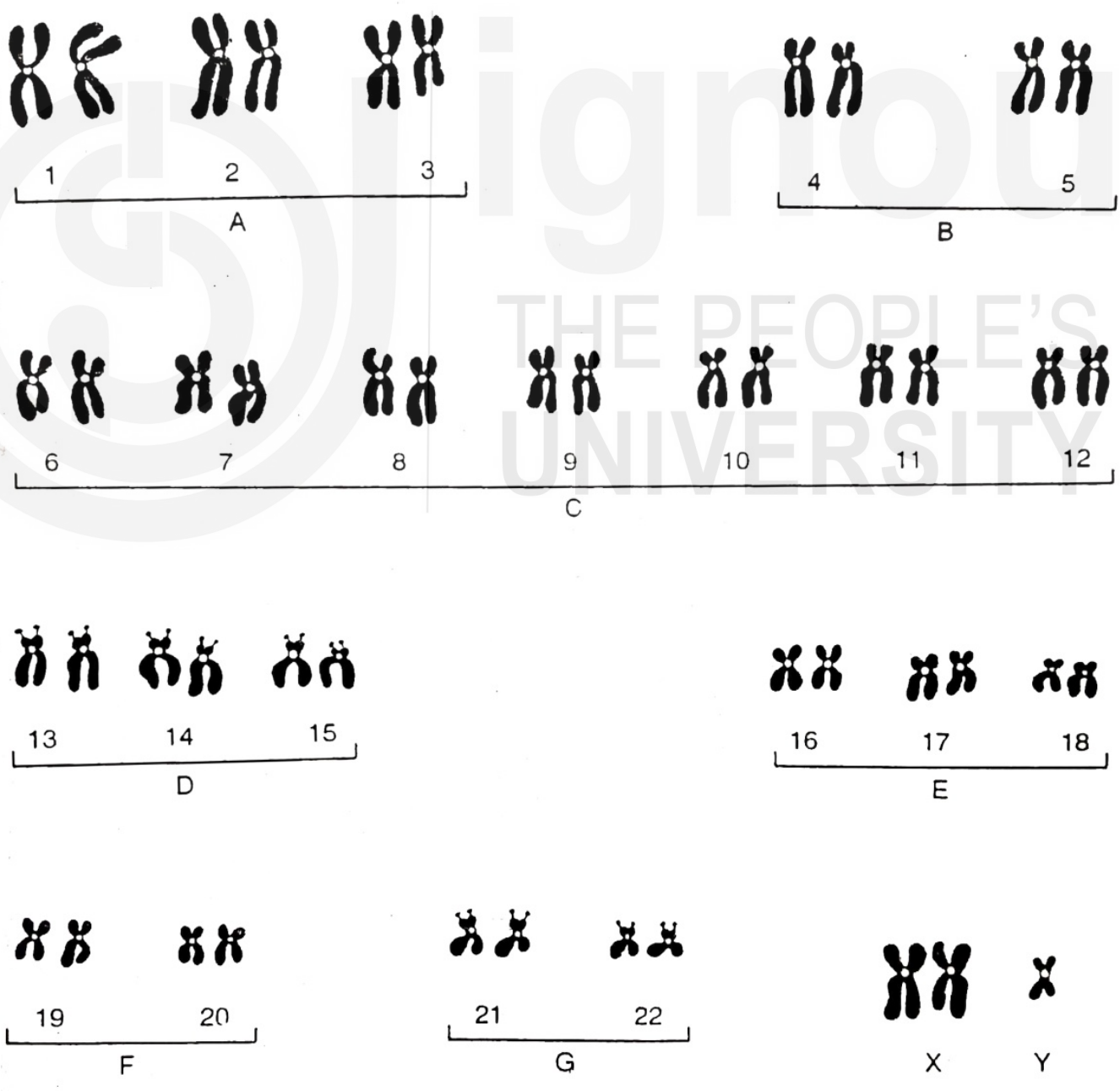
4) What are the prominent physical features exhibited by such persons?

……

4.10 STUDY OF AN UNKNOWN KARYOTYPE FROM SHEET-V

This is a karyotype of an individual. Observe it and answer the questions given in Section 4.11.

SHEET – V



4.11 QUESTIONS BASED ON SHEET-V

1) Does the sheet V show a normal karyotype or any abnormality?

……
……

2) If you have found it abnormal, then what is the difference?

……
……

3) This is a karyotype of an individual having which kind of genetic aberration?

……
……

4) What are the prominent physical features that help in identifying such individuals?

……
……

EXERCISE 5

STUDY OF PEDIGREE CHARTS

Structure

5.1 INTRODUCTION

In course on Genetics and Evolutionary Biology (BZYCT-137) you have studied certain traits of humans which follow the Mendelian concept of dominance-recessive relationship. You are aware that Mendel followed the inheritance of such traits in pea plant through successive generations by controlled crosses. Also, such crosses have led to the deduction of principles of segregation and independent assortment. These principles are applicable to all plants and animals including human beings. However, the type of crossing experiments done with plants and animals, cannot be performed on humans. Therefore, other methods are used to study the human inheritance patterns. One method that is extensively used by human geneticists and genetic counsellors is the construction and analysis of pedigree charts. The pedigree charts consist of a set of symbols which convey the details regarding the transmission of a trait over a number of successive generations. It is possible, after a careful study of pedigree chart to deduce whether a trait is dominant or recessive and whether it is an autosomal trait or a sex linked one. In this exercise we shall learn to draw pedigree charts and analyse them meaningfully. Make sure that you are familiar with the concepts of autosomal and sex chromosomal inheritance from your studies on Genetics and Evolutionary biology course.

Objectives

After performing this exercise you should be able to:

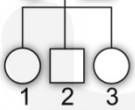
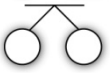
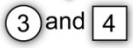
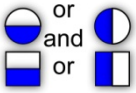
- explain various symbols that are commonly used in pedigree charts,

- ❖ draw a pedigree chart based on the data available or provided to you, and
- ❖ analyse and interpret pedigree chart and assign the trait to autosome or sex chromosome and say whether the concerned allele is dominant or recessive.

5.2 PROCEDURE FOR CONSTRUCTING PEDIGREE CHARTS

Before we begin with constructing or analysing the pedigree charts it is necessary to get yourself acquainted with different symbols that are commonly used in pedigree charts. Table 5.1 provides you the symbols used and their meanings. We shall begin with the construction of a simple pedigree chart and then start analysing the more complex ones.

Table 5.1: Symbols and Pedigree Charts.

1.		Normal female
2.		Normal male
3.		Indicates mating
4.		Indicates consanguinous mating or mating between close relatives.
5I.		Normal parents with 3 normal children, 2 daughters and one son.
II		Roman numerals denote generation numbers and arabic numerals, the order in which the children are born.
6.		Only a single parent is shown as the other parent is normal and is of no significance in pedigree analysis.
7.		Fraternal twins or twins arising from two different zygotes (dizygotic).
8.		Identical twins or twins arising from a single zygote (monozygotic).
9.		Sex unknown.
10.		Number of children for each sex.
11.		Shaded circle or square indicates affected daughter or son.
12.		An arrow below the affected individuals indicates that the analysis begins from that individual. The individual is an index case. The affected indivdual is thus a proband or propositus.
13.		Autosomal heterozygous recessive.
14.		Sex linked carrier individual.
15.		Deceased individual.
16.		Aborted foetus or a stillborn child

Humans fall into two categories depending on their ability to taste a chemical the phenylthiocarbamide (PTC). The tasters taste this chemical bitter and the non-tasters do not taste the chemical at all. The allele T determines the ability to taste PTC and people who are homozygous recessive to the allele (tt) are non-tasters. We have the following data on taste blindness from a family.

The female parent is a non-taster and the couple had five children, three daughters and two sons, of whom a son and a daughter are non-tasters. One of the taster daughters is married to a non-taster man. This couple had eight children, five sons and three daughters, of whom two sons and a daughter are non-tasters.

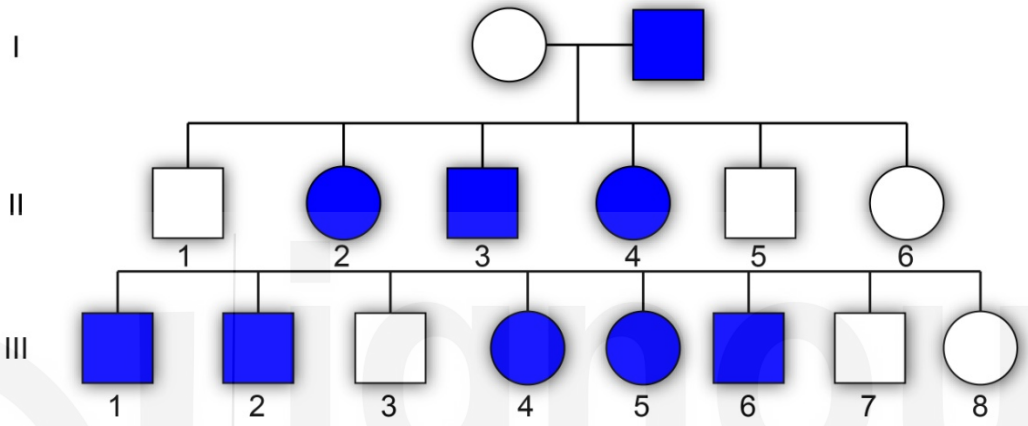


Fig. 5.1: A pedigree chart is constructed to show the inheritance pattern of non-taster allele in the family.

The pedigree (Fig. 5.1) shows that the female parent is a non-taster.Similarly II-1, II-5 and II-6 individuals are non-tasters; so also III-3, III-7 and III-8 individuals. It is possible to deduce from the pedigree that the trait is determined by an autosomal allele. The method of deducing is discussed below. Briefly it could be said here that if the allele were to be a sex linked trait, all the males of the second generation would have inherited it. You have already learnt that sex linked characters are generally transferred from mother to sons and from father to daughters, a phenomenon known as **criss-cross inheritance**. From the pedigree chart it is possible to deduce whether the trait is a dominant one or recessive. Assuming the trait is a dominant one and the father is homozygous for it (TT), then the mother will be homozygous recessive (tt). All the children of this couple, then would necessarily be taster and heterozygous (Tt). However, this is not the case here. Assuming the father is heterozygous dominant, then half of their children would be non-taster and the other half tasters. More or less this appears to be the case in the pedigree cited. The II-4 female, as taster is married to a non-taster. Assuming she is also heterozygous, dominant, then half of the children would be non-tasters and the other tasters. The data suggests that this is so in the III generation. So could the taster allele be dominant?

Assuming the taster to be a recessive one, and the female of generation is heterogyzous for non-taster trait, then again results similar to the one shown in the pedigree chart would be obtained. The pedigree chart is thus incomplete in the sense that it does not help us to decide whether the concerned allele is dominant or recessive. Essentially the chart has to be expanded by collecting

more data on the family. Let us say that we have additional data. The III-8 female, a non-taster is married to a taster male and has four children all of whom are tasters. Let us now redraw the pedigree chart with the additional information (Fig. 5.2).

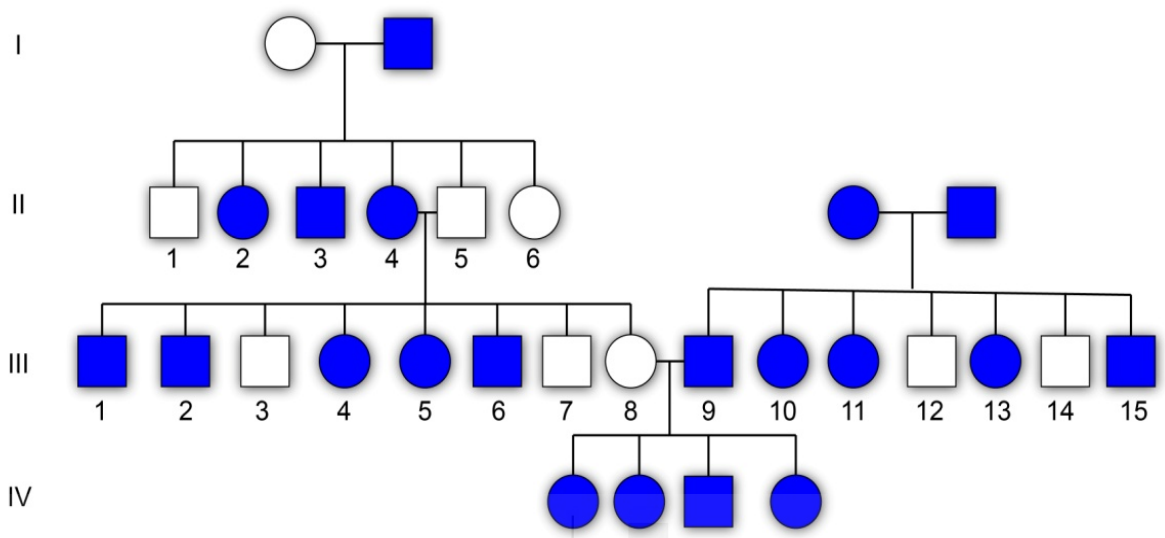


Fig. 5.2: The pedigree chart of III-8 female who is a non-taster is married to a taster male and has four children all of them are tasters.

Now, the III-8 and III-9 individuals are married and have four taster children. This part of pedigree chart helps us to determine beyond any doubt the dominance or recessiveness of the allele. The fact that all the four children born to III-8 and III-9 parents are tasters and that only one of the parents is a taster, clearly tells us that the father is homozygous dominant (TT) and the mother is recessive (tt) (Fig. 5.3).

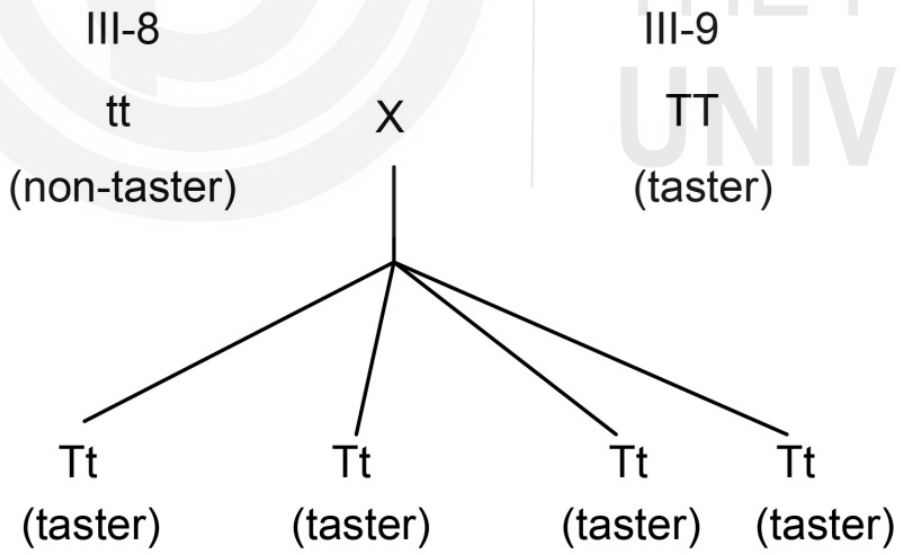


Fig. 5.3: Father homozygous dominant (TT) is married to mother who is recessive (tt).

Therefore, the allele in question is a dominant allele. Essentially PTC taster trait is an autosomal dominant trait.

Certain general patterns can be followed to identify whether a trait is autosomal or sex chromosomal and whether it is dominant or a recessive one. Let us categorise these patterns.

5.3 AUTOSOMAL DOMINANT TRAITS

a) Autosomal dominant traits make their appearance invariably in all generations. In other words, they do not skip generations.

b) An individual carrying the gene for the affected trait (heterozygous) married to a normal individual generally produces normal offspring to affected ones in the ratio of 1:1.

c) There is no discrimination between sexes and the trait is distributed equally in both the sexes.

The pedigree in Fig. 5.4 shows the general pattern of inheritance of an autosomal dominant trait. Study the pedigree carefully and make sure that the pattern of autosomal dominant inheritance conforms with the statements we have made above.

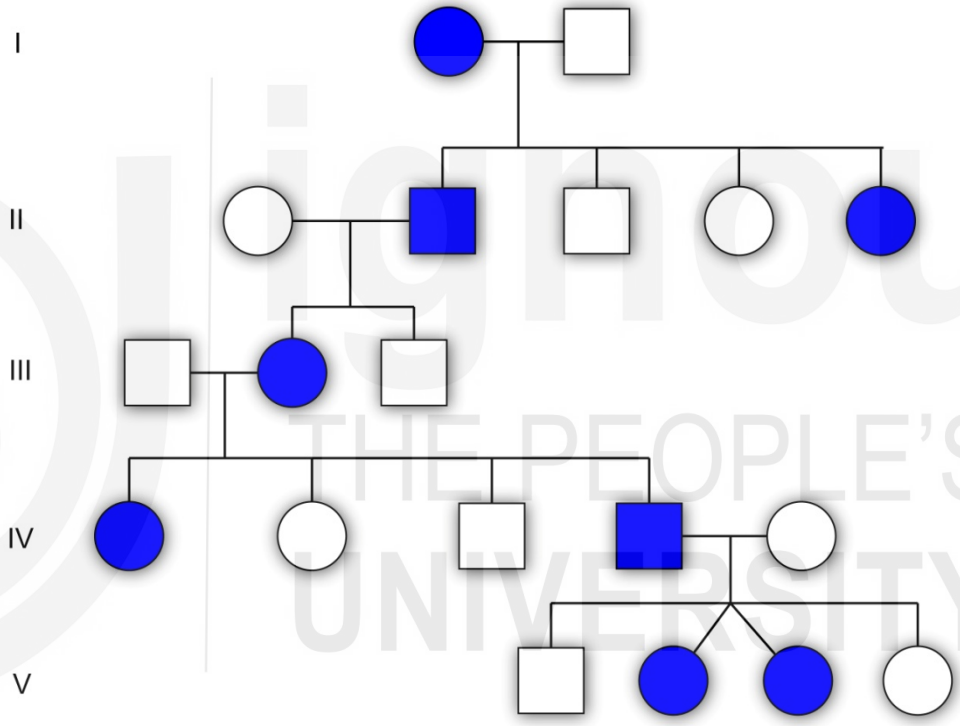


Fig. 5.4: General pattern of inheritance.

Brachydactyly, Huntington's disease, ability to taste phenylthiocarbamide and polydactyly are some of the autosomal dominant traits in humans.

5.4 AUTOSOMAL RECESSIVE TRAITS

a) Unlike the autosomal dominant traits, the autosomal recessive ones do not make their appearance in every generation. In other words, they may skip certain generations and appear only in certain others.

b) Like autosomal dominants, the distribution is equal between the two sexes.

c) They are more commonly found among children born to consanguineous married couples, that are couples who were first cousins before marriage or otherwise closely related.

d) If both the parents are affected, all the children born to them will also be affected.

e) The parents of the affected children may be normal.

f) The affected child born to normal parents essentially suggests that the parents are heterozygous and are carriers of the allele for the trait.

g) If both the parents are heterozygous, then the chances are that approximately 50% of the children born to them would inherit the recessive trait.

The pedigree given in Figure 5.5 is an example of the inheritance pattern of albinism in humans. Albinism is an autosomal recessive trait and refers to a condition in which a person can not synthesise the pigment melanin. Study the pedigree carefully and verify that the inheritance of albinism follows the pattern that is characteristic of an autosomal recessive trait.

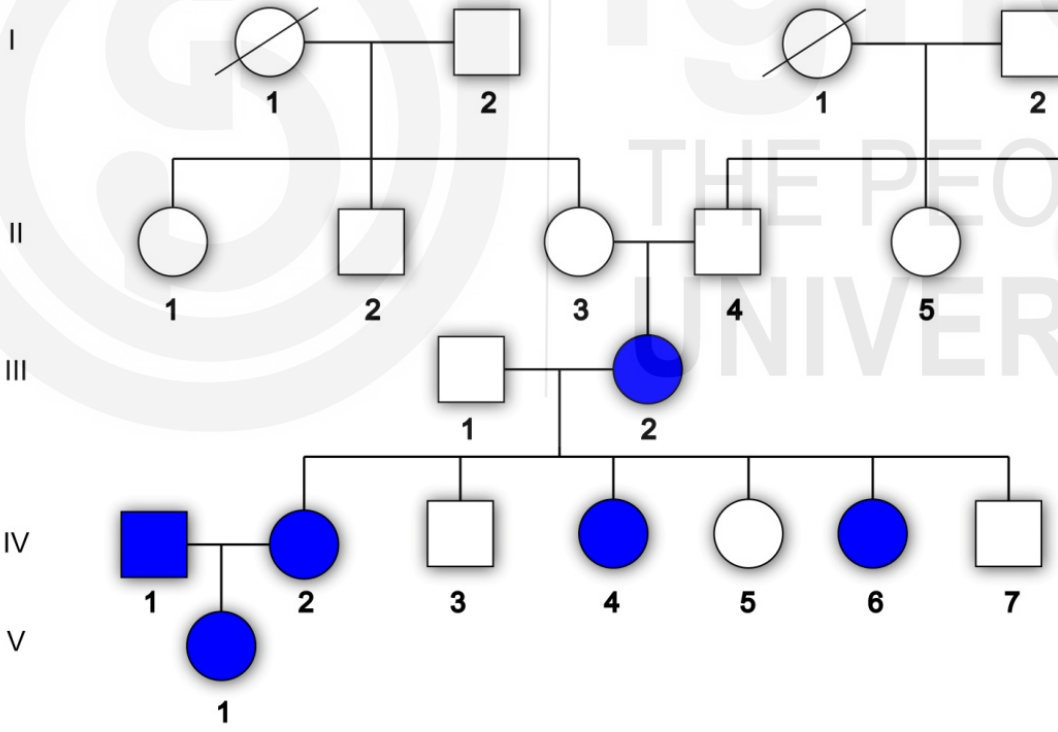
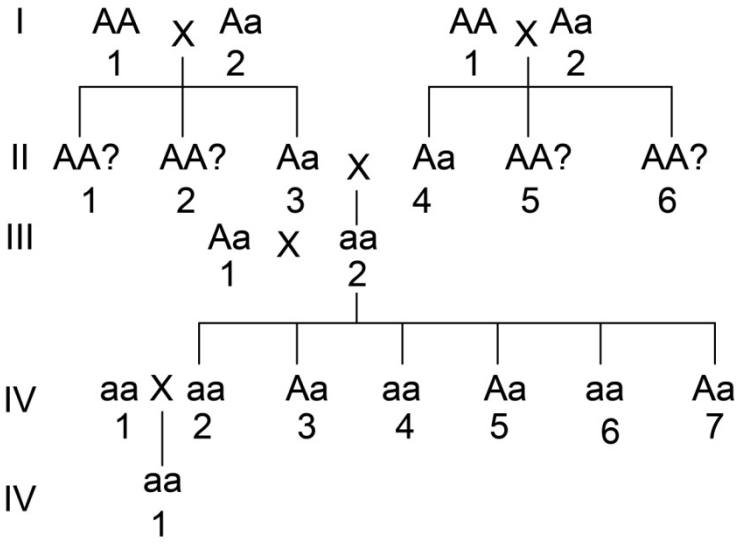


Fig. 5.5: Inheritance pattern of albinism in humans.

An analysis of the pedigree in Figure 5.5 shows that in individual there is an inheritance pattern of albinism in human albino, although both her parents are normal. Essentially they should be heterozygous and the daughter receives the two recessive alleles. Also in generation IV individuals 2, 4 and 6 are also albinos, suggesting that their father is also heterozygous. Since both 1 and 2 are generation IV albinos their daughter is also an albino. The possible genotypes of the above pedigree can be written as follows (Fig. 5.6).



The question mark suggests that the genotypes of individuals could be either AA or Aa.

Fig. 5.6: Genotype of the pedigree of albino.

5.5 SEX CHROMOSOMAL DOMINANT TRAITS

Sex linked dominant traits are very rare. A sex linked dominant trait is oral-facial-digital syndrome which results in cleft tongue, absence of teeth and mental retardation. Other sex linked dominant alleles responsible for certain diseases include the allele for Albright's hereditary osteodystrophy causing seizures, stunted growth and mental retardation, Goltz's syndrome whose symptoms are mental retardation, small eyes and flexed digits and incontinentia pigmenti which causes the non-retention of melanin in melanoblasts. Figure 5.7 shows a pedigree from one such sex-linked dominant trait.

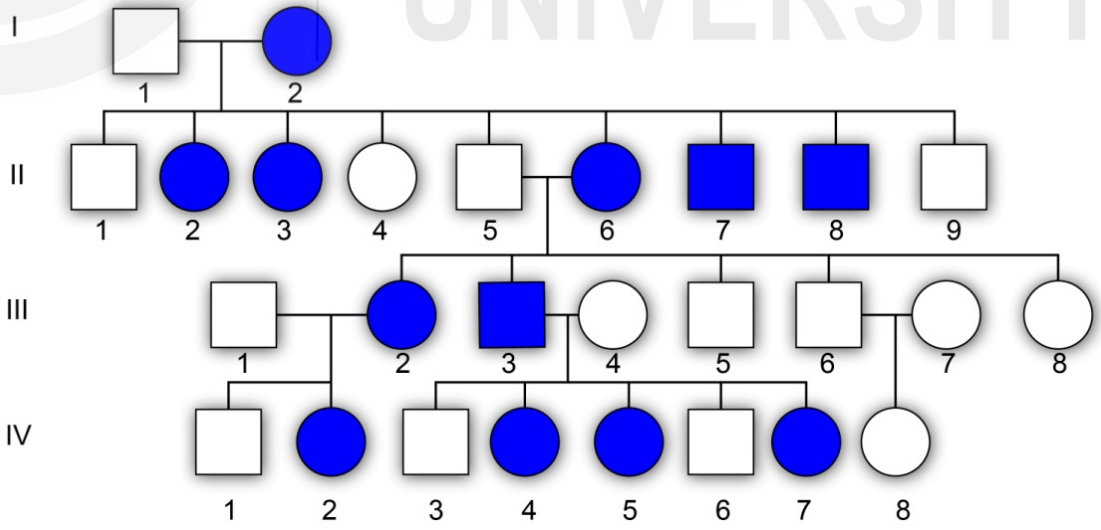


Fig. 5.7: Pedigree of sex linked dominant trait.

You may observe in the Figure 5.7 that the trait appears in all generations; and the trait always passes from the father to the daughters and not to the sons (Note that III-3 male has passed the trait to all his daughters but the two sons are normal).

5.6 SEX-LINKED RECESSIVE TRAITS

a) Since males have only one X-chromosome and receive the same from their mother, they are the ones most affected by sex linked recessive alleles.

b) Generally, the females are carriers of the recessive allele and thus heterozygous in genotype.

c) The females are known to be heterozygous because of affected brothers, fathers or maternal uncles.

d) Males receive the X-linked recessive trait from their mother.

e) Females express the trait only when they have a homozygous recessive genotype and the alleles are received from carrier mothers and affected fathers.

f) All sons of an affected female are affected and 50% of the daughters are carriers.

g) Nearly 50% of the sons of the carrier female would be affected.

Several sex linked recessive traits are known in humans and haemophilia is the most famous case of sex linked recessive inheritance. Colour blindness, the inability to distinguish red and green colours is another one. Many deficiencies relating to enzymes such as G-6-PD deficiency (glucose 6 phosphate dehydrogenase deficiency) are also sex linked. The pattern of inheritance of a linked recessive allele is shown in the Figure 5.8.

The GEPD provides instructions for making an enzyme called Glucose to phosphate dehydrogenase. This enzyme, is involved in the normal processing of carbohydrates. This enzyme helps protect red blood cells from damage and premature destruction.

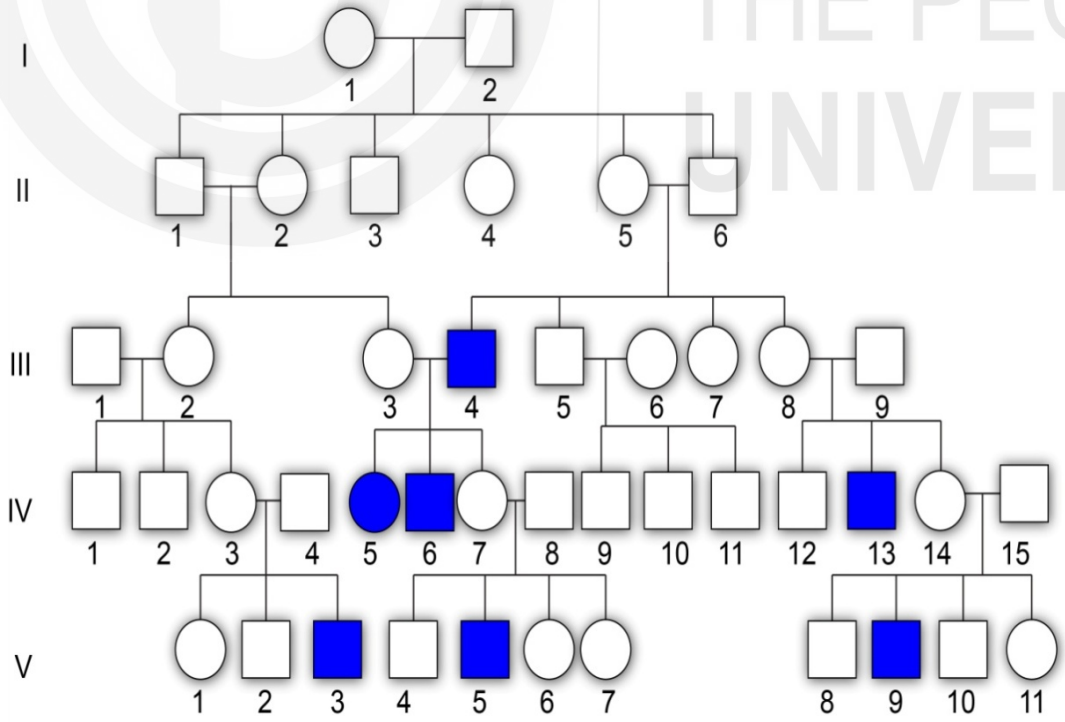


Fig. 5.8: The pattern of inheritance of a sex-linked recessive allele.

In the case of sex linked recessive inheritance, the number of males affected are always more than females. This is essentially due to the fact that the males receive only one X chromosome and that is received from the mother.

You can observe from Figure 5.9 that III-3, a normal female married to III-4, an affected male gives birth to 3 children, 2 daughters and one son. Since the son is affected, it is obvious that the mother is the carrier of the recessive allele and has passed the trait to him.

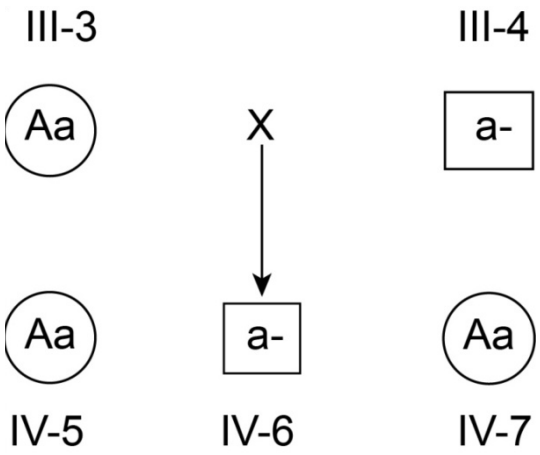


Fig. 5.9: III-3, a normal female married to III-4, an affected male gives birth to 3 children, 2 daughters and one son; Since the son is affected the mother is the carrier of recessive allele.

Similarly, III-8, IV-7 and IV-14 are heterozygous mothers who have passed the trait to their son.

We have thus been discussing the analysis of pedigree charts and assigning the traits to autosomes or sex chromosome and to identify them as dominant or recessive ones. You may now work on the following problems to test your understanding of pedigree charts.

5.7 TERMINAL QUESTIONS

1. Analyse the following pedigree and answer the questions as directed below: (Fig. 5.10).

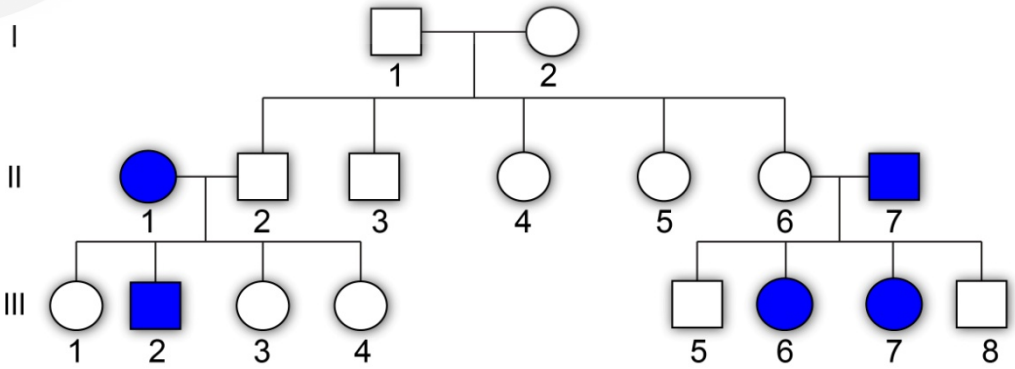


Fig. 5.10

a) Is the trait an autosomal or a sex chromosomal one?

b) Is the gene that causes the trait a dominant or a recessive one?

c) What could be the genotypes of I-1, I-2, II-1, II-6, II-7, III-5 and III-7 individuals, assuming the dominant allele is S and recessive allele is s.

2. Below is given the pedigree of a family (Fig. 5.11), certain individuals of which are affected by an inherited metabolic disorder alkaptonuria. The disease is caused by a defect in the metabolism of the amino acid phenylalanine, Answer the questions given below after a careful analysis of the pedigree.

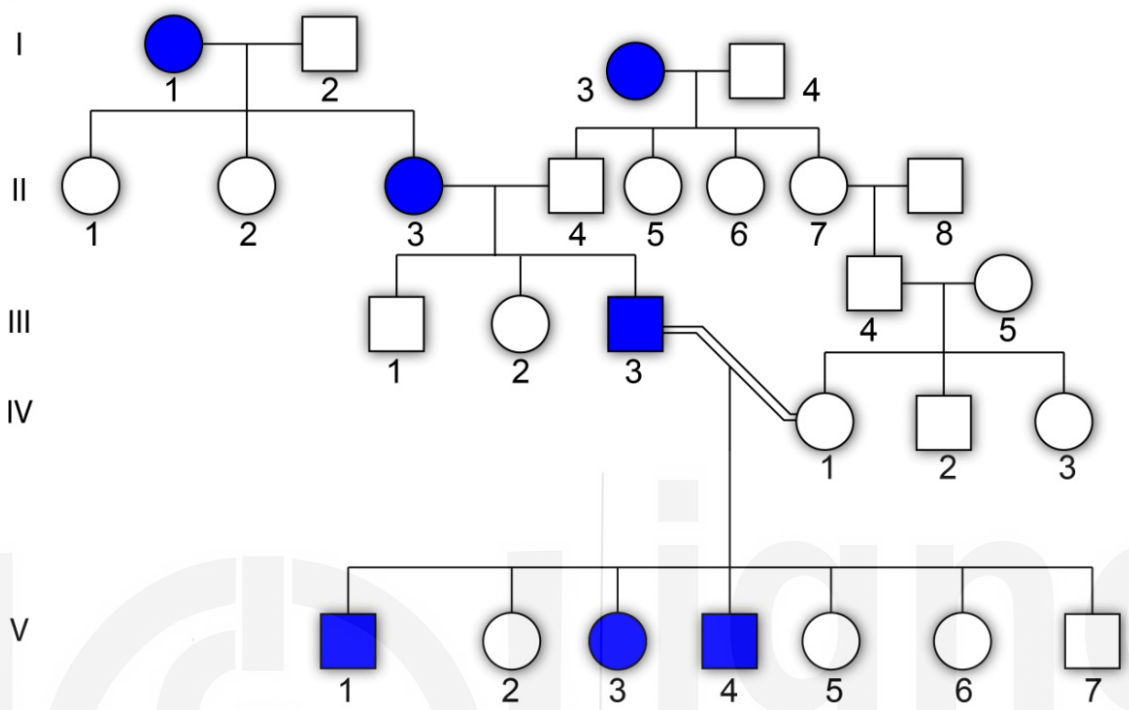


Fig. 5.11: Pedigree of family.

i) Does the above pedigree suggest an autosomal or sex chromosomal inheritance?

ii) Does the inheritance pattern suggest the involvement of a dominant allele or recessive allele?

iii) How would you explain the marriage between III-3 and a IV-1 individuals?

iv) Assuming the dominant and recessive alleles are designated as A and a what are the genotypes of I-2, II-3, II-4, III-3 and IV?

EXERCISE 6

APPLICATION OF PROBABILITY TO PROBLEMS IN GENETICS

Structure

6.1 INTRODUCTION

One of the effective tools for a geneticist and a genetic counsellor to assess the possible occurrence of a trait in a family is the application of probability theory. A probability is the ratio of the number of times a particular event occurs to the number of trials which the event could have happened. Assuming a man and his wife seek advice of a genetic counsellor on a genetic problem, the counsellor analyses the pedigree of the couple's family, establishes the genotypes of the couple and then makes calculations relating to the probability of the appearance of the trait in question in the children to be born to them in future. In this lab exercise, you will learn some of the basic rules of probability theory. You should try to apply them for problems in genetics.

Objectives

After the end of this lab exercise, you should be able to:

- describe the basic principles of probability;
- make use of the principles of probability to solve genetic problems;
- apply the formula for the binomial expansion to determine the probability of any combination of events;
- comprehend the Pascal's triangle to determine the coefficient of binominal expression that tells how many ways a particular combination may be obtained.

6.2 THE BASIC PRINCIPLES OF PROBABILITY THEORY

The probability theory, it can be said, helps you to make a proper and meaningful guess of the occurrence of an event. The probability *P* of the occurrence of an event is the number of favourable cases (a) divided by the total number of possible cases *n*.

$$P = \frac{a}{n}$$

In determining the probability of an event, one way is to observe a large number of cases and record the number of times an event occurs or does not occur. But this is an empirical method. A better way of determining the probability is the one that is generally used by the geneticists for making predictions on the occurrence of an event. The probability obtained by this method is *a priori* probability. You should look into the following example that explains the calculation of probability before we take up specific examples from genetics.

Take a die (plural = dice) with six faces numbered 1 to 6. When the die is rolled, the probability *p* of any one face showing up is $1/6$.

$$P = \frac{a}{n} = \frac{1}{6} = 0.167$$

The probability of picking up the nine of spades from a deck of 52 cards is

$$P = \frac{1}{52} = 0.0192$$

And the probability or drawing any one club from a deck of cards is

$$P = \frac{13}{52} = \frac{1}{4} = 0.25$$

Now let us cite one or two examples from genetics. The probability of an offspring to be of recessive genotype when a monohybrid is self-fertilised is

$$P = \frac{1}{4} = 0.25$$

and the probability of an offspring to be of dominant phenotype for both traits on self-fertilisation of a dihybrid is

$$P = \frac{9}{16} = 0.5625$$

When we say that the probability of the occurrence of an event is *P*, the combined probability of occurrence of all other events is $Q = (1 - P)$. Thus when the *P* of occurrence of dominant phenotypes $= 9/16$, the combined probability of occurrence of other phenotypes $= Q = 1 - 9/16 = 7/16$ and $P + Q = 9/16 + 7/16 = 1$. $P + Q$ is always equal to 1. In fact all probabilities must lie between 0 and 1. A probability of 1 means that the event is certain to occur; a probability of zero indicates that the event cannot occur.

Now, let us look into situations where we consider the occurrence of two events. By two events, we mean the occurrence of either one of the events or both the events simultaneously. Essentially in such cases we will be combining the probability of occurrence of the two events. There are three rules under which the combining of the probabilities can be done.

6.2.1 Addition Rule

When the occurrence of one event precludes the possibility of the occurrence of another event, the two events are said to be **mutually exclusive.** Essentially it means that when one event occurs, the other does not. And the probability of occurrence of one of several mutually exclusive events is the sum of the probabilities of individual events. For instance, when a die is thrown what is the probability that is shows either a two or a three?

P of getting a two $= 1/6 = 0.167$

P of getting a three $= 1/6 = 0.167$

Therefore, the two events are mutually exclusive. Theprobability of getting either a two or a three $= 1/6 + 1/6 = 2/6 = 1/3 = 033.$ Since the probability of occurrence of mutually exclusive events is summed up, the rule is called **addition rule**.

6.2.2 Product Rule

Product rule is used when the occurrence of one event is not dependent on the occurrence of another event; in other words we are dealing with independent events. For instance, when two dice are thrown simultaneously the probability of getting a two and a three in that order are

P of getting a two $= 1/6 = 0.167$

P of getting a three $= 1/6 = 0.167$

Probability of getting a two and a three $= 1/6 \times 1/6 = 1/36 = 0.028.$ Since the probability of occurrence of independent events is the product of their separate probabilities, this rule is called the **product rule**.

Look into this example,

What is the probability of getting a heat and a rail, when two coins are tossed simultaneously? This procedure as we are going to demonstrate to you, requires the use of both addition and product rules. For each coin the probability of getting a head H or tail T is

$P(H) = 1/2 = 0.5$

$P(T) = 1/2 = 0.5.$

When the coins are tossed one at a time, there are two ways of getting a head or a tail.

First head and then tail (HT)

First tail and then head (TH)

The results of each of the two tosses in a sequence are independent events.

The probability of getting HT and $\mathrm{TH} = 1/2 \times 1/2 = 1/4 = 0.25$

At the same time, the two sequences are mutually exclusive. The probability of getting either of two sequences of a set of mutually exclusive events is

$$\frac{1}{4} + \frac{1}{4} = \frac{1}{2} = 0.5$$

Thus when events are unordered, the probability can be obtained by combining addition and product rules.

6.2.3 The Binomial Theorem

The probability of unordered events can be determined by using binomial theorem. This theorem defines the probability of the occurrence of some arrangement of two mutually exclusive trials where the final order is not specified. According to this theorem the frequencies or the probabilities of the occurrence of various combinations correspond to the terms of the binomial expansion. The first three binomial expressions are as follows.

$$(a+b)^2 = a^2 + 2ab + b^2$$

$$(a+b)^3 = a^3 + 3a^2b + 3ab^2 + b^3$$

$$(a+b)^4 = a^4 + 4a^3b = 6a^2b^2 + 4ab^3 + b^4$$

A simple formula based on binomial theorem would help you to calculate the probability by a short-cut method. According to this formula:

$$P = \frac{n!}{s!t!} \times p\,q$$

Where *n* is the total number of events, *p* is the probability of the occurrence of an event (X), *q* is the probability of the occurrence of an alternate event (Y), *s* is the number of times the event X will occur and *t* is the number of times the event Y will occur out of n number of trials. Here, $s + t = n$ and $p + q = 1$.

Let us look into the previous example. when

$$n = 2$$

Probability of getting a head $= p = \frac{1}{2}$

Probability of getting a tail $= q = \frac{1}{2}$

and $s = t = 1$.

Substituting the above data in the formula,

$P =$ the probability of getting a head and a tail when two coins are tossed

$$\text{Simultaneously} = \frac{2}{1!1!}\left(\frac{1}{2}\right)^1\left(\frac{1}{2}\right)^1$$

$$= \frac{2\times1}{1\times1}\ \frac{1}{2}\ \frac{1}{2} = \frac{1}{2}$$

$$= 0.5\,.$$

Let us now look into more specific example from genetics. What is the probability that a family with five children will have 3 boys and 2 girl?

$$\text{The probability of a child to be a boy} = p = \frac{1}{2}$$

$$\text{The probability of a child to be a girl} = q = \frac{1}{2}$$

Applying the formula

$$P = \frac{5!}{3!2!}\left(\frac{1}{2}\right)^3\left(\frac{1}{2}\right)^2$$

$$= \frac{5\times4\times3\times2\times1}{3\times2\times1\times2\times1}\times\frac{1}{2}\times\frac{1}{2}\times\frac{1}{2}\times\frac{1}{2}\times\frac{1}{2} = 0.312$$

Assuming the parents want the 5 children to be born in a specific order say 2 boys, 1 girl 1 boy and a girl. Essentially this mean that you have to apply the product rule; in which case the probability would be

$$\frac{1}{2}\times\frac{1}{2}\times\frac{1}{2}\times\frac{1}{2}\times\frac{1}{2} = \frac{1}{32} = 0.312$$

Thus you can see from the two answers, that when an order is specified, the probability is 10 times less than when no order is specified. In other words, while there is only one way of getting 2 boys, 1 girl 1 boy and 1 girl, there are 10 different ways of getting 3 boys and 2 girls.

1	2	3	4	5	6	7	8	9	10
B	B	B	B	B	G	G	G	G	G
B	B	B	G	G	G	B	B	B	G
B	G	G	G	B	B	G	B	B	B
G	B	G	B	G	B	B	G	B	B
G	G	B	B	B	G	B	B	G	B

[B = boy; G = Girl]

Assuming a couple is heterogygous (Aa) for albinism, what is the probability that 4 children out of 6 born to them are normal?

Let A = allele for normal skin

a = allele for albinism

$Aa \times Aa$

AA Aa Aa aa

normal albino

Since the ratio of normal to albino is 3 : 1, the probability of a normal son being born is $\frac{3}{4}$ and an albino is $\frac{1}{4}$.

The probability of 4 children being normal is

$$\frac{3}{4} \times \frac{3}{4} \times \frac{3}{4} \times \frac{3}{4} = \frac{81}{256} = 0.316$$

What is the probability of 4 children being normal and 2 children albinos?

In this case,

$$P = \frac{6!}{4!\,2!} \cdot \left(\frac{3}{4}\right)^4 \cdot \left(\frac{1}{4}\right)^2$$

$$= \frac{6 \times 5 \times 4 \times 3 \times 2 \times 1}{4 \times 3 \times 2 \times 1 \times 2 \times 1} \times \frac{3}{4} \times \frac{3}{4} \times \frac{3}{4} \times \frac{3}{4} \times \frac{1}{4} \times \frac{1}{4}$$

$$= \frac{1215}{4096} = 0.297$$

Assuming you specify the order in which the normal and albino children are born say the first 3 children are normal, 1 albino, 1 normal and 1 albino, then the probability would be

$$\frac{3}{4} \times \frac{3}{4} \times \frac{3}{4} \times \frac{1}{4} \times \frac{3}{4} \times \frac{1}{4} = \frac{81}{4096} = 0.0198$$

You can again observe that that once the order is specified, the probability assumes a lower value. In other words, if no order is specified, the probability is fifteen times larger than when it is specified.

6.3 PASCAL'S TRIANGLE

We earlier said that in the formula $P = (n!/s!t!)\,pq$, $(p+q) = 1$ and $(s+t) = n$. This formula essentially separates the binomial equation $(p+q)^n = 1$ and gives the probability of one of the terms. The binomial expansion of $(p+q)^n$ contains $(n+1)$ terms. A device called Pascal's triangle is useful to get the coefficient of the terms. The coefficients tell you the number of ways by which one can obtain a particular combination of events. The following is the Pascal triangle upto $n = 7$.

1.	n = 0	$(p+q)^0$	1
2.	n = 1	$(p+q)^1$	1 1
3.	n = 2	$(p+q)^2$	1 2 1
4.	n = 3	$(p+q)^3$	1 3 3 1
5.	n = 4	$(p+q)^4$	1 4 6 4 1
6.	n = 5	$(p+q)^5$	1 5 10 10 5 1
7.	n = 6	$(p+q)^6$	1 6 15 20 15 6 1
8.	n = 7	$(p+q)^7$	1 7 21 35 35 21 7 1

The above triangle is built out of the coefficient of binomial expansion. You may observe that any given row beings with 1, then proceed by the addition of two adjacent coefficients from the preceeding row and finally ends in 1. For example, take the 7th row, $(p+q)^6$.

$$(p+q)^6 = (1)p^6 + (1+5)p^5q + (5+10)p^4q^2 + (10+10)p^3q^3 + (10+5)p^2q^4 + (5+1)pq^5 + (1)q^6$$

Let us get back to our earlier problems. What is the probability of 4 normal and 2 albino children born to a couple heterozygous for the albino allele?

$$n = 6; s = 4, t = 2, p = \frac{3}{4} \text{ and } q = \frac{1}{4}$$

The problem is completely described by the expression of $(p+q)$. In the equation $(p+q)^6$ (remember $n = 6$), the term that is of interest to us is $15p^4q^2$, since $s = 4$ and $t = 2$.

$$15p^2q^2 = 15\ (3/4)^4\ (1/4)^2$$

$$= 15 \times \frac{3}{4} \times \frac{3}{4} \times \frac{3}{4} \times \frac{3}{4} \times \frac{1}{4} \times \frac{1}{4}$$

$$= \frac{1215}{4096} = 0.297$$

This is the same answer that we obtained using the formula. But when the order of the binomial increases, its expansion to get all the terms becomes difficult. In such cases we can readily use the formula.

6.4 MULTINOMIAL EXPRESSION

The formula has one more advantage. It could be expanded to include more than two events. The multinomial expansion $(p+q+r...)^n$ can be represented as the general formula for calculating the probability.

$$p = \frac{n!}{s!\,t!\,u!} \times p^s q^t r^u \ldots$$

Where $p + q + r \ldots = 1$ and $s + t + u \ldots = n$.

Assuming a couple were told by the genetic counsellor that each of them carry an allele for albino trait. The couple wants to have six children. What is the probability that of the six children, two will be normal daughters, two normal sons, one albino son and one albino daughter?

In this problem, you first apply the product rule to know the probability for each item and then apply the formula to get the probability for the entire event.

For example,

Probability of getting a normal son $p = (3/4)(1/2) = 3/8$

Probability of getting a normal daughter $q = (3/4)(1/2) = 3/8$

Probability of getting a albino son $r = (1/4)(1/2) = 1/8$

Probability of getting a albino daughter $k = (1/4)(1/2) = 1/8$

Probability of getting 2 normal sons (s), 2 normal daughters (t), 1 albino son (u) and 1 albino daughter (v) =

$$P = \frac{n!}{s!\,t!\,u!\,v!} . p^s q^t r^u k^v$$

$$P = \frac{6!}{2!\,2!\,1!\,1!} . (3/8)^2 . (3/8)^2 . (1/8)^1 . (1/8)^1$$

$$= \frac{6 \times 5 \times 4 \times 3 \times 2 \times 1}{2 \times 1 \times 2 \times 1 \times 1 \times 1} . 3/8 \times 3/8 \times 3/8 \times 3/8 \times 1/8 \times 1/8$$

$$= \frac{14580}{262144} = 0.0556$$

6.5 TERMINAL QUESTIONS

1. Assuming a sex ratio of 1:1, what is probability that a family of 4 children will consist of

   i) 3 daughters and 1 sons

   ii) All daughters

   iii) Alternating sexes

   iv) All sons

   v) Atleast two daughters

2. In lab exercise 5, we discussed about phenylthiocarbamide tasters. PTC tasting, as you are aware is dominant to non-tasting. A taster man

whose mother is a non-taster marries a taster woman who in a previous marriage had a non-taster daughter. What would be the probability of the couple having

a) Their first child a taster?

b) Their first child a non-taster boy?

c) 7 children with 1 taster and 3 non-tasters?

d) 5 children, of whom 2 taster boys 1 taster girl 1 non-taster boy and 1 non-taster girl in that order.

3. Two parents have genotype Mm and suffer from migraine headache. What is the probability that

a) Their first child will be a girl with migraine and their second a boy without the disorder.

b) 4 children are born to them with 3 children born without migraine and one child with it.

EXERCISE 7

STUDY OF FOSSIL EVIDENCES

Structure

7.1 INTRODUCTION

In the previous exercise you have learnt the construction and analysis of pedigree charts which consist of a set of symbols conveying the details with regard to transmission of trait over a number of successive generations. In the present exercise we will study the fossil evidences provided to us in the form of plaster cast models or pictures to buttress the theory of evolution put forth by Charles Robert Darwin (1809-1882). Our interest in the geological record is owing to the preservation in the earth's crust of living organisms from ages past in the form of fossils. Rock formation has been going on ever since the birth of the planet earth and fossilization has been taking place since the advent of life. In the course of such processes, mud, sand or stones are transported to the floor of depressions, lakes or oceans and accumulate there. Sometimes various organisms also die and get buried at the bottom of such water bodies. Material so deposited later becomes compressed and solidified, forming layers or strata of the rocks entrapping the remains of life in the form of fossils. Thus geological records are written in the rocks in the language of fossils.

Examination of the nature and distribution of stratified rocks enable the construction of geological history of the area from which the rocks were obtained. By analyzing the rocks it is also possible to discover environmental condition under which they were formed and also estimate their age. In this exercise we will examine some of fossil evidences available in your institution in the form of plaster cast model or pictures. Before performing this exercise it is desirable for you to go through the unit on fossil evidence in the course on Genetics and Evolutionary Biology (BZYCT-137).

Objectives

After performing this exercise you should be able to:

- relate the presence of fossils on the earth's crust to the occurrence of organic evolution,
- piece together the major events which took place during the course of evolution of animals, and
- link the geographical distribution of various species of contemporary plants and animals to the course of evolution.

7.2 MATERIAL REQUIRED

Plaster cost models or pictures of the following fossils:

1. Trilobite
2. *Ichthyosaurus*
3. *Dimetrodon*
4. *Brontosaurus*
5. *Diplodocus*
6. *Tyrannosaurus*
7. *Stegosaurus*
8. *Rhamphorhynchus*
9. *Pteranodon*
10. *Archaeopteryx*

7.3 EXAMINATION OF FOSSILS EVIDENCE (AVAILABLE IN THE FORM OF PLASTER CAST MODEL OR PICTURE)

The most convincing and direct evidence for evolution comes from the study of fossils. Fossils are the record of organisms of the past, preserved by burial in rocky layers. A fossil may be the product of preservation of an entire organism or of a part. The organism itself may be dissolved away, leading a **natural mould** or the mould may be filled with deposited material, forming a **natural cast.** Sometimes a fossil may be a mere animal **foot print** or **imprint** of a leaf on the rock. Fossils that only represent an activity, characteristic of

an organism are called trace fossils. Some example of trace fossils are tracks or trails, burrows and also fossil dung. As a general rule, hard parts are necessary for fossilization for example, teeth and bones of vertebrates, shells of invertebrates, woody parts of plants. It is rare to find soft parts preserved in fine-grained sediments. ***Archaeopteryx*** is the first bird whose feather in present have survived the ages. Sometimes, even fossil faeces called **coprolites** may give us important clues about the food habits of extinct animals.

You will study some of the fossil evidences that are available in the institution where you study, in the form of plaster cast model or pictures. Study them carefully and draw the diagram in your exercise book and note down the characters that are clearly visible to you in the model or picture and try to link up with evidences available.

Note: Before you observe the model or cast of Trilobite you must go through characters and classification of Phylum Arthropoda so that you can correlate the trilobites with the Arthropods.

7.4 PHYLUM ARTHROPODA

7.4.1 Characteristic Features of Arthropoda

1. Body bilaterally symmetrical and metamerically segmented; segments show a tendency to combine or fuse together to form functional units called **tagmata**, like cephalothrorax and abdomen; head and trunk, or head, thorax and abdomen.
2. Segments carry jointed appendages.
3. Exoskeleton consists of a tough cuticle made up of chitin, protein and lipid, sometimes strengthened with calcium carbonate. The cuticle, secreted by the underlying epidermis, is shed periodically to permit growth of body.
4. Absence of cilia.
5. Coelom present but highly reduced and obliterated in the adult. The main body cavity is haemocoel, a characteristic space between organs and tissues, filled with blood.
6. Circulatory system, open type.
7. Well-developed muscular system with striated muscles attached to the exoskeleton, and visceral organs having smooth muscles.
8. Mouth parts modified from appendages; well developed alimentary canal.
9. Respiratory organs are usually tracheae, book lungs or gills.
10. Excretory organs either **Malpighian tubules**, **coxal glands**, **antennary glands** or **maxillary glands**.
11. Nervous system is of the annelidian plan.
12. Sexes are separate; fertilization internal; development often involves metamorphosis.

7.4.2 Classification of Phylum Arthropoda

The phylum Arthropoda has been divided into four sub-phyla as under:

Sub-phylum-Trilobitomorpha

Sub-phylum-Chelicerata

Sub-phylum-Crustacea

Sub-phylum-Uniramia

You will study the characteristic features of sub-phylum Trilobitomorpha in detail in the following subsection.

7.4.3 Subphylum Trilobitomorpha

Subphylum Trilobitomorpha includes the trilobites (Fig. 7.1). **All species are extinct and the fossils indicate that they were all marine forms belonging to Palaeozoic era**. They are the most primitive of all arthropods. Body was divided into three lobes by means of two furrows longitudinally; distinct head, thorax and abdomen were present. Appendages biramous. It appears that trilobites had a variety of habits; they included burrowing, epibenthic. crawling, planktonic and swimming forms.

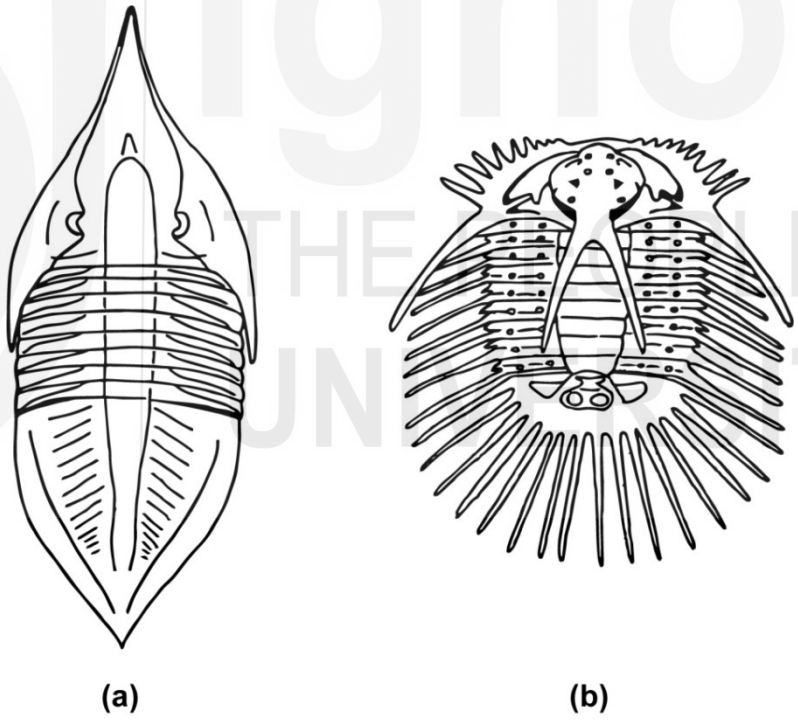


Fig. 7.1: Extinct trilobites (a) *Megalaspis* sp.-burrowing form (b) *Radiaspis* sp.-planktonic form.

7.5 CLASS REPTILIA

7.5.1 Characteristics of Class Reptilia

i) Body is covered with horny epidermal scales. Skin is dry and with few glands.

ii) Two paired limbs are generally present (except in amphisbaenians, legless lizards and snakes).

iii) Skull with one occipital condyle, skeleton is well ossified and ribs are with sternum.

iv) Respiration is by lungs.

v) Heart is three chambered, being four chambered in crocodiles.

vi) Kidneys are metanephric and uric acid is the main excretory product.

vii) There are twelve pairs of cranial nerves.

viii) Sexes are separate and fertilization is internal.

ix) Eggs are laid on land and covered with calcareous or leathery shell.

x) Amnion, chorion and allantois are present during embryonic life. No larval stages.

7.5.2 Classification of Reptilia

The outline of class Reptilia is given in Fig. 7.2.

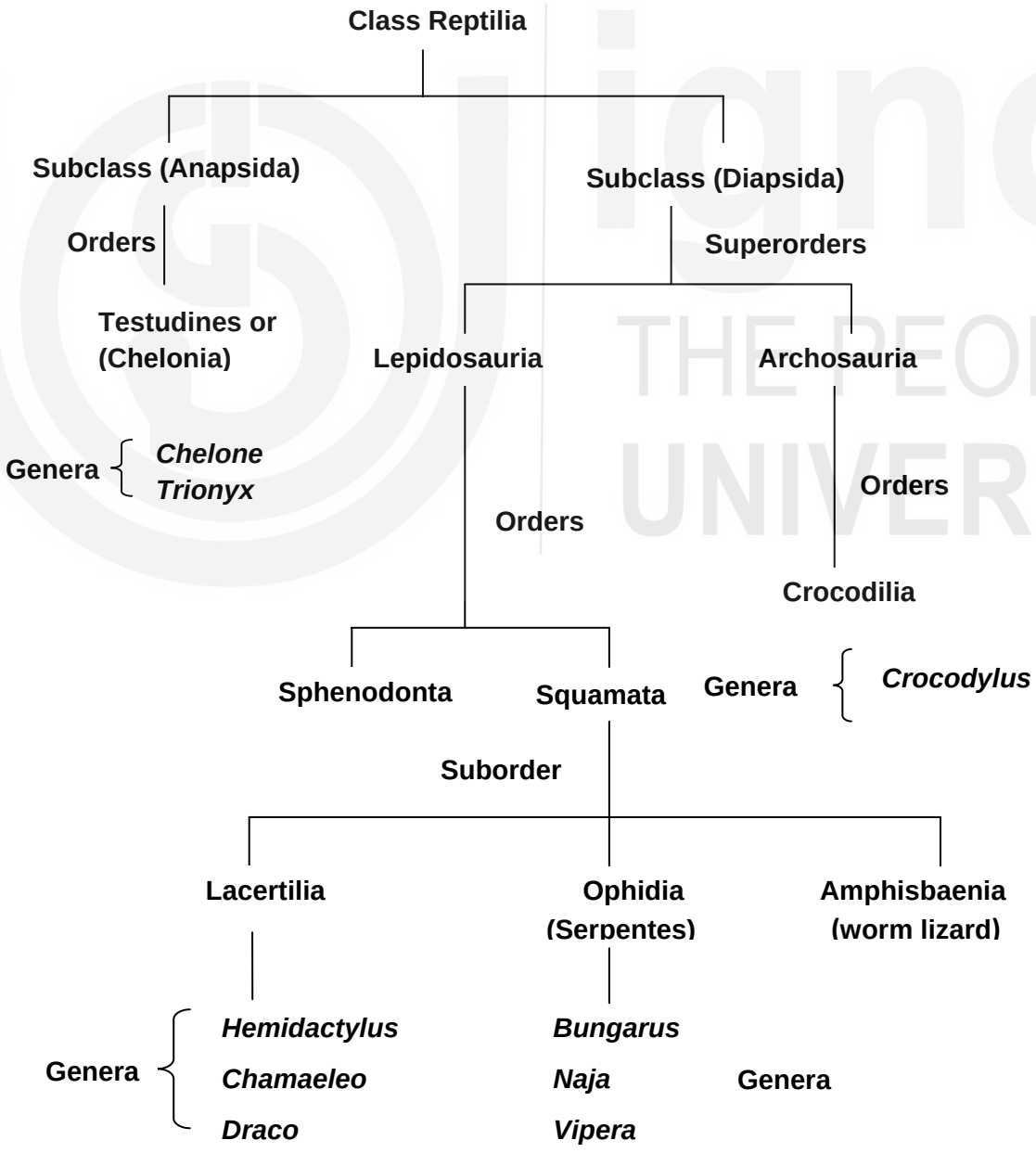


Fig. 7.2: Classification scheme of extant reptiles.

7.5.3 Extinct Reptiles (Dinosaurs): Models

The rise and fall of reptiles occurred during Mesozoic era. Huge reptiles of that era became extinct and fossilised. A few extinct reptiles are as follows represented either by models or pictures.

Class Reptilia

Subclass : **Parapsida** → Temporal openings bounded by supra-temporal and post-frontal.

Genus : *Ichthyosaurus*

1. *Ichthyosaurus*

Comments

i) *Ichthyosaurus* (Fig. 7.2) is a fish-like extinct reptile.

ii) Body divided into head, neck, trunk and forked tail.

iii) Head with large eyes surrounded by sclerotic plates, nostrils, ear openings and long, teethed beak.

iv) Paired limbs paddle-like.

v) Dorsal and caudal fins fish-like.

They appeared in Triassic, reached their climax in Jurassic and became extinct by the end of Upper Cretaceous.

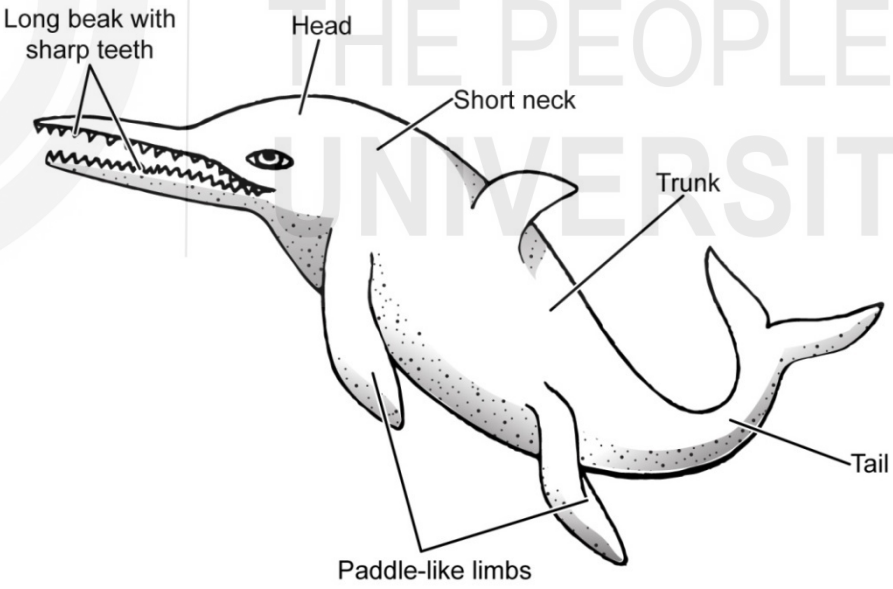


Fig. 7.2: *Ichthyosaurus*.

2. *Dimetrodon*

Class Reptilia

Subclass : **Synapsida** → One Temporal opening behind each eye bounded by post-orbital and squamosal bone.

Genus : *Dimetrodon* (= *Pelycosaur*)

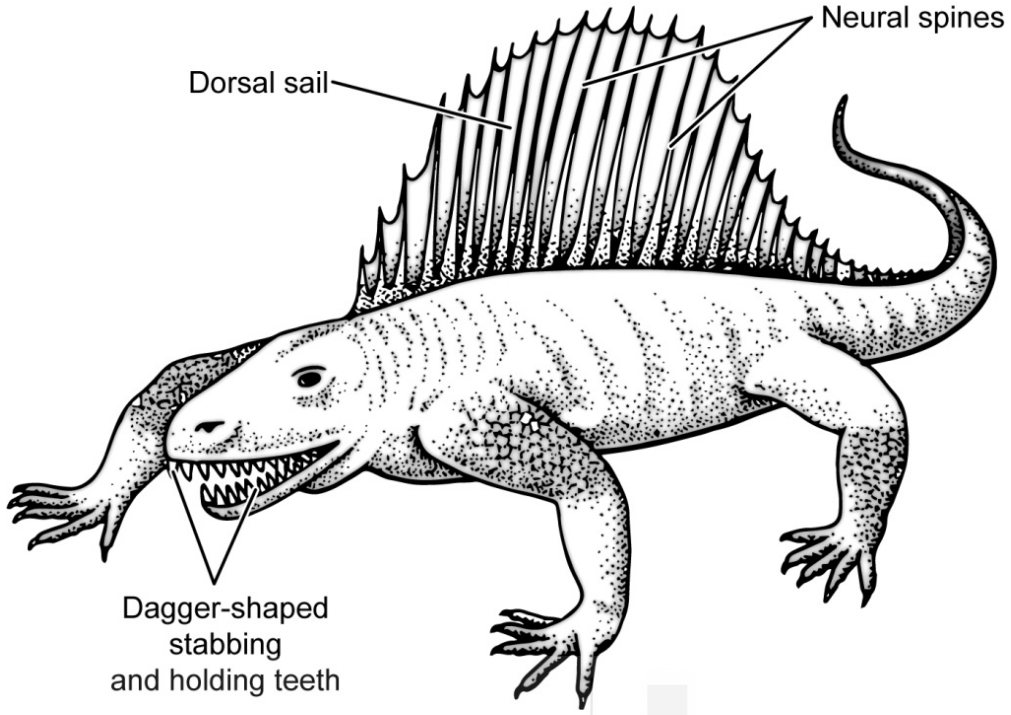


Fig. 7.3: *Dimetrodon*.

Comments

i) *Dimetrodon* (Fig. 7.3) is extinct mammal-like reptile having single lateral temporal opening on either side in the skull.

ii) Body more than 10 feet long and divisible into small head, neck, large trunk and comparatively small tail.

iii) Jaws were provided with dagger-shaped teeth.

iv) Limbs well developed.

v) Dorsal sail over trunk formed by elongated neural spines covered with skin.

vi) Their extinction took place in Jurassic period. They might have given rise to ancestors of mammals.

3. *Brontosaurus*

Class Reptilia

Subclass : **Diapsida** → 2 temporal openings behind each eye separated by post-orbital and squamosal.

Order : **Saurischia** →Ischium and pubis diverge.

Genus : *Brontosaurus*

Comments

i) *Brotosaurus* (Fig. 7.4) is commonly called **giant dinosaur** whose body was very massive.

ii) Body divided into small head, elongated neck, massive trunk and long pointed tail.

iii) Their body size was about 20-25 metre long and weighs about 50 tons.

iv) Hindlimbs large, thick and stout than forelimbs.

v) Pelvis triradiate, Backbone massive with hollowed lateral vertebrae.

vi) Brain small in size.

vii) They lived in swamps in shallows water.

viii) They faced extinction due to small brain and huge massive body.

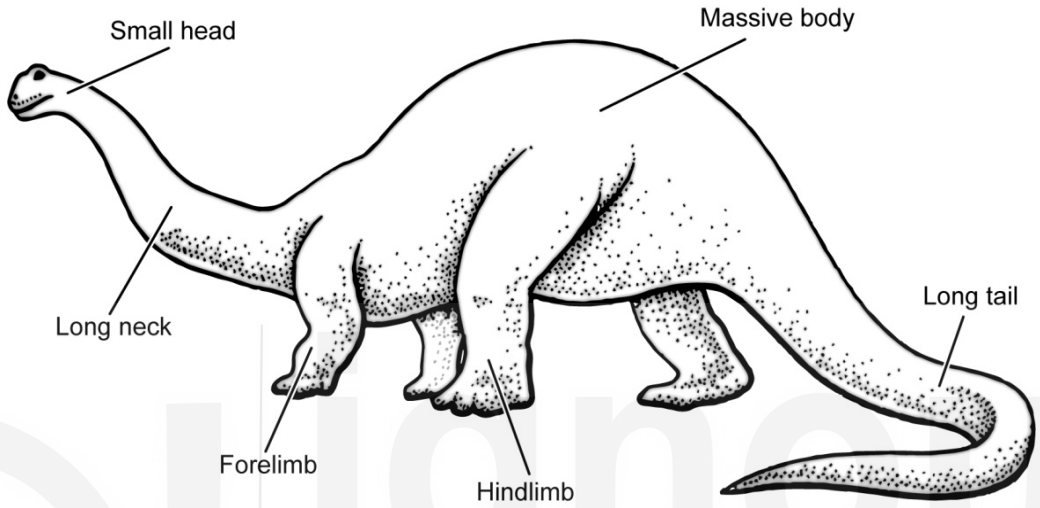


Fig. 7.4: *Brontosaurus.*

4. *Diplodocus*

Class Reptilia

Subclass : **Diapsida**

Order : Saurischia

Comments

i) This was the longest dinosaur of Jurassic period measuring 25 metres long and 50 tons in weight.

ii) Body was divisible into very small head, very long and tapering neck, massive trunk and long massive tail (Fig. 7.5).

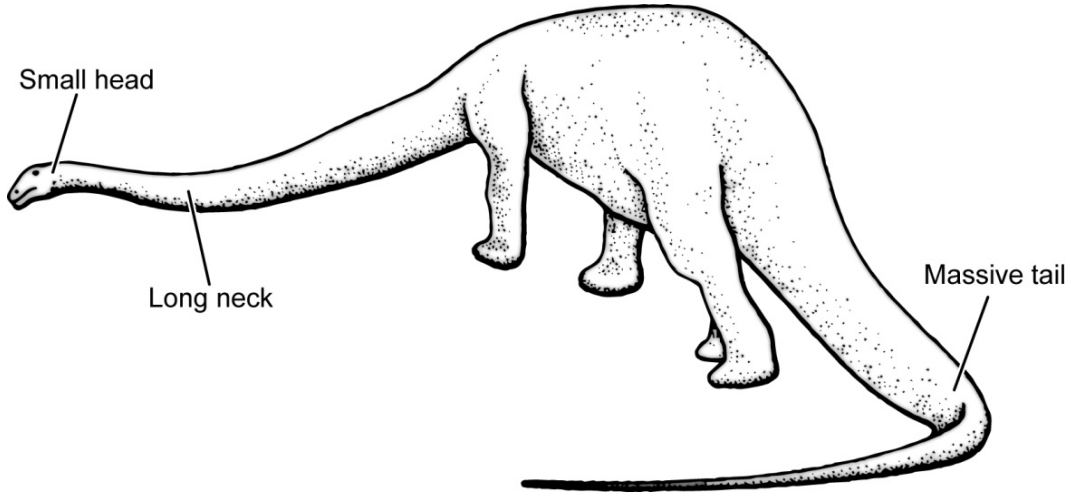


Fig. 7.5: *Diplodocus.*

iii) Jaws small and brain exceptionally small (6 inches long).

iv) Limbs pillar-like and hip region huge.

v) Quadrupedal, herbivorous and swamp dwellers.

vi) They always needed constantly lush green pastures to support their generation due to bulky body.

5. *Tyrannosaurus*

Class Reptilia

Subclass : ***Diapsida***

Order : Saurischia

Comments:

i) This was commonly called **tyrant dinosaur,** lived in plains of Western North America.

ii) Body divided into large head, small neck, massive trunk and tail (Fig. 7.6).

iii) Body was about 15 metres long and 6-7 metres in height.

iv) Forelimbs very small used to hold prey, while hindlegs were adapted for running. Feet were like those of mammoth bird having three powerful claws.

v) Jaws large with 3-6 inches long dagger-like teeth.

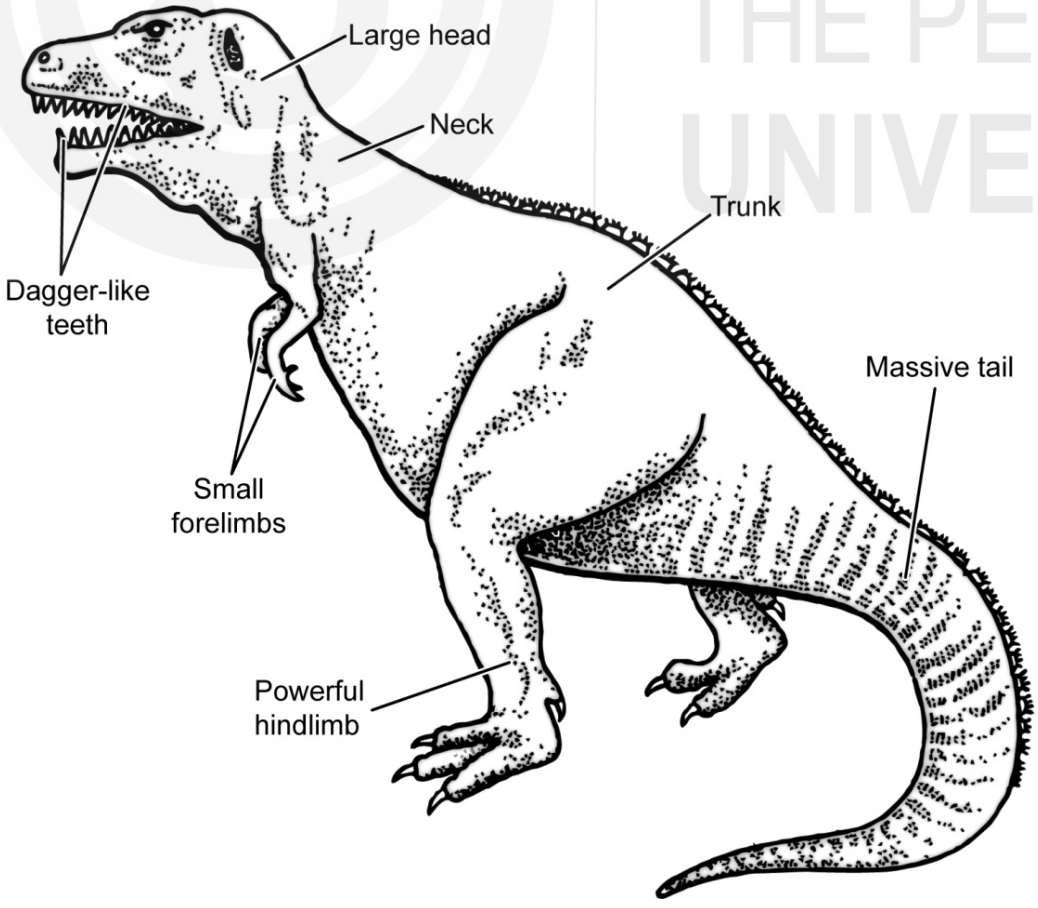


Fig. 7.6: ***Tyrannosaurus.***

6. *Iguanodon*

Class Reptilia

Subclass : **Parapsida**

Genus : *Iguanodon*

Comments

i) *Iguanodon* (Fig. 7.7) is the most primitive bird-like dinosaur with typical tetraradiate pelvis.

ii) Fairly large measuring approximately 5 metres in length.

iii) Body divided into head, neck, trunk and tail.

iv) Head and neck small, while trunk very heavy.

v) Hindlegs massive and forelegs not much reduced.

vi) Peculiar feature of *Iguanodon* was its sharp dagger-like defensive thumb.

They were prevalent in Upper Cretaceous. Descendents of primitive thecodont stock.

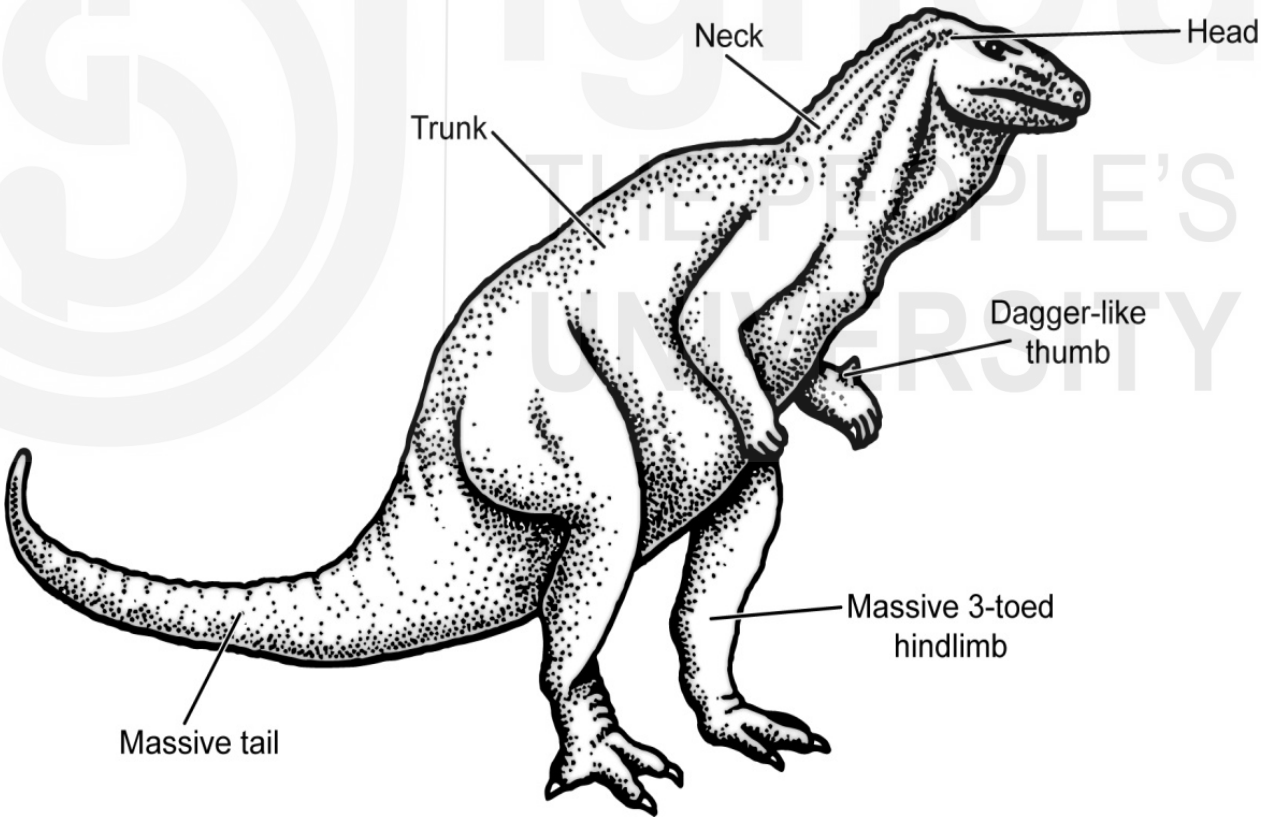


Fig. 7.7: *Iguanodon.*

7. *Stegosaurus*

Class Reptilia

Subclass : **Diapsida**

Order : Saurischia

Comments

i) *Stegosaurus* (Fig. 7.8) is an extinct Jurassic dreaded quadupedal dinosaur.

ii) Body measuring about 8 metres in length and 10 tons in weight.

iii) Head very small with a minute brain, neck and massive trunk and tail were provided with armature of heavy plates and spines.

iv) Trunk's dorsal plates were large and horizontally flattened forming roof. Powerful massive tail had two or more pairs of powerful defensive spines.

v) Spinal cord at the base of tail was tremendously enlarged.

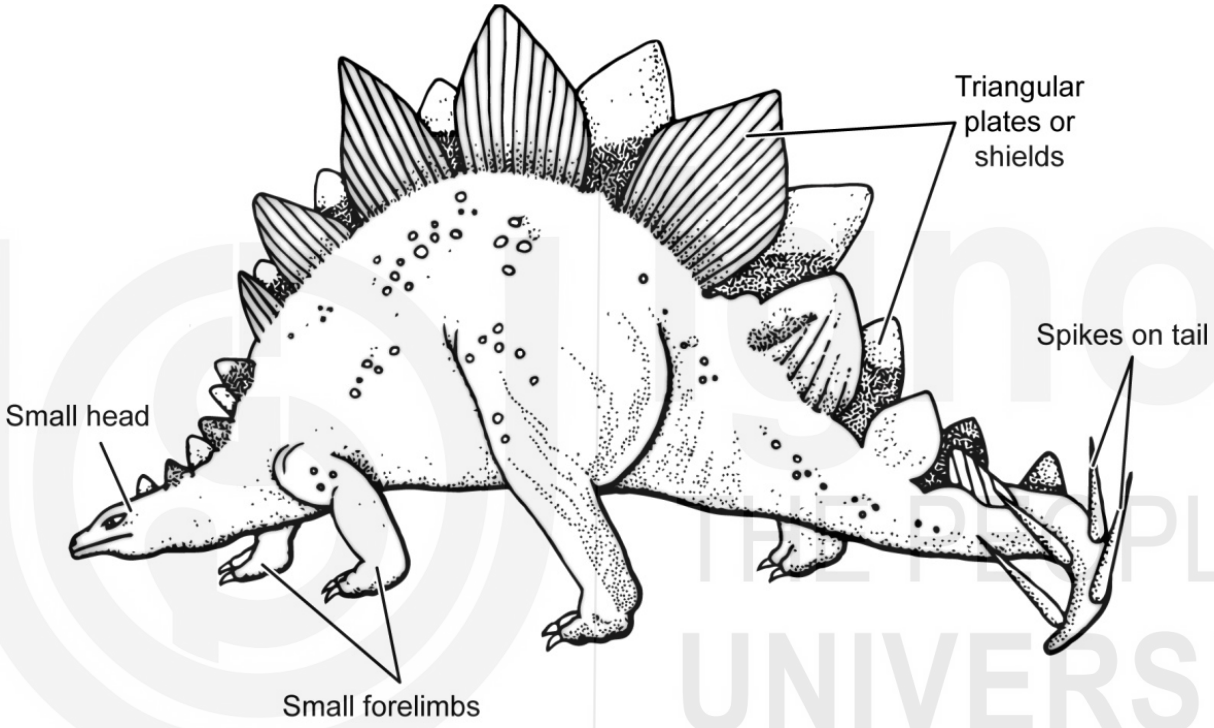


Fig. 7.8: *Stegosaurus.*

8. *Rhamphorhynchus*

Class Reptilia

Subclass : **Semapsida**

Order : **Pterosauria** → Flying reptiles. Forelimbs with wing membranes.

Genus : *Rhamphorhynchus*

Comments:

i) *Rhamphorhynchus* (Fig. 7.9) is commonly called **flying reptile** or **flying dragon.**

ii) Primitive pterosaur had one metre wingspan.

iii) Head with eyes, nostrils, ear openings and beak provided with teeth.

iv) In forelimbs thumbs and three fingers were small and clawed, while fifth finger was very much elongated and attached with body skin forming patagium used as wings during flight.

v) Legs slender with five clawed toes, Tail very much elongated.

vi) Their fossils were obtained from marine rocks.

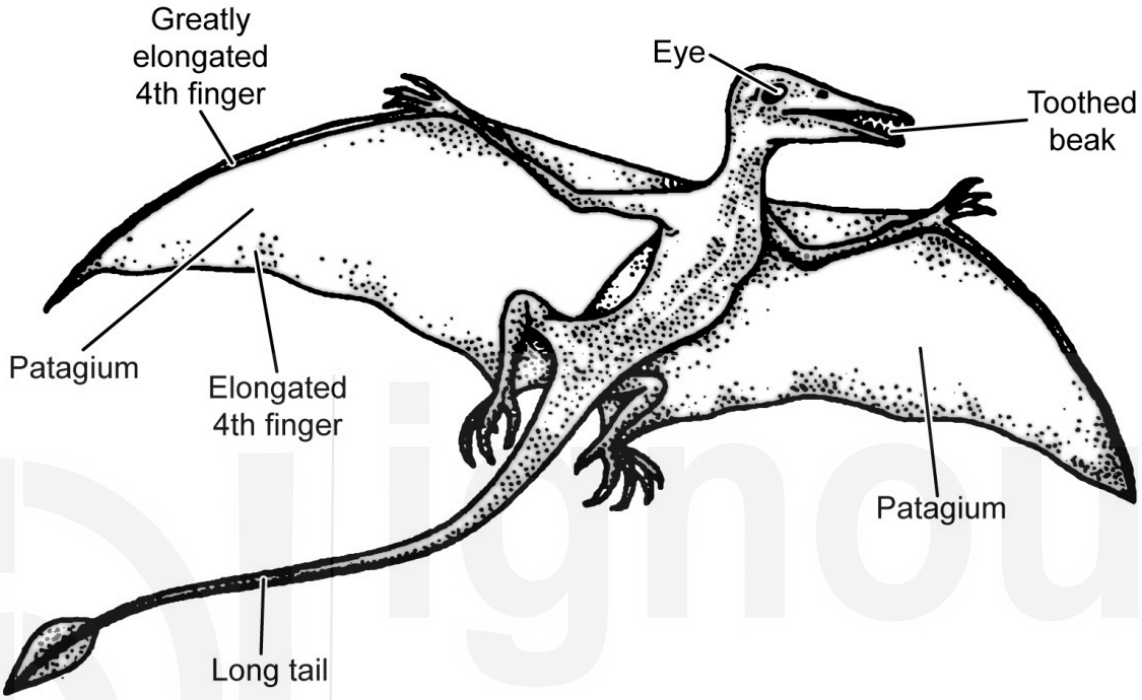


Fig. 7.9: ***Rhamphorhynchus.***

9. ***Pteranodon***

Class Reptilia

Subclass : **Synapsida**

Order : **Pterosauria**

Comments:

i) *Pteranodon* is bird-like giant extinct reptile having wing span about 27 feet.

ii) Body divided into huge head, neck and trunk (Fig. 7.10).

iii) Head contained large eyes with sclerotic plates, nostrils, ear openings and toothless spear-like beak. Head also bears a posterior bony process.

iv) On the back was present a long bony backwardly directed crest.

v) Fourth finger of forelimbs elongated having patagia for soaring in the air.

vi) Brain large with cerebellum very much like that of birds.

vii) They appeared in Jurassic period and became extinct in late Cretaceous, thus, survived throughout Cretaceous.

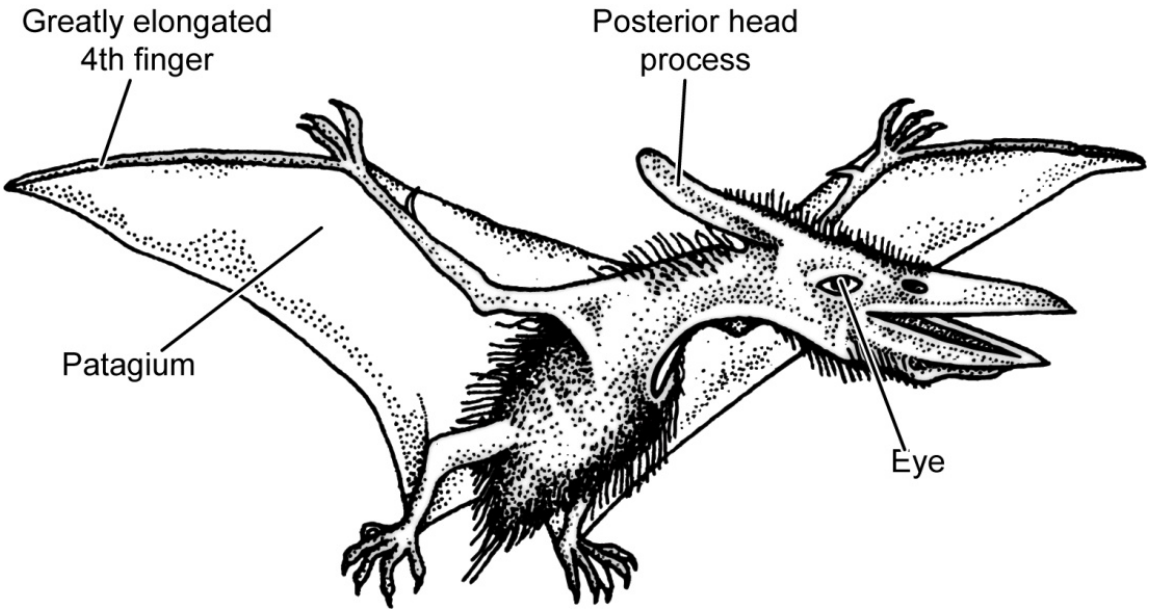


Fig. 7.10: ***Pteranodon.***

7.6 *ARCHAEOPTERYX*: CONNECTING LINK BETWEEN REPTILIA AND AVES

Archaeopteryx lithographica (Fig. 7.11) was discovered in 1861 from Bavaria (Germany), belonged to Jurassic period (140 million years ago).

Class : **Aves** → Warm blooded with an exoskeleton of feathers; forelimbs modified into wings; hindlimbs for walking.

Subclass : **Archaeornithes** → Jaws with teeth and wings present. Tail long with feathers.

Genus : *Archaeopteryx*

Comments

i) Body was divisible into head, neck, breast, abdomen and elongated tail with rectrices.

ii) Head was provided with eyes, nostrils, ear openings and long beak. Jaws were teethed like lizards.

iii) Body was covered with scales. But forelimbs were modified into wings with remiges.

iv) Legs were scaly and toes clawed one behind and 3 anterior.

Archaeopteryx had both reptilian and avian features.

Reptilian features were body covered with epidermal scales, brain simple with cylindrical cerebrum and small cerebellum, jaws with homodont teeth in sockets, vertebrae amphicoelous, sternum without keel, cervical vertebrae 9-10 and caudal vertebrae 20.

Avian characters were feathers over forelimbs (wings) and tail, beak, skull monocondylic, bones spongy, scapula elongated and curved, clavicles fused to form V-shaped furcula, tarsometatarsus present, hallux opposable, and sclerotic ossicles around eyes.

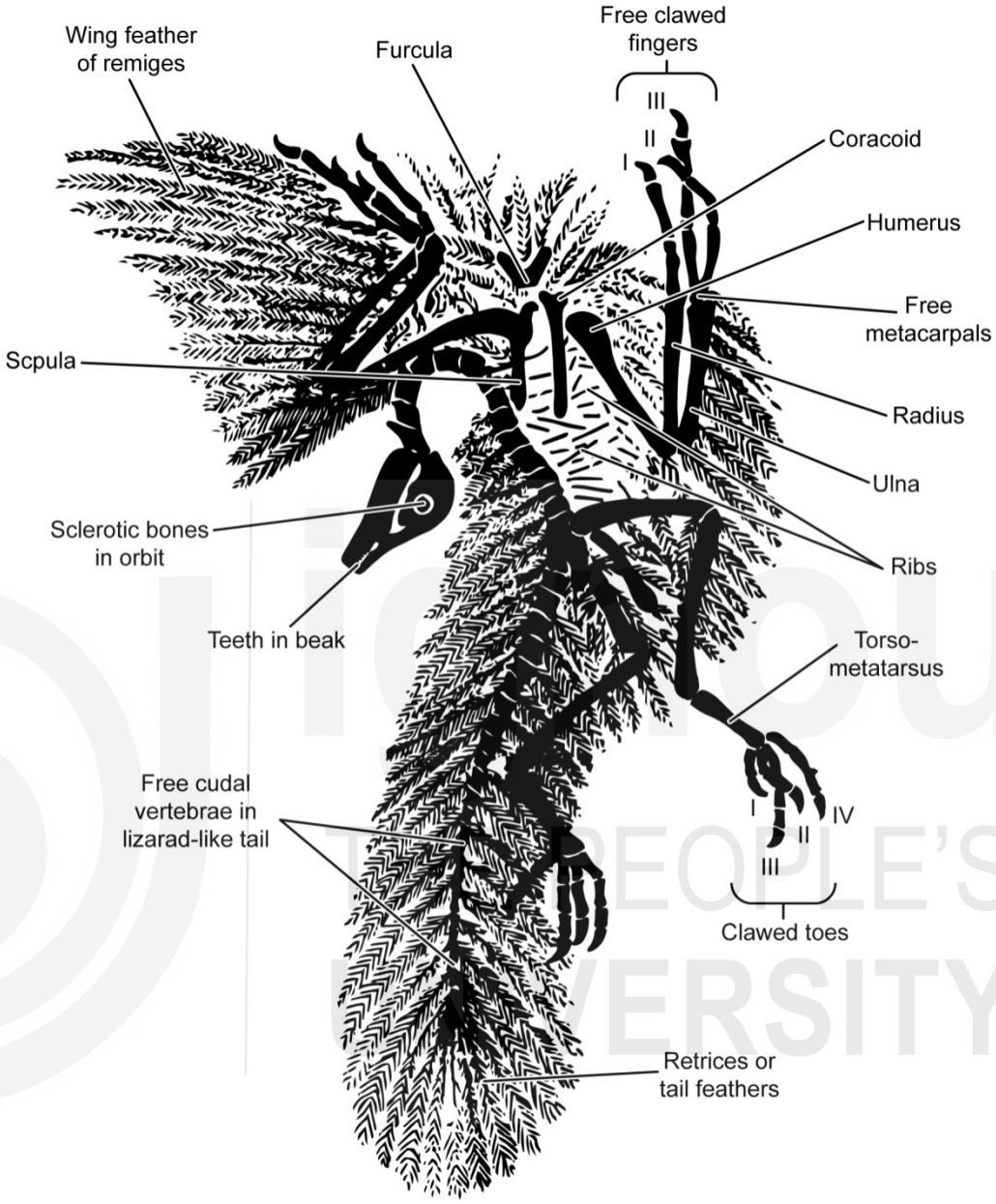


Fig. 7.11: ***Archaeopteryx.***

7.7 TERMINAL QUESTIONS

1. State the significance of *Archaeopteryx* in evolution.
2. Write important characteristics of Trilobites.
3. Write general characteristics of four important dinosaurs of class Reptilia.

EXERCISE 8

STUDY OF HOMOLOGY AND ANALOGY

Structure

8.1 INTRODUCTION

In the previous exercise you have learnt that how the most convincing and direct evidence of evolution comes from the study of fossils that happened to be the records of the organisms of past which remained preserved being buried in the rocky layers. In the present exercise we will try to learn as to how the study of comparative anatomy provides evidence in support of evolution. As you know while classifying organisms, they are put together on the basis of their similarities. But question arises what type of similarities? You know that fish and whales swim in water whereas birds and butter flies pass most of the time in air during flying. One of the early biologists, Pliny, in fact classified animals on the basis of such analogous organs which are organs that serve the same functions and have similar appearance but have different evolutionary origins. With advancement in knowledge about the structure of animals it is revealed that fish and whale or birds and butterflies have more differences than similarities between their body characters. For example fish respire using gills while the whales have lungs for this function. Similarly, birds have an internal skeleton whereas insects like butterflies lack it, instead they have an exoskeleton.

Carl Linnaeus recognised these basic differences and based his classification on the principle of homology. Homologous organs are those with the same evolutionary origin but which may not have the same appearance or serve the same function. For example, in the fore limbs of a human, the wing of bat and flipper of the whale (all mammals) you will observe that in each case, though the function and the appearance of each of the fore-limbs are different, their

basic skeletal plan is same and they are homologous, furthermore, fossil records show that they all have a common evolutionary origin as indicated by the fore-limbs of ancient amphibians. Thus, it becomes evident from a comparative study of the anatomy of different species of organism that they show many structural similarities. This clearly suggests their evolution has taken place by divergence from one or more common ancestors.

In this exercise, you will learn homology and analogy from suitable specimens/pictures and their significance in relation to evolution.

Objectives

After having performed this exercise you should be able to:

- ❖ explain analogous and homologous structures, and
- ❖ interpret their significance in relation to evolution.

8.2 MATERIAL REQUIRED

1. Charts/pictures of homologous and analogous organs
2. Note book
3. Pencil and eraser.

8.3 STUDY OF COMPARATIVE ANATOMY

Based on similarities in the structure and/or function of organs and organ systems in different organisms, biologists have identified three types of structures: **homologous**, **analogous** and **vestigial**. In the following sub-sections you will study these structures and interpret their significance in relation to evolution.

8.3.1 Homologous Organs

Homology is the similarity of structures in various related organisms arising from common ancestory and is usually reflected in common embryological origin. Such structures, called homologous organs, are the consequence of divergent evolution and enable the organisms to carry out different functions.

Superficially, they look different. The study of homologies is a major aspect of comparative anatomy. You may find examples of homologies in organs and organ systems of all groups of living organisms. These homologies provide important evidence for evolution. The forelimbs of vertebrates provide an excellent example of organs with similar structure but different function. The forelimbs of several vertebrates such as those of amphibians, lizards, birds, bats, and men are all constructed on the same basic plan and include similar bones in the same position.

But they have become morphologically different in the course of evolution as a result of modifications to suit different requirements. In Fig. 8.1, you can see some examples of vertebrate forelimbs, modified to suit various functions.

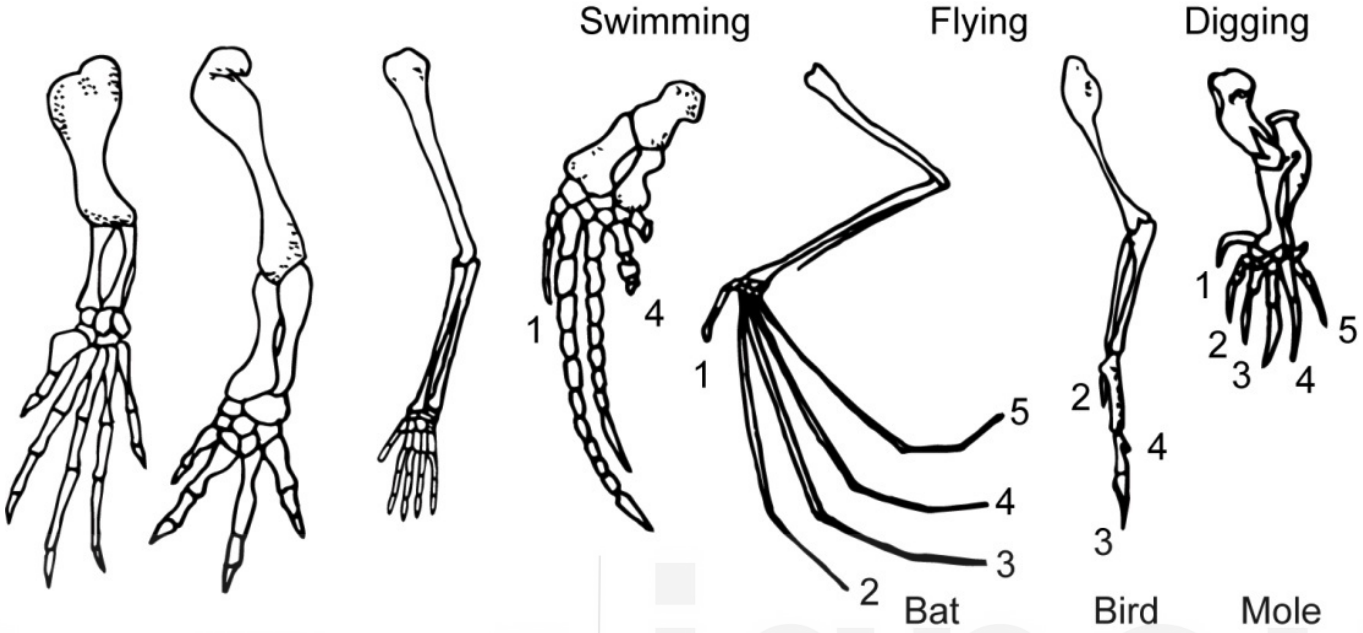


Fig. 8.1: Homology in the bones of the left forelimb in the vertebrates. Similar bones in each animal are adapted for special use by differences in length, shape and bulk of various bones.

A special type of homology is shown by metameric animals. A striking example is that of the appendages of arthropods – especially those of crustaceans. In the typical crustacean, one pair of appendages is borne by each segment of the body. **These appendages have evolved from a single structural plan and are modified in a serial order so as to perform various functions. This is known as serial homology.** You can observe an example of serial homology in the prawn in Figure 8.2.

Metamerism (Segmentation) is the condition of being constructed of linear series of repeating parts, each being a metamere and each being formed in sequence in the embryo.

Homology represents the consequences of **adaptive radiation** from a common ancestor enabling organisms to adapt successfully to different ecological niches. A classical example of adaptive radiation is the structure of the forelimb of mammals (Fig. 8.3). There is always a single long bone, the humerus in the upper arm. In the forearm there are two parallel bones, the ulna and the radius. In the wrist there are typically eight carpal bones arranged as two rows of four. Five parallel metacarpals form the skeleton of the palm of the hand, and rows of three phalanges each from the skeleton of the digits, excepting the first digit which has only two phalanges. Now, you can look at Figure 8.3 for examples. The tenrec (scaly ant eater) of the order Insectivora shows the primitive **pentadactyl** arm structure. The moles, which are their relatives, are highly modified for digging in order to adapt to a subterranean habitat. All the bones of the limbs are short and broad and give the limb a shovel-like appearance. In the bats of the order Chiroptera, the humerus, radius and ulna, as well as four of the digits are greatly elongated so as to support the wing membrane. The forelimb is thus modified for fight. In ungulates like the horse, which are adapted for running or a cursorial habit, the humerus is short and heavy. The remaining bones of the forelimb are generally elongated and the digits are reduced in number. Fusion of bone is

Bats are mammals of the order chiroptera with their forelimbs adapted as wings, they are the only mammals capable of true flight. Their long spread out digits are covered with a thin membrane or **patagium**.

quite common in adults. The details of the structure differ considerably among the various families of ungulates. In aquatic mammals, like the whale, the forelimbs are modified into flippers to aid in swimming.

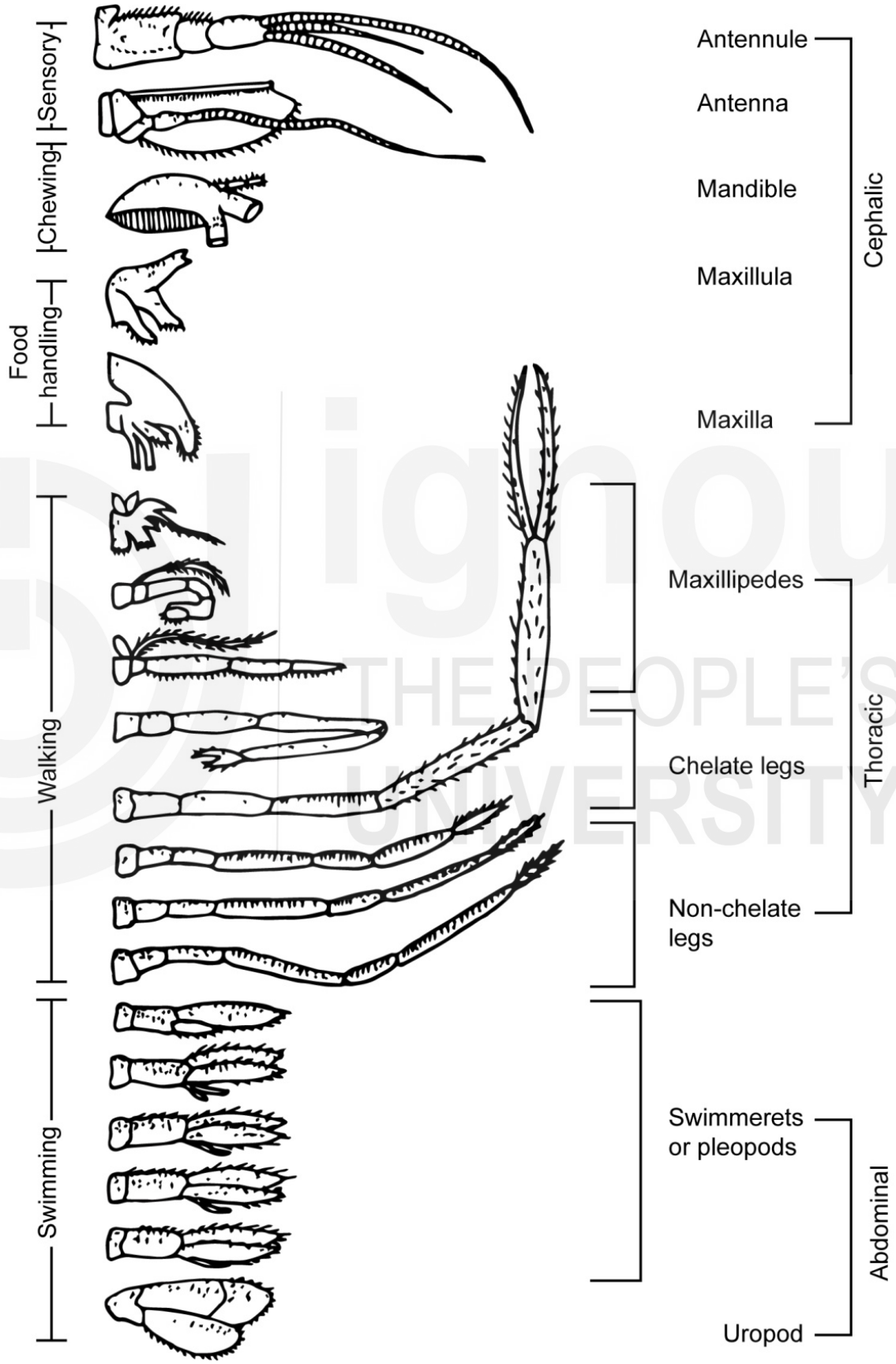


Fig. 8.2: Serially homologous appendages of prawn. Note the structural modifications to suit the various functions performed.

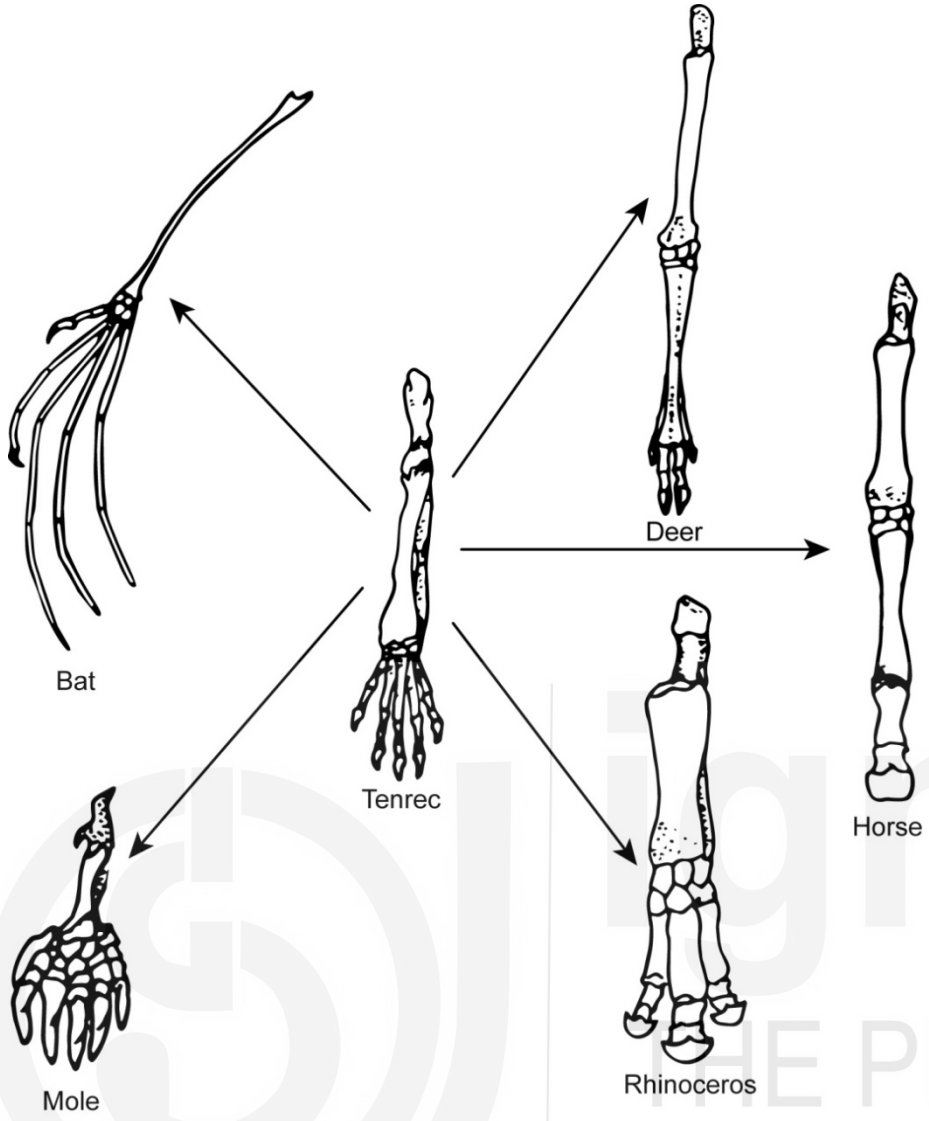


Fig. 8.3: Adaptive radiation in the forelimbs of mammals.

In plants also, homologous structure can be observed. Examples are: thorns of bougan villia, thorns of rose and tendril in cucurbits because they grow in axillary position.

Many examples are found where diversity of structures is there but their fundamental organisation is same. Organisms of a taxonomic category have same basic plan, but there are differences among members to suit their specific needs.

Higher is the taxonomic class, more are the differences among members most of the biologists after Darwin are the assumptions that closely related anatomical structural resemblance is due to close genetic similarity. More the genetic differences, less similarities will be there.

8.3.2 Analogous Organs

Analogous organs are morphologically different structures which develop in various unrelated organisms serving similar functions. However, there are a certain similarities in these structures which are based on adaptations to perform the same functions. These organs, called analogous organs, are the consequence of convergent evolution.

A classical example of analogous organs is the set of wings developed independently by insects, some extinct reptiles as well as birds and bats. As shown in Figure 8.4 the insect wing is a membrane supported by chitinous veins. The entire wing is a lifeless structure operated by muscles attached to its base. The wings of pterosaurs, extinct flying reptiles, were formed by a fold of skin supported by an enormously enlarged fourth digit of the forelimb. In birds, the planing surface of the wing is composed of feathers.

The feathers are supported by an internal skeleton of bones of the forelimb. In bats the wing is formed by a membrane modified from skin. The wing in the bat is supported by the elongated and outspread phalanges of the last four digits of the forelimbs.

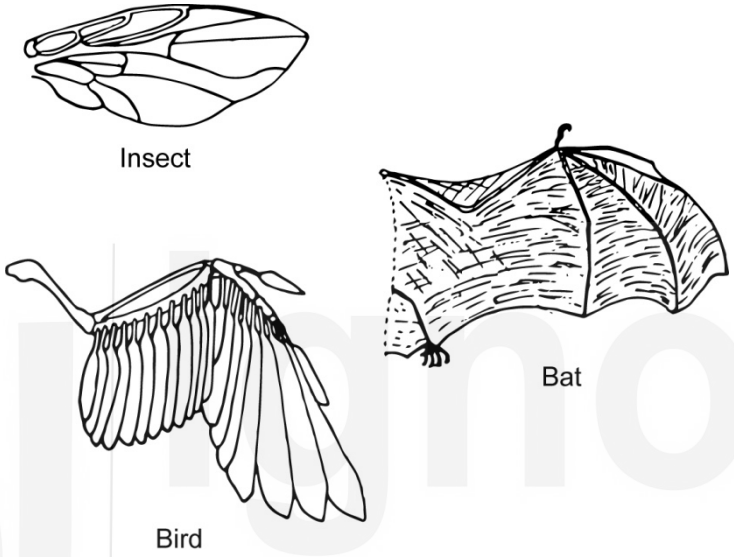


Fig. 8.4: Analogy between wings of insect, a bird, bat and pterosaur. In each the planning surface is formed from different materials and the resemblances are only due to functional adaptations

Analogy in body shapes

A fish, an ichthyosour and whale have stream-lined body and are adapted for aqueous existence but these belong to three different classes of vertebrates with no ancestry traces.

Analogy in fin of fishes and flippers of whale

Anal fins of fish and flippers of whale are completely unrelated structures but have similar appearance and perform the same function to help in swimming. Their structural details are totally different.

8.4 INFERENCE

With the examples given above we can infer that evidences provided to us in the form of homologous and analogous structures support the organic evolution.

8.5 TERMINAL QUESTIONS

1. Compare homologous and analogous organs with suitable examples.
2. Explain the adaptive radiation in the forelimbs of mammals.

EXERCISE 9

TO STUDY THE PHYLOGENY OF HORSE WITH DIAGRAMS / CUTOUTS OF LIMBS AND TEETH OF HORSE ANCESTERS

Structure

9.1 INTRODUCTION

In the previous exercise you have learnt the homology and analogy from the comparative anatomy of different species of organisms as they exhibit many structural similarities suggesting their evolution from divergence from one or more common ancestors. In this unit you will learn the phylogeny of horse with diagrams/cutouts of limbs and teeth of horse ancestors.

As you have studied an important line of evidence which Darwin carefully built in support of his thesis of evolution through variation and natural selection that comes from a knowledge of the distribution of contemporary as well as species of plants and animals; found as fossils.

The phylogeny of the horses was the first to be deduced from the fossil record. The record shows us that during the course of evolution every part of the skeleton of the horse was affected. In this exercise we will trace evolutionary history of the horse with emphasis on the events that led to modification of limb and teeth of horse.

Objectives

After having performed this exercise you should be able to:

- relate the presence of fossils on the earth's crust to the occurrence of organic evolution of horse limbs and teeth, and
- piece together events in development that took place during the evolution of horse.

9.2 MATERIALS REQUIRED

1) Chart depicting evolution of horse
2) Note book
3) Pencil and eraser

9.3 EVOLUTION OF HORSE

The evolution of the horse that is a mammal of the family **Equidae,** took place over geological period of 50 million years which transformed the small sized dog to a forest dwelling ***Eohippus*** and finally transforming into a modern horse. Those paleontologists working in the field have been able to piece evidences together to a more complete outline of evolutionary lineage of the modern horse as compared to any other animal. Much of this evolution occurred in North America where the origin of horse had taken place but they became extinct about 10,000 years ago (Fig. 9.1).

As a matter of the fact, the horse belongs to the order **Perissodactyla** (odd toed ungulates), of which all the members share hooved feet having odd number of toes on each foot as well as mobile upper lips with similar structure of tooth. This alludes that **horses share a common ancestry with tapirs and rhinoceros**. The perissodactyls originated in late Paleocene, less than ten billion years after the Cretaceous – Paleocene extinction event. The perissodactyle group of animals seem to have been originally specialised for life in tropical forests, whereas tapirs and rhinoceros retained their specialisation for jungle. Modern horse is adapted to life on drier land in the much harsher climatic condition of the steppes. The other species of ***Equus*** are adapted to a variety of intermediate conditions.

Thus early ancestors of modern horse walked an several spread out toes, an accommodation to life spent walking on the soft moist grounds of primeval forests. By then species of grass began to appear and flourish. The equids diets shifted from foliage to grasses, leading to longer and more durable teeth. And at the same time the steppes began to appear the horse's predecessors required to be capable of greater speeds to outrun predators. This was attained through the lengthening of limbs and the lifting of some toes from the ground in such a way so that the weight of the body was gradually placed on one of the longest toes, the third toe.

9.4 EVENTS IN EVOLUTIONARY HISTORY OF HORSE

The history of horse evolution dates back to the beginning of **Eocene period** of **Coenozoic Era.** The primary centre of their evolution was great plains Region in North America, from where some of the species hove spread out to Europe and Asia from time to time. First known ancestors of horse were fox like, living on moist ground and browsing soft leafy vegetation. The history of evolution of horse presents definite changes in certain organs in a particular direction. These directional changes are known as **evolutionary trends.**

In this Section we will briefly go through the events that took place in the evolutionary history of horse.

- A general increase in size.
- Enlargement of the brain (especially the cerebral hemispheres) and a corresponding increase in the size of the head.
- Increased length and mobility of the neck.
- An enlargement of the last two, and finally, of the last three premolars until they became comparable to the molars.
- Elongation of the limbs for speedy running, but with a loss of rotational movement.
- Fusion of bones in the limbs to provide better hinge joints. This also makes for more efficient support of the body weight.
- Reduction of the toes from five to one long toe (third) on each foot, which is covered by a hoof (claw). The lateral toes are gradually reduced and finally only small bones of the second and fourth toes persist as splints.
- Thus, in the limbs of horse the number of digits is reduced to one, usually the 3rddigit is retained. The limbs are elongated by fusion of persisting metacarpals to form cannon bones. This has resulted in unguligrade type of locomotion. Since the part of limb contracting the ground is reduced there is less friction attaining a greater speed. In this way the horse is able to gallop at greater speed.
- By these changes during its long evolutionary history, the horse became a long-legged, swift-running mammal adapted to live and feed on open grasslands. The prominent teeth having many enamel ridges help grind tough grassy vegetation.
- You can see in Figure 9.1, a brief sketch of the fossil record of the horse which covers 60 million years and includes five continents. It documents evolutionary trends and patterns of diversity through time, patterns of origin and extinction and even migration between the continents.

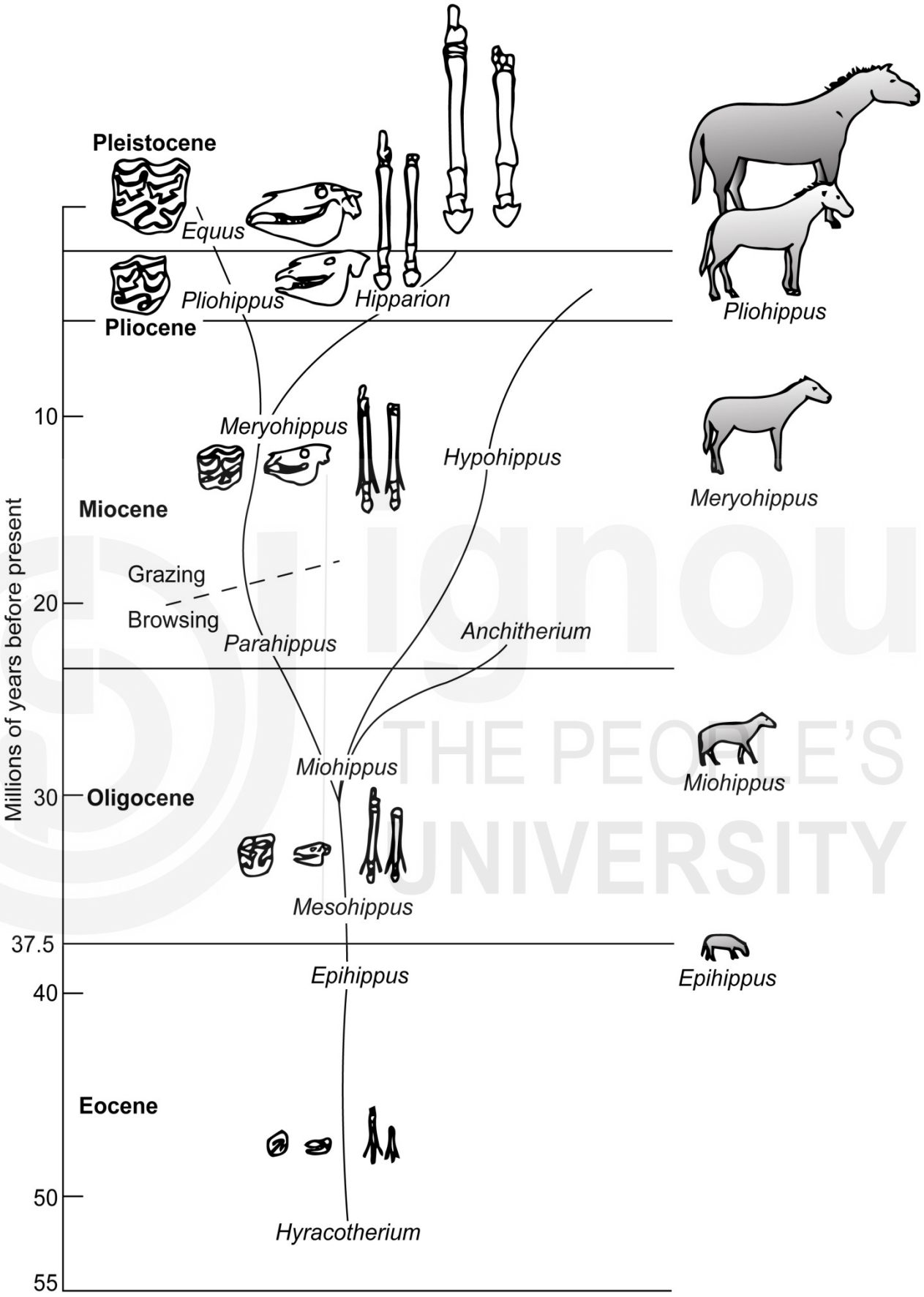


Fig. 9.1: Schematic diagram showing the evolutionary trends in horse within a span of 60 million years. Note the diversity of horse during Miocene and Pliocene period and extinction of all other horses except for single toed species by the end of Pliocene.

9.5 INFERENCE DRAWN

Thus we can say that the present day modern horse *Equus* has evolved from *Eohippus* through transitory phases of *Miohippus, Merychippus, Pliohippus*.

9.6 TERMINAL QUESTIONS

1. Explain the modifications of teeth and limbs during the course of evolution of horse.
2. “Horses share a common ancestry with tapirs and rhinoceros”. Explain this statement.

EXERCISE 10

STUDY OF DARWIN'S FINCHES WITH DIAGRAMS/CUTOUT BEAKS OF DIFFERENT SPECIES

Structure

10.1 INTRODUCTION

In Exercise 9 you have learnt as to how the horses have evolved from miniature forest-dwelling *Eohippus* into modern horse. In the present exercise you will study about the Darwin's finches that are also known as the **Galapagos finches** with the help of diagrams/cut outs of beaks of different species.

During his voyage on the **Beagle** (name of ship of Darwin), Darwin observed that oceanic islands lying beyond the continents account for much less number of naturally occurring species of organisms. Many of the species on these islands are **endemic,** that is they are found nowhere else.

Darwin found 15 species of land birds in the islands of the Galapagos archipelago. Of these 21 to 23 were found to be endemic. But of 11 species of marine birds found there, only 2 were endemic. In Figure 10.1, you will see the heads of the birds of the subfamily Geospizinae, showing the different beak structures. This difference is correlated with different feeding habits, which enabled them to occupy different ecological niches within a restricted area.

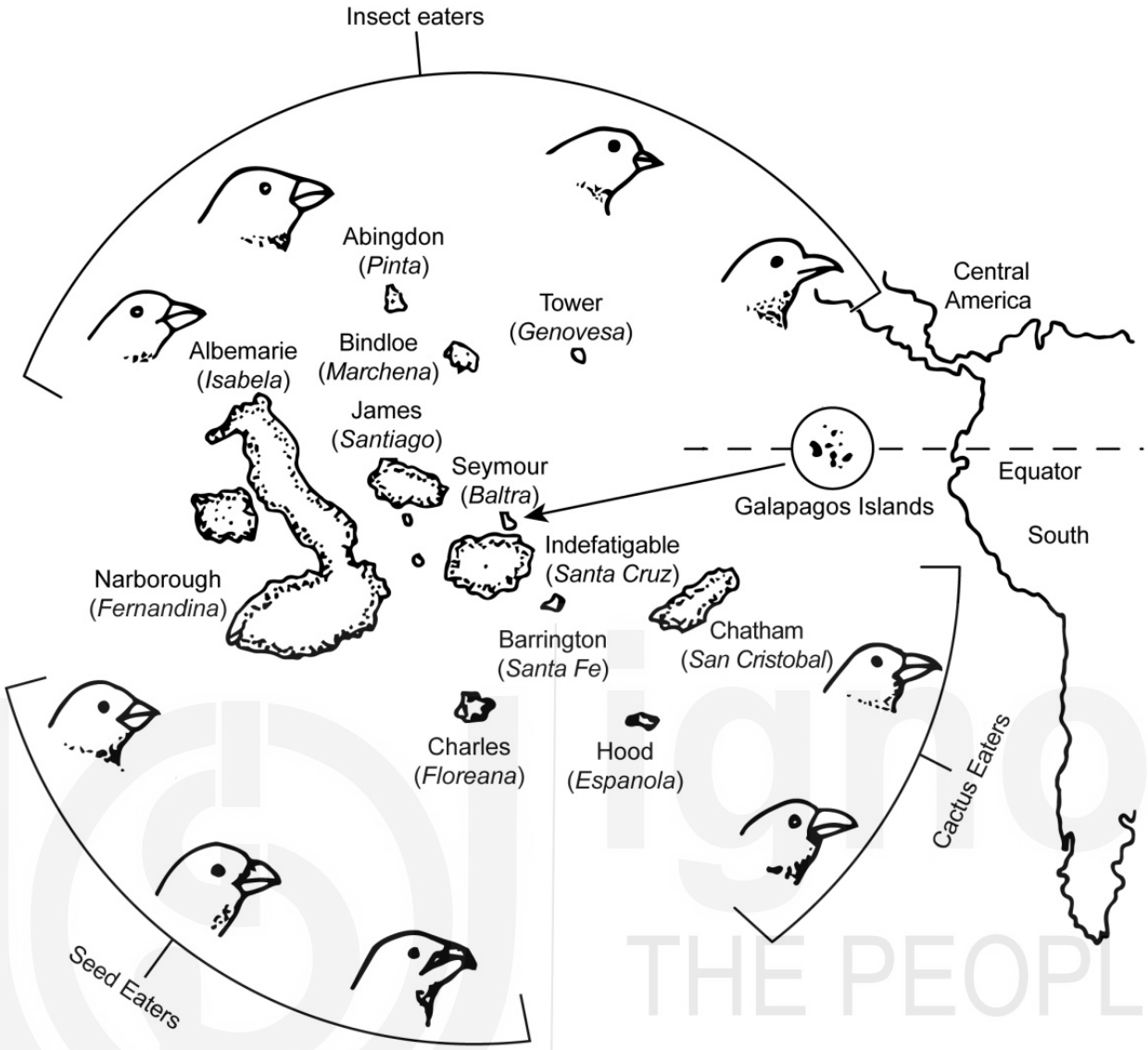


Fig. 10.1: Galapagos islands and different beak structures of birds of subfamily Geospizinae, owing to different feeding habits. Galapagos islands are situated between 500 and 600 miles west of South America.

In another example, Galapagos Islands include 436 species of flowering plants. Of these 223 species are endemic. Many of these are restricted to one or a few islands in the archipelago. Struck by the endemic nature of the flora and fauna of Galapagos Islands, Darwin proposed that islands were colonized by occasional migrants from the mainland. In due course of time their descendants would have been modified, eventually evolving into new and distinct species. As they spread to the various islands of the archipelago, each isolated population would have been modified independently, thereby forming groups of closely related endemic species. Also the wide water barrier would greatly reduce the probability of these new species spreading to other localities. For marine birds, such a barrier is less formidable. It is because of this that you find a smaller proportion of endemics amongst the marine birds.

At first glance the contemporary species on the Archipelago and their continental counterparts may appear wholly unrelated. However, on the basis of their similarities it should be clear to you that sometime in the geological past they must have shared a common ancestor.

A long-term research study was carried out for more than 40 years by the research of the Princeton University. However, Peter and Rosemary Grant have documented evolutionary changes in beak size affected by El Nino/La Nina cycles in the Pacific ocean. In the present exercise we will try to learn about Darwin's finches with the help of diagrams/cutouts of beaks of different bird species.

Objectives

After having performed this exercise you should be able to:

- ❖ define the term Darwin's finches,
- ❖ analyse the large collection of museum specimens/cut outs of beaks of different birds species,
- ❖ differentiate between different species on the basis of size and shape of their beaks, and
- ❖ correlate their adaptation to different food habits.

10.2 MATERIAL REQUIRED

1. Chart/diagrams/cutout of beaks of different species of birds
2. Note book
3. Pencil and eraser.

10.3 DARWIN'S FINCHES

Darwin's finches also known as Galapagos finches happen to be a group of about 15 species of passerine birds that are known for their diversity in beak form and function. They are classified as the sub-family **Geospizinae** or tribe Geospizini, the closest known relative of Galapagos finches is South American ***Tiarisobscurus***. These birds were first collected by Charles Darwin on Galapagos Islands during his second voyage on the Beagle. Besides Cocos finch that was collected from Cocos Island, the others were found on the Galapagos Islands (Fig. 10.2). Darwin observed that the finches on mainland of South America were all of one type, possessing short, straight beaks of seed crushing. These birds from various islands of Galapagos were different in size and shape of the bill due to adaptation to different food types available. Darwin differentiated 13 species of finches and grouped them into 6 main types:

- Large ground finches
- Cactus ground finches feeding on cacti
- Warble finches
- Insectivorous tree finches
- Vegetarian tree finches
- Wood pecker finches

Darwin's finches offer an excellent example of adaptive radiation. The ancestral finches on reaching different islands occupied all empty ecological niches in absence of competition and evolved into different species.

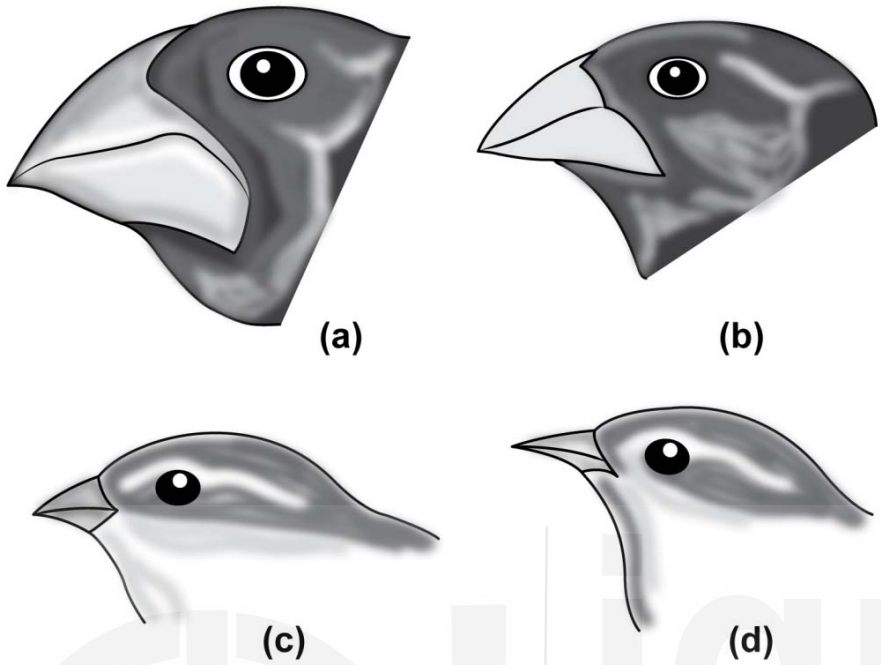


Fig. 10.2: Darwin's finches. a) Large ground finch, b) Medium ground finch, c) Small tree finch, and d) Green warbler- finch.

Scientific classification

Kingdom	**:**	**Animalia**
Phylum	**:**	**Chordata**
Class	**:**	**Aves**
Order	**:**	**Passeriformes**
Family	**:**	**Thraupidae**
Genera	**:**	***Geospiza***
		Camarhynchus/Patyspiza
		Certihidea
		Pinaroloxias

The term "Darwin's finches" was first used by Percy Lowe in 1936, and popularized in 1947 by David Lack in his book *Darwin's Finches*. David Lack based his analysis on the large collection of museum specimens collected during 1905-06 Galapagos expedition of the California Academy of Sciences, for which Lack dedicated his book in 1947. The birds varied in size from 10 to 20 cm and weighed between 8 and 38 grams. **The smallest are the warbler-finches and the largest is the vegetarian finch**. The most important difference are noted between species in the size and shape of their beaks, that were highly adapted to different food sources (Fig. 10.3). The birds were all dull-coloured.

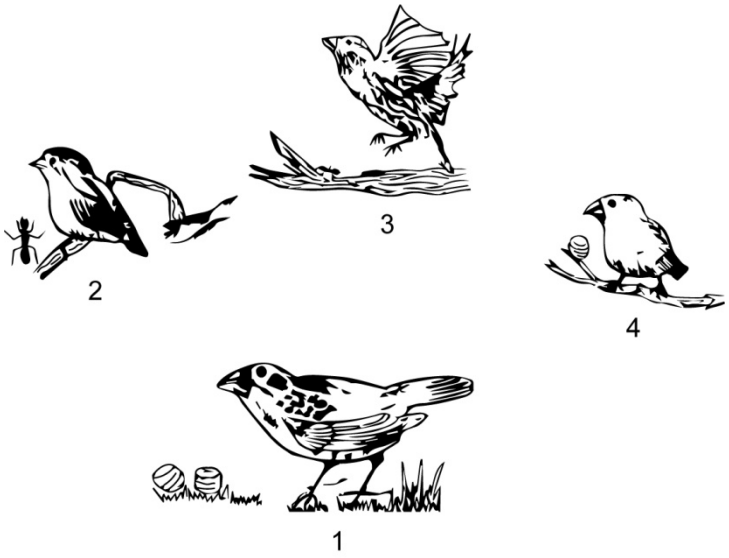


Fig. 10.3: Shown here is adaptive radiation of finch A. (*Geospiza magnirostris*) into three other species of finches found on the Galapagos Islands. Due to the absence of other species of birds, the finches adapted to new niches. The finches beaks and bodies changed allowing them to eat certain types of foods such as nuts, fruits, and insects. 1. *Geospiza magnirostris,* 2. *Geospizaparvula*, 3. *Certhideaolivacea* and 4. *Geospizafortis*.

10.4 INFERENCE

While going through diagrams/cut outs of beaks of different species of birds it is inferred that the diversity in the size and shape of beaks are owing to different feeding habits as suggested by the biologists.

Evolution favours appearance of similar forms under similar environmental conditions. **Convergence or convergent evolution is the evolution of similar forms of different lineages when exposed to the same selective pressure. Divergence or divergent evolution is the evolution of different forms in the same lineage when exposed to different selective pressure.**

Tortoise and finches on Galapagos Islands present divergent evolution.

10.5 TERMINAL QUESTIONS

1. Which factors influenced the size and shape of Darwin Finches?
2. Explain adaptive radiations of Galapagos finches.

EXERCISE 11

AN EXERCISE TO DEMONSTRATE THE ROLE OF NATURAL SELECTION IN EVOLVING ADAPTATIONS

Structure

11.1 INTRODUCTION

In course on Evolutionary Biology (BZYCT-133) you have learnt about the role of natural selection in the evolutionary process. You have studied that in a population natural selection promotes those alleles that confer an adaptive advantage on the individuals who possess such alleles. We further discussed the examples of *Biston betularia* (peppered moth) and sickle-cell anemia to illustrate the positive role played by natural selection in evolving adaptations. You may recall that in England the melanic (dark coloured) forms of *Biston betularia* resting on soot covered trees increased in numbers in post-industrial revolution period while the number of non-melanics was higher in pre-industrial revolution times when the trees were lichen covered. The reason is that the melanics easily escaped predation in an environment to which they were adapted best. Natural selection promotes in a population those characters that are better adapted to the environment. African populations maintain the sickle cell allele in heterozygous condition (HP^A/HP^A – normal homozygous genotype, Hb^A/Hb^s-heterozygous genotype and Hb^S/Hb^S sickle cell genotype). The hetetozygotes do not contract either malarial disease or sickle cell anemia. They are better adapted to live in an environment in which both the killer diseases are prevalent. In this simple exercise you will learn the probable role of selection process in evolving meaningful adaptations in a population.

Objectives

After having performed this exercise you should be able to:

- make use of simple devices to illustrate the concept of natural selection,
- discuss that evolution of adaptations is a non-random process, and
- relate the illustration presented here to real-life situation.

11.2 MATERIAL REQUIRED

1. White card-board squares of side 2 cm – 260 pieces.
2. Plastic bowls-2.

11.3 PROCEDURE

You require 260 white card-board squares of 2 cm side, 10 for each of the 26 alphabets. This means you have with you 10 As, 10Bs. 10Cs and so on upto 10Zs. You have 260 letters with you.

Situation I

1. Leave all the 260 cards in a plastic bowl; mix well. Your task would be to pick out any three cards at a time and get a meaningful word, let us say CAT.
2. If you do not get the word CAT, discard the three cards into another bowl. You continue to do this exercise until you get the required word. You will find that the three letters C, A and T shortly disappear without the word CAT being formed. Instead, you may form all types of meaningless combinations such as ALC, TXP, BYA, so on and so forth. After 86 draws, the bowl is almost empty with only two cards lying there and the word CAT not yet formed. Barring a miracle, you may not be able to form the word even in hundreds of such a draw.

Situation II

Now let us have the rules of the game slightly changed. When you make your draw of three letters and when any of the required letters C, A and T appear in any combination, return these letters to the bowl and discard only the other letters. Assuming you draw XAP, return A to the bowl and put aside P and X. Or, if you draw TUC return T and C to the bowl and put aside U. By this process, the letters C, A and T will start accumulating in the bowl, even as the other letters are gradually lost. Sooner you will have 10Cs, and l0As and l0Ts in the bowl. Naturally, the meaningful word CAT will be picked up in a few draws. Record the number of draws you have made to get the right word.

Situation III

Now let us introduce yet another change in the rules of the game. When you draw three letter combinations such as QTC, AWT etc., discard the non-essential letters such as Q and W as you did earlier. But before returning the essential letters back to the bowl, clip them together. Use a gem clip for this purpose. Thus, T and C will be clipped together and so also the letters A and T. Now when you draw the word CAT from the bowl, you will succeed in doing so in less number of draws. Record this observation also.

11.4 INFERENCE DRAWN

What do you infer from the above three situations? In situation I, the natural selection is not operating. The letters C, A and T do not have any special advantage over the other letters. As a result the meaningful word CAT or the adaptation is not emerging.

In situation 11, natural selection is operating. The letters C, A and T which contribute to the adaptation are retained in the population. In other words, the three letters are favoured more than the rest of the alphabets. The fact that you return the three letters to the bowl symbolises the retention of the adaptation in the population. Under the influence of natural selection, a meaningful adaptation (here the meaningful word CAT) emerges.

In situation III, you clip together the two essential letters as and when they are picked up (such as C and T, and A and T). This clipping together signifies another process that naturally occurs in the population-a natural genetic phenomenon known as **inversion**. It is known that inversions are suppressors of crossing over. Inversions therefore hold together adaptive genes in a tight linkage. The two letters C and T or A .and T clipped together represent linked genes and are prevented from recombining with other letters.

1. What do the three situations considered together signify?
2. Why are inversions regarded as suppressors of crossing over event?

Pericentric
Inversions include the centromere while **paracentric** inversions occur outside of the centromere. A pericentric inversion can change the length of the chromosome arms above and below the centromere.

11.5 TERMINAL QUESTIONS

1. “Evolution of adaptations is a non-random process”, Discuss this statement.
2. What is the relationship between sickle shaped RBCs and malaria?

EXERCISE 12

AN EXERCISE TO DEMONSTRATE THE ROLE OF NATURAL SELECTION IN FIXING FAVOURED ADAPTATIONS AND ELIMINATING MALADAPTATIONS

Structure

12.1 INTRODUCTION

In the previous exercise we illustrated the role of natural selection in evolving adaptations. In effect this means that occurrence of those variations which are less adapted to an environment will be slowly minimized in the population. In the example of *Biston betularia*, the non-melanics in a non-conducive environment dwindled in numbers over a period of time since they were easily located by the birds and were predated upon. This was also true of melanics which rested on lichen covered trees and therefore were easily sighted by birds. **The elimination of an allele does not occur unless there is an alternate allele superior in terms of survival**. In an environment where malaria is not prevalent the frequency of sickle cell allele would be very low. It is also true that the selection process does not totally eliminate an undesirable allele from the population since other evolutionary forces such as mutations continue to generate the allele. This experiment illustrates the role of natural selection in promoting or reducing the frequency of alleles that are better adapted or ill adapted respectively to a given environment.

Objectives

At the end of this exercise you should be able to:

- discuss that natural selection does not totally promote (100%) any allele except when it is adapted to the environment, and
- describe that the alleles for maladaptations continue to occur at very low frequencies in the population.

12.2 MATERIAL REQUIRED

Plastic beads (bigger sized ones) of red, black, blue and green colour-500 of each colour.

A plastic bowl.

A white cloth towel with rough texture.

12.3 PROCEDURE

1. Place the 2000 beads – 500each of blue, red, black and green – in a plastic bowl and mix thoroughly.
2. At random, pick out 100 beads from a total of 2000 in the bowl without looking into it and place them on the white towel spread on the table. The purpose of using the white towel is that coloured beads will be seen clearly on a white background and the beads will not roll off. You may pick more than 100 but picking out 100 would make percentage calculations much easier.
3. In the next step, separate 100 beads according to the colour on the towel itself. Since you have picked 100 beads randomly from a population of 2000 beads, you may have picked close to 25 numbers of each colour. Assume that the number of heads you have picked is as follows:

Green	26
Red	24
Blue	26
Black	24

Record these numbers in your record note book.

4. Now let us say that natural selection is operating. Assume that the selection favours green beads and opposes black beads in the population. To represent this concept, add 10 more green beads to the 100 you have picked out and remove 10 black beads. The new frequencies will be as follows:

Green	36
Red	24
Blue	26
Black	14
	100

Record these numbers in your note book.

5. With these frequencies let us make a new population of 1000 individuals. This means you will mix up 360 green beads, 240 red ones, 260 blue ones and 140 black ones, to make a total of 1000. These 1000 individuals belong to the 2nd generation. From this population of 1000 beads, pick out a 100 beads.

Suppose the sample of 100 individuals has the following distribution:

Green	34
Red	27
Blue	23
Black	16
	100

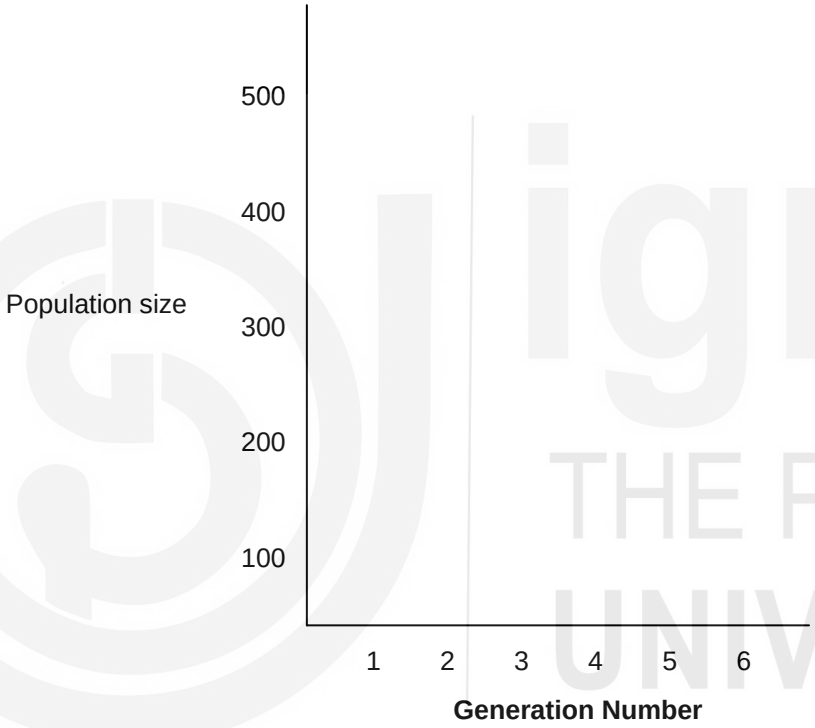
Record the numbers in your observation note book. Because of the selection process, the frequencies of beads have changed. There is an increase in the numbers of favoured beads (characters or alleles) and a decrease in the number of not so favoured ones. Let the selection continue to act. Add 10 more of green beads and remove ten of the black beads. The new population that constitutes the third generation will be as follows:

Green	44
Red	27
Blue	23
Black	06
	100

Record these results in your note book. You may continue to do the experiment for a number of generations. But confine yourself to six or seven generations and record the results in the following table.

Generation	No. of greens	No. of reds	No. of blues	No. of blacks
I				
II				
III				
IV				
V				
VI				

Plot the results in a graphical form



12.4 INFERENCE DRAWN

You may recall that one definition of evolution, the definition proposed by population geneticists is that **evolution is a systematic change in gene frequencies**. You may say that such changes in gene frequencies are a measure of evolution. You may also observe that after a few generations, the frequencies of various traits (colours in this exercise) more or less remain the same. The green colour after a few generations would end up with a frequency ranging from 40 to 60%. The red and blue with twenties or less; and the black registering less than 5%. At the same time the black may not be eliminated totally from the population. In other words, at the end of the six or so generations, all the traits (colours), the 'fittest' green, the 'fitter' red and blue and the 'least fit' black continue to exist in the population. Natural selection may not completely fix (100% presence) or eliminate (0%) any allele in the

population. When environmental conditions are favourable, natural selection promotes the frequency of favoured alleles but does not eliminate totally the least adapted ones from the population. This is how the role of Darwinian selection is perceived in recent times.

12.5 TERMINAL QUESTIONS

1. In what ways evolution is influenced by natural selection?
2. Does natural selection eliminate all alleles for maladaptations? Explain.

EXERCISE 13

AN EXERCISE TO ILLUSTRATE THE CONCEPT OF GENETIC DRIFT

Structure

13.1 INTRODUCTION

In randomly mating large populations, according to Hardy-Weinberg principle the frequency of any given pair of alleles tend to remain constant in the absence of selection and mutations. If selection and mutations do occur then the frequencies do change and such changes are usually small and slow. We have discussed in the theory course about the behaviour of genes in **small populations,** and the concept of **genetic drift** was illustrated with a small population of mice living as four or five extended families in the rice barn of a farmer. In small populations due to sampling error there will be random changes in the frequencies of genes in the population. Such changes or drift in gene frequencies may eliminate well adapted traits from the population and fix the less adapted ones.

The two forms of genetic drift are the **bottleneck effect** and the **founder effect**.

We mentioned in the previous exercise that under the influence of natural selection neither the fixation (100%) nor elimination (0%) of a trait occurs. Under the influence of the genetic drift, a trait may become fixed (100%) or even eliminated (0%) from the population. In this exercise we shall illustrate the concept of genetic drift with a simple exercise.

Objectives

After the end of this exercise you should be able to:

- distinguish the specific role of natural selection and genetic drift in the evolutionary process, and
- illustrate the concept of genetic drift.

13.2 MATERIAL REQUIRED

Plastic beads (big sized ones) of green, red, blue and black colours–500 each, two plastic bowls.

13.3 PROCEDURE

As in the previous exercise, let us assume the green colour is favoured by selection and black colour is opposed by it.

1. From a population of 2000 beads (500 of each colour) kept well mixed in a plastic bowl, just pick out 10 beads with your finger tips. This should be done at random without looking into the bowl. Let us say that the 10 beads belong to the four colours in the following way:

Green	4
Red	1
Blue	3
Black	2

2. Record these results in your note book. Now, with the above frequencies make a population of 100 heads.

   This distribution will be

Green	40
Red	10
Blue	30
Black	20
	100

3. Out of these 100 beads, once again pick up ten beads with your finger tips. Suppose you get the following.

Green	0
Red	2
Blue	3
Black	5
	10

Record your results.

4. Now there are just three colours left. Once again make a total of 100 heads based on new frequencies. This would mean no greens, 20 reds, 30 blues and 50 blacks to make a hundred. Out of these hundred pick up 10 beads. Suppose, you get the following distribution.

Red	0
Blue	4
Black	6

Record your result.

5. Now with the new frequencies, make up a population with 40 blues and 60 blacks. Pick out ten beads out of these hundred and you may get the following figures.

Blue	3
Black	7

Record your results.

6. Repeat the experiment after they were made upto 100 beads with 30 blues and 70 blacks. Supposing we get the following:

Blue	0
Black	10

Record you results.

7. Put you results on graph sheet.

13.4 INFERENCE DRAWN

You may observe here that the green colour favoured by the selection process is eliminated from the population by the third generation. On the contrary the black colour which is normally opposed by the selection gets fixed in the population by about sixty generation.

Both natural selection and genetic drift are mechanisms for evolution (they both change allele frequencies overtime). The key distinction that in **genetic drift** allele frequencies change by chance whereas **natural selection** allele frequencies change by differential reproductive success. The effect of genetic drift is strongest in small populations as chance events will significantly change the frequencies of alleles in small population.

1. Why this happened?
2. Do you think that effects of genetic drift are opposite to that of selection? Why this is so?
3. Can genetic drift be an important agent during the process of evolution?

13.5 TERMINAL QUESTIONS

1. Do you think that the effects of genetic drift are quite opposite to the ones produced by natural selection. Why?
2. Can genetic drift be a significant factor in the evolutionary process? Discuss.

SUGGESTED READINGS

Exercises 1 to 6

Recommended Books

- Snustad, D.P., Simmons, M.J. (2009). *Principles of Genetics*. V Edition. John Wiley and Sons In.
- Klug, W.S., Cummings, M.R., Spencer, C.A. (2012). *Concepts of Genetics*. X Edition. Benjamin Coming.
- Pierce B. A. (2012), *Genetics-A Conceptual Approach*. IV Edition. W. H. Freeman and Company.

Suggested Readings

- Russell, P.J. (2009), *Genetics-A Molecular Approach*. III Edition. Benjamin Cummings.
- Griffiths, A.J.F., Wessler, S.R., Lewontin, R.C. and Carroll, S.B. *Introduction to Genetic Analysis*. IX Edition. W. H. Freeman and Co.
- Gardner, E.J., Simmons, M.J., Snustad, D.P. (2008). *Principles of Genetics.* VIII Edition. Wiley India.

Online Tools and Web Resources

- https://swayam.gov.in/courses/4922-genetics-and-genomics
- https://swayam.gov.in/course/96-genetics
- https://www.coursera.org/learn/genetics-evolution
- https:/onlinelearning.hms.harvard.edu/hmx/courses/hmx-genetics/
- https://learn.genetics.utah.edu/

Exercises 7 to 13

Recommended Books

- Ridley, M. (2004). Evolutin.III Edition, Blackwell publishing.
- Hall, B.K. and Hallgrimson, B. (2013). Evolution. V Edition. Jones and Barlett Publishers.
- Douglas, J. Futuyma (1997). Evolutionary Biology. Sinauer Associates.

Suggested Readings

- Pevsner, J. (2009). Bioinformatics and Functional Genomics. II Edition, Wiley-Blackwell.
- Campbell, N.A. and Reece J.B. (2011). Biology. IX Edition. Pearson, Benjamin, Cummings.

Online Tools and Web Resources

- https://www.coursera.org/learn/molecular-evolution
- https://www.coursera.org/learn/genetics-evolution
- https://swayam.gov.in/courses/4062-environmental-biology-genetics-and-evolution