A practice book for Introductory Genetics

Student's guide

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Contents

Practical 1: Study of features of mitosis

Objectives

1. To study about different stages of mitotic cell division

Theory

- **Mitosis** is the division of the genetic material in the nucleus, usually followed immediately by **cytokinesis**.
- Walther Flemming coined the terms *mitosis* and *chromatin*.
- Mitosis and cytokinesis produce cells that make up most of our bodies and same process continues to generate new cells to replace dead and damaged ones.
- Mitotic (M) phase, including both mitosis and cytokinesis, is the shortest part of a cell's life cycle.
- M phase alternates with **interphase** (accounts for 90% of the cycle).
- Interphase can be divided into subphases (with time lapse for a normal human cell, about **24 hours**):
  - the $\mathbf{G_1}$ phase ("first gap") $\longrightarrow$ 5-6 hours,
  - the S phase ("synthesis") $\longrightarrow$ 10-12 hours, and
  - the $\mathbf{G_2}$ phase ("second gap") $\longrightarrow$ 4-6 hours.
- During all three subphases, a cell that will eventually divide grows by producing proteins and cytoplasmic organelles such as mitochondria and endoplasmic reticulum.
- Chromosomes are duplicated only during the S phase.

Mitosis usually takes up only a small proportion of the cell cycle, approximately 5-10 percent. The remaining time in the interphase, composed of G1, S, and G2 stages. The DNA is replicated during the S phase, although the duplicated DNA does not become visible until later in mitosis. The chromosomes cannot be seen during iterphase. Mainly because they are in an extended state and are intertwined with one another like a tangle of yarn.

A type of nuclear division (occurring at cell division) that produce two daughter nuclei identical with that of the parent nucleus is known as mitosis. The cell undergoing mitotic division goes through two distinct phases namely – interphase and mitotic phase.

Interphase comprises of following events:

1. Structural and functional protein are synthesized
2. RNA and ribosomes are synthesized
3. DNA of each chromosome replicates and each chromosome becomes two sister chromatids which lay side by side
4. Cell volume increases

The G phases were misnamed as "gaps" when they were first observed because the cells appeared inactive, but we now know that intense metabolic activity and growth occur throughout interphase. During all three phases of interphase, in fact, a cell grows by producing proteins and cytoplasmic organelles such as mitochondria and endoplasmic reticulum. Duplication of the chromosomes, crucial for eventual division of the cell, occurs entirely during the S phase. Thus, a cell grows (G1), continues to grow as it copies its chromosomes (S), grows more as it completes preparations for cell division (G2), and divides (M). The daughter cells may then repeat the cycle.

A particular human cell might undergo one division in 24 hours. Of this time, the M phase would occupy less than 1 hour, while the S phase might occupy 10–12 hours, or about half the cycle. The rest of the time would be apportioned between the G1 and G2 phases. The G2 phase usually takes 4–6 hours; in our example, G1 would occupy about 5–6 hours. G1 is the most variable in length in different types of cells. Some cells in a multicellular organism divide very infrequently or not at all.

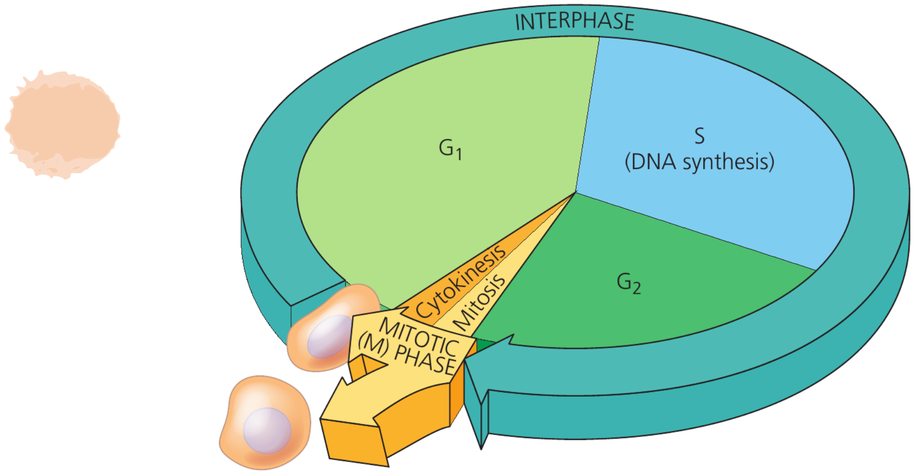
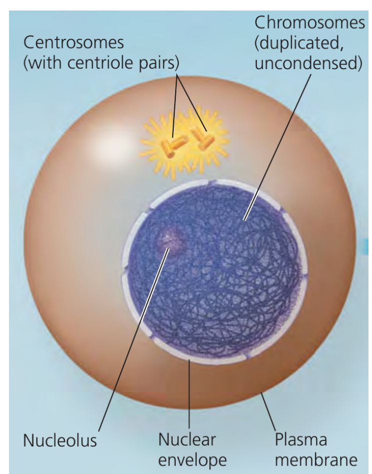


Figure 1.1: In a dividing cell, the mitotic (M) phase alternates with interphase, a growth period. The first part of interphase (G1) is followed by the S phase, when the chromosomes duplicate; G2 is the last part of interphase. In the M phase, mitosis distributes the daughter chromosomes to daughter nuclei, and cytokinesis divides the cytoplasm, producing two daughter cells.

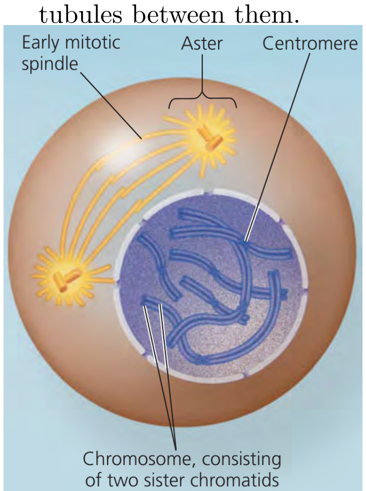


G2 of Interphase

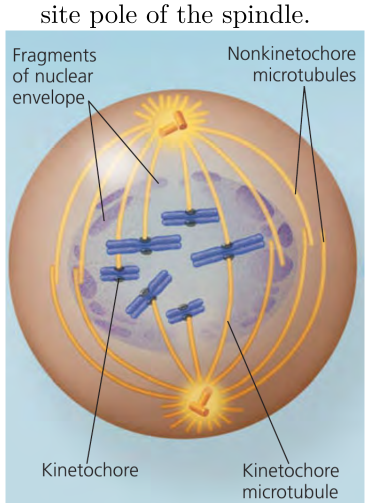
- A nuclear envelope encloses the nucleus
- The nucleus contains one or more nucleoli (singular, nucleolus)
- Two centrosomes have formed by duplication of a single centrosome. Centrosomes are regions in cell that organize the microtubules of the spindle. Each centrosome contains two centrioles.
- Chromosomes, duplicated during S phase, cannot be seen individually because they have not yet condensed.

Following events describe mitotic phase:

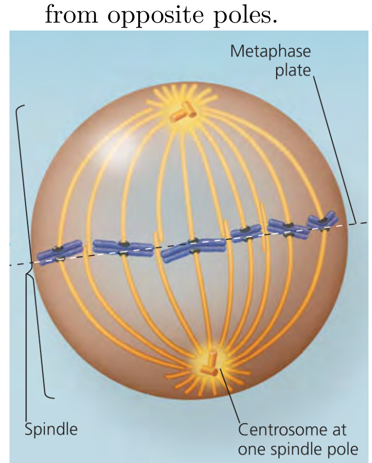
1. Prophase
   - The chromatin fibers become more tighly coiled, condensing into discrete chromosomes observable with a light microscope.
   - The nucleoli disappear.
   - Each duplicated chromosomes appear as two identical sister chromatids joined at their centromeres and, in some species, all along their arm by cohesins (sister chromatid cohesion)
   - The mitotic spindle (named after its shape) begins to form. It is composed of the centrosomes and the microtubules that extend from them. The radial arrays of shorter microtubules that extend from the centrosomes are called asters ("stars").
   - The centrosomes move away from each other, propelled partly by the lengthing microtubules between them.



2. Early metaphase
   - The nuclear envelope fragments.
   - The microtubules extending from each centrosome can now invade the nuclear area.
   - The chromosomes have become even more condensed.
   - Each of the two chromatids of each chromosome now has a kinetochore, a specialized protein structure at the centromere.
   - Some of the microtubules attach to the kinetochores, becoming "kinetochore microtubules", which jerk the chromosomes back and forth.
   - Nonkinetochore microtubules interact with those from opposite pole of the spindle.

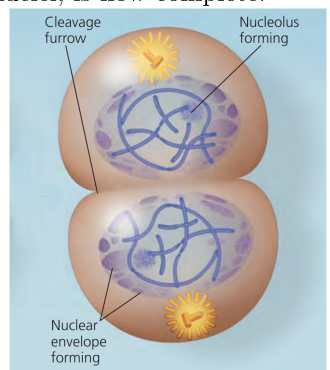


3. Late metaphase
   - The centrosomes are now at opposite poles of the cell.
   - The chromosomes convene at the metaphase plate, a plane that is equidistant between the spindle's two poles. The chromosomes' centromeres lie at the metaphase plate.
   - For each chromosome, the kinetochores of the sister chromatids are attached to kinetochore microtubules coming from opposite poles.



4. Anaphase
   - Anaphase is the shortest stage of mitosis, often lasting only a few minutes.
   - Anaphase begins when the cohesin proteins are cleaved. This allows the two sister chromatids of each pair to part suddenly. Each chromatid thus becomes full-fledged chromosome.
   - The two liberated daughter chromosomes begin moving toward opposite ends of the cell as their kinetochore microtubules shorten. Because these microtubules are attached at the centromere region, the chromosomes move centromere first (at about 1 micron/min).
   - The cell elongates as the nonkinetochore microtubules lengthen.
   - By the end of anaphase, the two ends of the cell have equivalent – and complete – collections of chromosomes.

5. Telophase
   - Two daughter nuclei form in the cell. Nuclear envelopes arises from the fragments of the parent cell's nuclear envelope and other portions of the endomembrane system.
   - Nucleoli reappear.
   - The chromosomes become less condensed.
   - Any remaining spindle microtubules are depolymerized.
   - Mitosis, the division of one nucleus into two genetically identical nuclei, is now complete.



Beside these four phases, cytokinesis (a part of cell cycle) occur after completion of telophase. In cytokinesis, following events are noticeable:

- The division of the cytoplasm is usually well under way by late telophase, so the two daughter cells appear shortly after the end of mitosis.
- In animal cells, cytokinesis involves the formation of a cleavage furrow, which pinches the cell in two.

Conclusion

Hence, after study of mitotic diagrams and comparing and contrasting among several stages of a cell cycle, we understand that mitotic division of cell results into equal and identical allocation of genetic material from a parent cell to daughter cells.

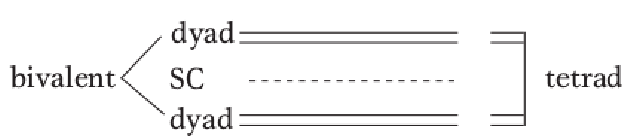
Practical 2: Study of features of meiosis

Objectives

1. To study about different stages of meiotic cell division

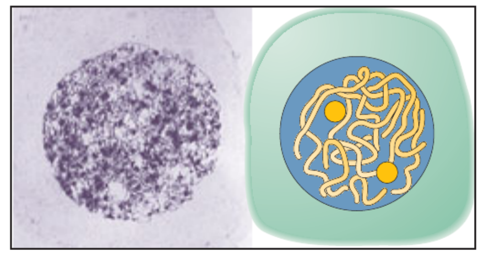
Theory

- **Meiosis** occurs in humans only in gonads (ovaries or testes) to produce eggs or sperm.
- In each division meiosis reduces the chromosome number from 46 (two sets of chromosomes) to 23 (one set), hence called the reductional division.
- Fertilization fuses gametes together and returns chromosome number to 46.
- Before meiosis, chromosome replication takes place to form sister chromatids, which become visible at meiosis.
- Contrasting mitosis and meiosis:
  - Centromere appears not to divide at this stage, whereas it does in mitosis.
  - The homologous pairs of sister chromatids now unite to form a bundle of four homologous chromatids, they do not in mitosis.
- Joining of the homologous pairs is called **synapsis**, and it relies on the properties of a macromolecular assemblage called the synaptonemal complex (SC), which runs down the center of the pair.
- Replicate sister chromosomes are together called a **dyad** (from the Greek word for two). The unit comprising the pair of synapsed dyads is called a **bivalent**.
- The four chromatids that make up a bivalent are called a **tetrad** (Greek for four), to indicate that there are four homologous units in the bundle.

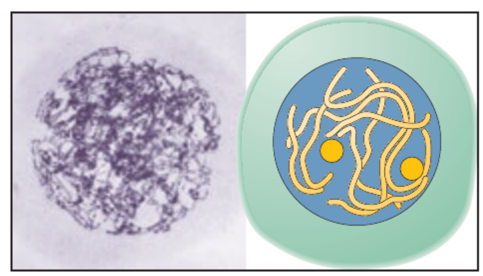


- The bivalents of all chromosomes move to the cell's equator, and, when the cell divides, one dyad moves into each new cell, pulled by spindle fibers attached to the centromeres.
- In the second cell division of meiosis, the centromeres divide and each member of a dyad (each member of a pair of chromatids) moves into a daughter cell. Hence, although the process starts with the same genomic content as that for mitosis, the two successive segregations result in four haploid cells.
- Each of the four haploid cells that constitute the four products of meiosis contains one member of a tetrad; hence, the group of four cells is sometimes called a tetrad, too.

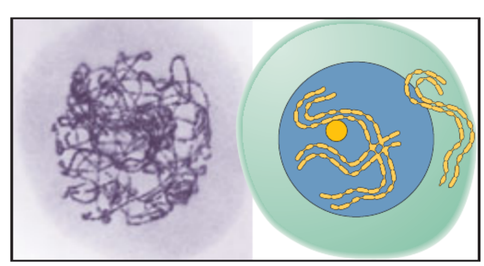
Stages of meiosis



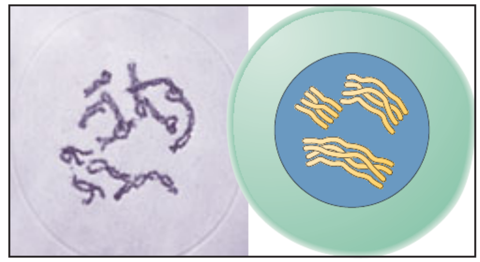
Leptotene
The chromosomes become visible as long, thin single threads. Chromosomes begin to contract and continue contracting throughout the entire prophase.

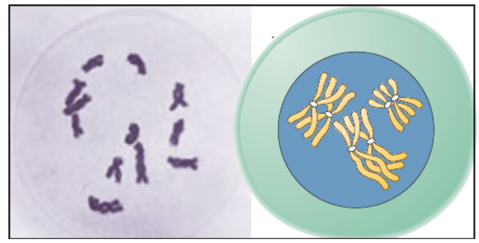


Zygotene
The threads form pairs as each chromosome progressively aligns, or synapses, along the length of its homologous partner.

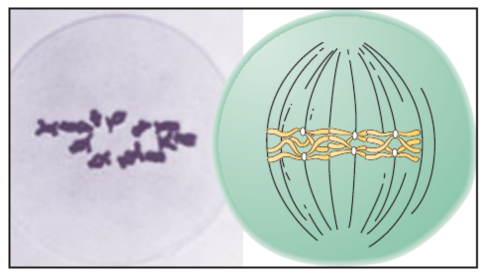


Pachytene
Chromosomes are thick and fully synapsed. Thus, the number of pairs of homologous chromosomes is equal to the number n.





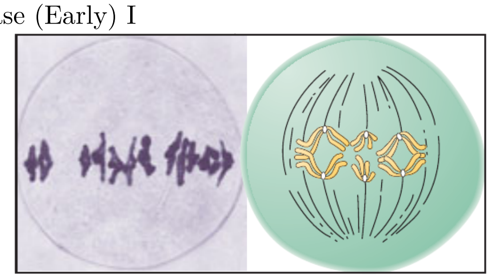
Diplotene
Although the DNA has already replicated during the premeiotic S phase, this fact first becomes manifest only in diplotene as each chromosome is seen to have become a pair of sister chromatids. The synapsed structure now consists of a bundle of four homologous chromosomes. The paired homologs separate slightly, and one or more cross-shaped structures called chiasmata (singular, chiasma) appear between nonsister chromatids.
Diakinesis
Further chromosome contraction produces compact units that are very maneuverable.



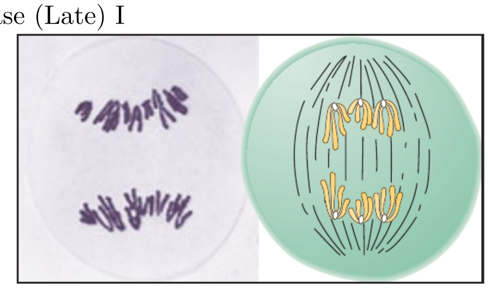
Metaphase I
The nuclear membrane has disappeared, and each pair of homologs takes up a position in the equatorial plane. At this stage of meiosis, the centromeres do not divide; this lack of division is a major difference from mitosis. The two centromeres of a homologous chromosome pair attach to spindle fibers from opposite poles.

Anaphase I
The members of each homologous pair move to opposite poles.

Anaphase (Early) I

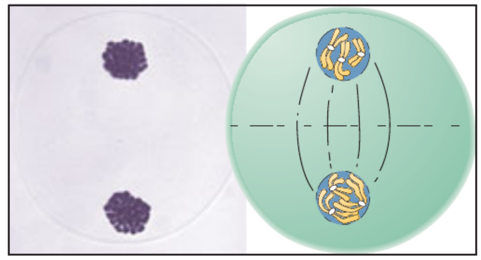


Anaphase (Late) I

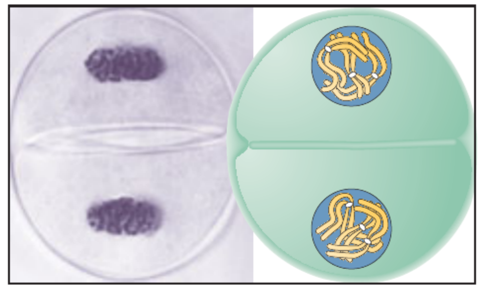


Telophase I and Interphase
The chromosomes elongate and become diffuse, the nuclear membrane re-forms and the cell divides. After telophase I, there is an interphase, called interkinesis. In many organisms, telophase I and interkinesis do not exist or are brief in duration. In any case, there is never DNA synthesis at this time, and the genetic state of the chromosomes does not change.

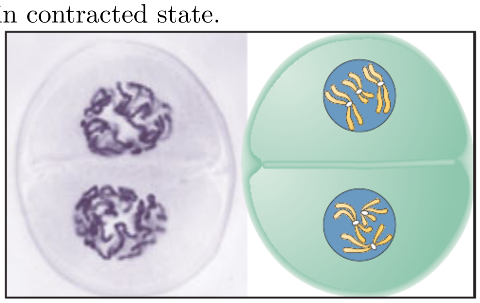
Telophase



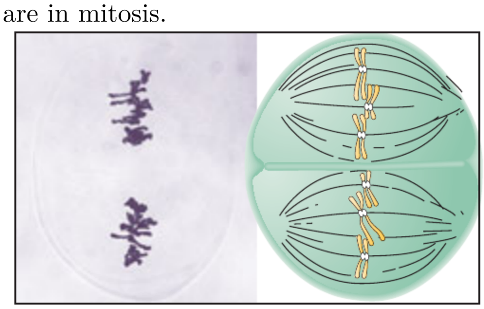
Interphase II



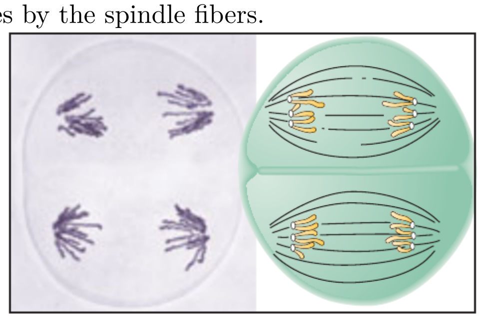
Prophase II
The haploid number of sister chromatid pairs are now present in contracted state.



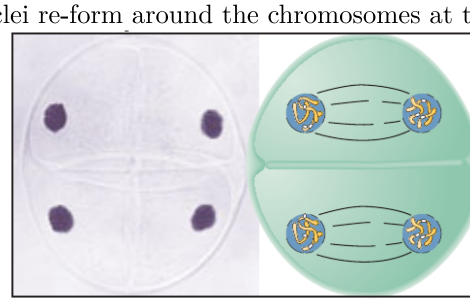
Metaphase II
The pair of sister chromatids arrange themselves on the equatorial plane. Here the chromatids often partly dissociate from each other instead of being closely pressed together as they are in mitosis.



Anaphase II
Centromeres split and sister chromatids are pulled to opposite poles by the spindle fibers.

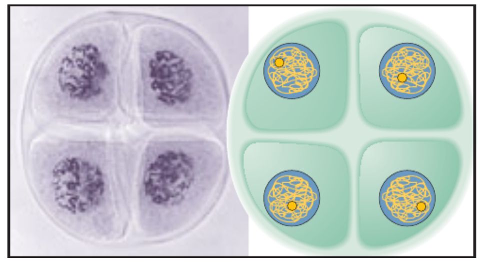


Telophase II
The nuclei re-form around the chromosomes at the poles.

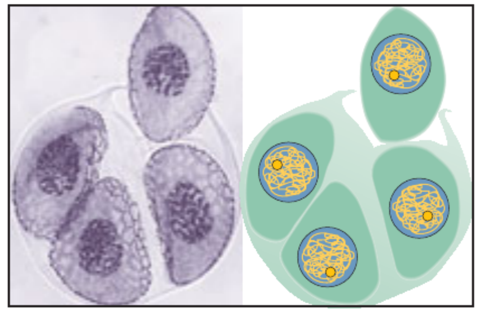


The tetrad and young pollen grains
In the anthers of a flower, the four products of meiosis develop into pollen grains. In other organisms, the products of meiosis differentiate into other kinds of structures, such as sperm cells in animals.

Tetrad



Young pollen grain



Conclusion

Hence, after study of meiotic diagrams and comparing and contrasting among several stages of a cell cycle, we understand that meiotic division of cell results into haploid/gametic chromosome number containing daughter cells.

Practical 3: Solution of numerical problems related to Mendel's law of segregation

Question 1

Phenylketonuria (PKU) is a human hereditary disease resulting from the inability of the body to process the chemical phenylalanine, which is contained in the protein that we eat. PKU is manifested in early infancy and, if it remains untreated, generally leads to mental retardation. PKU is caused by a recessive allele with simple Mendelian inheritance.

A couple intends to have children but consult a genetic counselor because the man has a sister with PKU and the woman has a brother with PKU. There are no other known cases in their families. They ask the genetic counselor to determine the probability that their first child will have PKU. What is this probability?

Question 2

While performing a large scale grow out experiment of pea germplasm, a plant with three cotyledons was discovered (normally, there are 2 cotyledons). This plant was crossed with a normal pure-breeding wild-type plant, and 600 seeds from this cross were planted. There were 298 plants with two cotyledons and 302 with three cotyledons. What can be said about the inheritance of three cotyledons ?

Question 3

On the basis of Mendel's hypothesis and observations predict the results from following crosses in garden peas.

a. A tall (dominant and homozygous) variety crossed with a dwarf variety
b. The progeny of (a) selfed
c. The progeny from (a) crossed with the original tall parent.
d. The progeny from (a) crossed with the original dwarf variety.

Question 4

Two black pigs were mated and over several years produced 29 black and 9 white offerings. Explain the results giving the genotypes of parents and progeny.

Practical 4: Solution of numerical problems related to Mendel's law of independent assortment

Question 1

In tomatoes, one gene determines whether the plant has purple (P) or green (G) stems, and a separate, independent gene determines whether the leaves are "cut" (C) or "potato" (Po). Five matings of tomato-plant phenotypes give the following results.

1. Which alleles are dominant ?
2. What are the most probable genotypes for the parents in each cross ?

Mating	Parental phenotypes	*Number of progeny* P, C	P, Po	G, C	G, Po
1	P, C × G, C	323	102	309	106
2	P, C × P, Po	220	206	65	72
3	P, C × G, C	723	229	0	0
4	P, C × G, Po	405	0	389	0
5	P, Po × G, C	71	90	85	78

Question 2

(Effective crossing) A corn geneticist has three pure lines of genotypes a/a; B/B; C/C, A/A; b/b; C/C, and A/A; B/B; c/c. All the phenotypes determined by a, b and c will increase the market value of the corn; so, naturally, he wants to combine them all in one pure line of genotype a/a; b/b; c/c.

1. Outline an effective crossing program that can be used to obtain the a/a; b/b; c/c pure line.
2. At each stage, state exactly which phenotypes will be selected and give their expected frequencies.
3. Is there more than one way to obtain the desired genotype ? Which is the best way ? (Assume independent assortment of the three gene pairs.)

Question 3

How many different types of F_1 gametes, F_2 genotypes and F_2 phenotypes would be expected from:

(a) AA x aa, AABB x aabb, AABBCC x aabbcc
(b) What general formula can be applied for F_1 gametes, F_2 genotypes and F_2 phenotypes ?

Question 4

In tomato red fruit flower is dominant to yellow, two loculed fruit is dominant to many loculed and tall vine is dominant to dwarf. A breeder has two pure lines; red two loculed dwarf and yellow many loculed tall. From these two lines, he wants to produce new pure line of yellow, two loculed and tall. How exactly should he go about doing this ? Show not only which crosses to make, but also how many progeny should be sampled in each case?

Question 5

In dogs, dark coat color is dominant over albino, and short hair is dominant over the long hair. If these effects are caused by two independently segregating gene pairs, write the most preferable genotypes for the parents of each of the following crosses.

Parental phenotypes	Dark short	Dark long	Albino short	Albino long
Dark short x Dark short	89	31	29	11
Dark short x Dark long	19	19	0	0
Dark short x Albino short	20	0	21	0
Albino short x Albino short	0	0	28	9
Dark long x Dark long	0	32	0	10
Dark short x Dark long	29	31	9	11

Practical 5: Study of gene action and interaction

Objective

- To understand how alleles of a gene interact to give rise to phenotypes in diploid organism
- To understand how non-allelic genes interact to affect trait phenotypes in diploid organism.

Theory

In Mendelian phenotypic ratio, one gene is involved in expression of a single phenotype. However, some phenotype develop through gene interaction between two or more genes. i.e. two or more genes may also be involved in determination of a single trait and the phenotypic ratio deviates from the expected Mendelian phenotypic ratio.

- There are a variety of phenotypic ratios which are the result of a variety of gene interaction. Steps in inferring gene interaction :
  1. Obtain many single-gene mutants and test for dominance
  2. Test the mutants for allelism
  3. Combine the mutants in pairs to form double mutants to see if the genes interact.

<u>Types of gene interaction</u>

- No interaction (Each gene pair affects a different character; gene action) (9:3:3:1)
- Duplicate gene action (15:1)
- Complementary gene action (9:7)
- Supplementary gene action (9:3:4)
- Inhibitory gene action (13:3)
- Masking gene action (12:3:1)
- Polymeric gene action (9:6:1)
- Additive gene action (1:4:6:4:1)

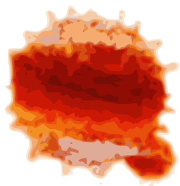
Inter allelic or intra-genic gene interaction

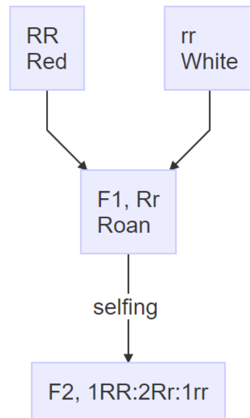
1. Incomplete or partial dominance

The phenotypic expression of heterozygous for a gene being intermediate to those of the two concerned homozygous. e.g., four o' clock plant (*Mirabilis jalapa*), we can cross a plant of red flower petals with another plant with white petals. The offspring of this cross will have pink colored petals. If these pink flowered F1 plants are selfed and F2 are obtained the progeny phenotypes of red:pink:white occur in the ratio 1:2:1.

2. Codominance

Both the alleles of a gene express themselves in the heterozygous. e.g. coat color in cattle, blood type in human coat color in cattle;





3. Dominance
4. Overdominance

Table 5.1: Each gene pair affecting a different character

Type of gene/s	Phenomena	Individuals	Phenotypes	Genotypes
Each gene pair affecting a different character	Complete dominance at both gene pairs	$\frac{9}{16}$	yellow round	1, 2, 3, 4, 5, 7, 9, 10, 13
	Example: Mendel's peas	$\frac{3}{16}$	yellow wrinkled	6, 8, 14
	Gene pair A: (seed color) yellow dominant over green	$\frac{3}{16}$	green round	11, 12, 15
	Gene pair B: (seed shape) round dominant over wrinkled	$\frac{1}{16}$	green wrinkled	16

Gamete	AB	Ab	aB	ab
AB	ABAE	AbAB	aBAB	abAB
Ab	ABAb	AbAb	aBAb	abAb
aB	ABaB	AbaB	aBaB	abaB
ab	ABab	Abab	aBab	abab

No interaction

- Two dominant genes controlling the development of a single trait.

Table 5.3: Each gene pair affecting the same character

Type of gene/s	Phenomena	Individuals	Phenotypes	Genotypes
Each gene pair affecting the same character	Complete dominance at both gene pairs; new phenotypes resulting from interaction between dominants, and also from interaction between both homozygous recessives	$\frac{9}{16}$	walnut	1, 2, 3, 4, 5, 7, 9, 10, 13
	Example: comb shape in poultry	$\frac{3}{16}$	rose	6, 8, 14

	Gene pair A: rose comb dominant over nonrose Gene pair B: pea comb dominant over nonpea Interaction: Dominants for rose and pea produce walnut comb. Homozygous recessives for rose and pea produce single comb	$\frac{3}{16}$ $\frac{1}{16}$	pea single	11, 12, 15 16

Gamete	AB	Ab	aB	ab
AB	ABAB	AbAB	aBAB	abAB
Ab	ABAb	AbAb	aBAb	abAb
aB	ABaB	AbaB	aBaB	abaB
ab	ABab	Abab	aBab	abab

Supplementary gene action

- The dominant allele of one gene produces a phenotypic effect.
- The dominant allele of the other gene does not produce any phenotypic effect on its own, but when present with dominant allele of the first gene, it modifies the phenotypic effect produced by the first gene.
- For e.g. development of grain (aleurone) color in maize producing 9:3:4 ratio in F_2.

Table 5.5: Each gene pair affecting the same character

Type of gene/s	Phenomena	Individuals	Phenotypes	Genotypes
Each gene pair affecting the same character	Complete dominance at both gene pairs, but one gene, when homozygous recessive, is epistatic to the other	$\frac{9}{16}$	agouti	1, 2, 3, 4, 5, 7, 9, 10, 13
	Example: mouse coat color Gene pair A: color dominant over albino Gene pair B: agouti color dominant over black Interaction: homozygous albino is epistatic to agouti and black	$\frac{3}{16}$ $\frac{4}{16}$	black albino	6, 8, 14 11, 12, 15, 16

Gamete	AB	Ab	aB	ab
AB	ABAB	AbAB	aBAB	abAB
Ab	ABAb	AbAb	aBAb	abAb
aB	ABaB	AbaB	aBaB	abaB
ab	ABab	Abab	aBab	abab

Complementary gene action

- Production of one phenotype requires the presence of dominant alleles of both genes controlling the character.
- When any one of the two or both the genes are present in homozygous recessive state, the contrasting phenotype is produced.

Table 5.7: Each gene pair affecting the same character

Type of gene/s	Phenomena	Individuals	Phenotypes	Genotypes
Each gene pair affecting the same character	Complete dominance at both gene pairs, but either recessive homozygote is epistatic to the effects of the other gene	$\frac{9}{16}$	purple	1, 2, 3, 4, 5, 7, 9, 10, 13
	Example: sweet pea flower color Gene pair A: purple dominant over white Gene pair B: color dominant to colorless (white) Interaction: homozygous recessives at either gene A or B produce white	$\frac{7}{16}$	white	6, 8, 11, 12, 14, 15, 16

Gamete	AB	Ab	aB	ab
AB	ABAB	AbAB	aBAB	abAB
Ab	ABAb	AbAb	aBAb	abAb
aB	ABaB	AbaB	aBaB	abaB
ab	ABab	Abab	aBab	abab

Masking gene action

- Dominant alleles of the two genes affecting a character produce distinct phenotypes when they are alone. But when dominant alleles of both the genes are present together, the expression of dominant allele of one gene masks the expression of the other. When both the genes are present in recessive state, a different phenotype is produced.

Table 5.9: Each gene pair affecting the same character

Type of gene/s	Phenomena	Individuals	Phenotypes	Genotypes
Each gene pair affecting the same character	Complete dominance at both gene pairs, but one gene, when dominant, epistatic to the other	$\frac{12}{16}$	white	1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 13, 14
	Example: fruit color in summer squash Gene pair A: white dominant to color Gene pair B: yellow dominant to green Interaction: Dominant white hides the effect of yellow or green	$\frac{3}{16}$ $\frac{1}{16}$	yellow green	11, 12, 15 16

Gamete	AB	Ab	aB	ab
AB	ABAB	AbAB	aBAB	abAB
Ab	ABAb	AbAb	aBAb	abAb
aB	ABaB	AbaB	aBaB	abaB
ab	ABab	Abab	aBab	abab

Inhibitory gene action

- One dominant gene produces the concerned phenotype or the character, while its recessive allele produces the contrasting phenotype.
- The second gene has no effect of its own on the character in question, but it stops the expression of the dominant allele of the first gene.
- As a result, when two dominant genes are present togethe, they produce the same phenotype as that produced by the recessive homozygote of the first gene.
- The recessive allele of the second gene does not affect the development of the character in any way.
- For e.g. genes controlling seed color in maize.

Table 5.11: Each gene pair affecting the same character

Type of gene/s	Phenomena	Individuals	Phenotypes	Genotypes
Each gene pair affecting the same character	Complete dominance at both gene pairs, but one gene, when dominant, epistatic to the second, and the second gene, when homozygous recessive, epistatic to the first	$\frac{13}{16}$	white	1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 13, 14, 16
	Example: feather color in fowl Gene pair A: color inhibition is dominant to color appearance Gene pair B: color is dominant to white Interaction: dominant color inhibition prevents color even when color is present: color gene, when homozygous recessive, prevents color even when dominant inhibitor is absent	$\frac{3}{16}$	color	11, 12, 15

Gamete	AB	Ab	aB	ab
AB	ABAE	AbAB	aBAB	abAB
Ab	ABAb	AbAb	aBAb	abAb
aB	ABaB	AbaB	aBaB	abaB
ab	ABab	Abab	aBab	abab

Polymeric gene action

- Two completely dominant genes controlling a character produce the same phenotype when their dominant alleles are alone. But when dominant alleles of both genes are present together, their phenotypic effect is enhanced as if the effect of the two genes are present together, their phenotypic effect is enhanced as if the effect of the two genes were cumulative or additive.

Table 5.13: Each gene pair affecting the same character

Type of gene/s	Phenomena	Individuals	Phenotypes	Genotypes
Each gene pair affecting the same character	Complete dominance at both gene pairs; interaction between both dominants to give new phenotypes	$\frac{9}{16}$	disc	1, 2, 3, 4, 5, 7, 9, 10, 13
	Example: fruit shape in summer squash	$\frac{6}{16}$	sphere	6, 8, 11, 12, 14, 15
	Gene pair A: sphere shape dominant over long shape	$\frac{1}{16}$	long	16
	Gene pair B: sphere shape dominant over long shape			
	Interaction: dominants at A and B, when present together, form disc-shaped fruit			

Gamete	AB	Ab	aB	ab
AB	ABAE	AbAB	aBAB	abAB
Ab	ABAb	AbAb	aBAb	abAb
aB	ABaB	AbaB	aBaB	abaB
ab	ABab	Abab	aBab	abab

Additive gene action

- Each positive allele of the two genes governing a trait produces equal and identical effect on the character.
- This gene action is the basis for **multiple factor hypothesis** and the gene action sometimes aka polygenic action.

Table 5.15: Each gene pair affecting the same character

Type of gene/s	Phenomena	Individuals	Phenotypes	Genotypes
Each gene pair affecting the same character	Partial dominance at both gene pairs; additive effects for each partially dominant gene	$\frac{1}{16}$	purple shade 10	1
	Example: flower color in beans (Mendel)	$\frac{2}{16}$	purple shade 8	2, 5
	Gene pair A: purple flower color partially dominant to white; additive effect on color for each A gene (i.e., value of 3)	$\frac{2}{16}$	purple shade 7	3, 9
	Gene pair B: purple flower color partially dominant to white; additive effect on color for each A gene (i.e., value of 2)	$\frac{1}{16}$	purple shade 6	6
		$\frac{4}{16}$	purple shade 5	4, 7, 10, 13
		$\frac{1}{16}$	purple shade 4	11
		$\frac{2}{16}$	purple shade 3	8, 14
		$\frac{2}{16}$	purple shade 2	12, 15

$\frac{1}{16}$	purple shade 0 (white)	16

Gamete	AB	Ab	aB	ab
AB	ABAB	AbAB	aBAB	abAB
Ab	ABAb	AbAb	aBAb	abAb
aB	ABaB	AbaB	aBaB	abaB
ab	ABab	Abab	aBab	abab

Conclusion

Thus by studying case scenarios in different traits of different organisms, we arrive at conclusion that genes have their effects on phenotype through interacting allelic action as well as through interacting action of non-allelic action gene loci.

Practical 6: Solution of numerical problems related to gene actions

Question 1

Two curly winged fruit flies are mated. The F_1 consists of 341 curly and 170 normal. Explain.

Question 2

In families in which both parents have sickle cell trait. What is the probability of their having

(a) A child with sickle cell trait
(b) A normal child

Question 3

If a man of blood group AB marries a woman of blood group A whose father was of blood group O, to what different blood groups can this man and woman expect their children to belong ?

Question 4

In cattle the gene, R for red coat is not dominant over white r. The heterozygous Rr produces roan coat color. What phenotypes might be expected from the following mating ?

(a) Red x red, roan x roan, red x roan, roan x white, red x white, and white x white. Give the genotypes of the parents and progeny.

Question 5

From the following table, can the paternity of the children be established ?

Person	Antigen AB	Antigen MN	Antigen RH
Husband	O	M	RH+
Wife's lover	AB	MN	RH-
Wife	A	N	RH+
Child 1	O	MN	RH+
Child 2	A	N	RH+
Child 3	A	MN	RH-

Practical 7: Solution of numberical problems related to gene interactions

Question 1

In summer squash, fruits may be white, yellow or green. In one case the cross of **Y x W** produced an F_1 of all **W** fruited plants that, when selfed, gave an F_2 segregating 12 W, 3 Y, and 1 G,

(a) suggest genotypes for the **W**, **Y** and **G** phenotypes.
(b) give genotypes of the parents, F_1 and F_2 of this cross.
(c) if the F_2 progenies were found as 135 W and 105 Y phenotypes, explain the results giving genotypes of the parents, F_1 and F_2 of this cross.

Question 2

Consider the production of flower color in the *Pharbitis*. Dominant alleles of either of two separate genes (**A__bb** or **aaB__**) produce purple petals. **A__B__** produce blue petals and **aabb** produces scarlet petals. Deduce the genotypes of parents and progeny in the following crosses.

Parents	Progenies
Blue x Scarlet	$\frac{1}{4}B$, $\frac{1}{2}P$, $\frac{1}{4}S$
Purple x Purple	$\frac{1}{4}B$, $\frac{1}{2}P$, $\frac{1}{4}S$
Blue x Blue	$\frac{3}{4}B$, $\frac{1}{4}P$
Blue x Purple	$\frac{3}{8}B$, $\frac{4}{8}P$, $\frac{1}{8}S$
Purple x Scarlet	$\frac{1}{2}P$, $\frac{1}{2}S$

Question 3

Radishes may be long, round or oval and they may be red, white or purple. You cross a long white variety with round red one and obtain an oval purple F_1. Following nine phenotypic classes are observed:

- 9 long red,
- 15 long purple,
- 19 oval red,
- 32 oval purple,
- 8 long white,
- 16 round purple,
- 8 round white,
- 16 oval white and
- 9 round red

(a) Provide a genetic explanation of these results. Be sure to define the genotypes and show the constitution of parents in F_1 and F_2.
(b) Predict the genotypic and phenotypic proportions in the progeny of a cross between a long purple radish and an oval purple one.

Question 4

In sweet pea, dominnat genes **C** and **R** together produce colored flowers. Determine the gametes, and the genotypes and phenotypes of progeny obtained in the following crosses.

(a) **CCRR** x **ccrr**
(b) **CcRr** x **CcRr**
(c) **CcRr** x **ccrr**
(d) **CcRr** x **CCrr**
(e) **ccRr** x **Ccrr**
(f) **CCRr** x **ccRr**
(g) **CCRR** x **CCRr**

Question 5

In maize, gene **R** produces red aleurone color, while gene **P**, when present together with **R**, produces purple color. Determine the phenotypes fo parents, and the genotypes and phenotypes of the progeny produced from the following crosses.

(a) **RRpp** x **Rrpp**
(b) **RRpp** x **RrPP**
(c) **RrPp** x **Rrpp**
(d) **rrPP** x **RrPP**
(e) **RrPp** x **RrPp**
(f) **RrPp** x **rrPp**

Practical 8: Probability

Problem 1

If 5 coins are tossed together, determine the probability of getting

1. 3H and 2T
2. At least 3H
3. More than 3H
4. Less than 3H
5. Not more than 3H

Problem 2

Two heterozygous brown-eyed (Bb) individuals have five children. What is the probability that two of the couple's five children will have blue eye ?

Problem 3

How often in families of 5 children where one parent is heterozygous for malaria resistance and other is homozygous for malaria resistance will the children be 3 resistant girls and 2 resistant boys ?

Problem 4

Albinism in human is controlled by a recessive gene c. From a marriage between two partners, if both partners are carriers, Cc for albinism, what is the chance that:

i. All 4 children of them are normal
ii. 3 normal and 1 albino
iii. 1 normal and 3 albino
iv. All 4 are albinos

Problem 5

How often in families of 3 children where both parents are heterozygous for non-red hair (Rr) and have wavy hair (h_1h_2) will children be

i. a red haired girl, a non-red curly haired boy and a red straight haired girl ?
ii. two red wavy haired boy and one non-red straight haired girl ?

Practical 9: χ^2 test

Problem 1

The F1 generation from a cross between albino and black strains of mice produced 75 black and 33 albino progeny.

i. What is the expected ratio of offsprings in this case ?
ii. State the null hypothesis.
iii. Determine the degree of freedom.
iv. Carry out χ^2 test.
v. Is any correction required in emperical formula for computation of χ^2 statistic in this case ? Why ? vi Draw the conclusion.

Problem 2

A presumed dihybrid in Drosophila, B/b ; F/f is test- crossed with b/b ; f/f. (B = black body ; b = brown body; F = forked bristles; f = unforked bristles.) The results are,

- Black, forked (BF) : 230
- Black, unforked (Bf) : 210
- Brown, forked (bF) : 240
- Brown, unforked (bf) : 250

Use the Chi-square test to determine if these results fit the results expected from testcrossing the hypothesized dihybrid.

Practical 10: Solution of numerical problems related to linkage analysis and crossing over

Question 1

In Drosophila the allele *dp*+ determines long wings and *dp* determines short wings. At a separate locus *e*+ determines the gray body and *e* determines ebony body. Both loci are autosomal. Pure line of long ebony parent was crossed to pureline of short gray and produced long gray F_1. The F_1 was crossed to pure line of short ebony and the following progeny were obtained.

- Long ebony: 54
- Long gray: 47
- Short gray: 52
- Short ebony: 47

Use χ^2 test to determine if these loci are linked.

Question 2

Two different plants heterozygous for the concerned genes are self-pollinated and in F_2 following data was recorded:

Plant 1		Plant 2	
Phenotypic class	Frequency	Phenotypic class	Frequency
A_B_	250	C_D_	510
A_bb	45	C_dd	240
aaB_	35	ccD_	240
aabb	70	ccdd	10

i. Are the genes *a* and *b*, and *c* and *d* linked ? If so, in which phase ?
ii. Determine the frequency of recombination between the genes a and b, and c and d.

Question 3

In maize, F1 heterozygous plants were test crossed with colourless, shrunken, waxy plants and the following types of progeny were obtained.

Progeny type	Frequency
CfS	50
cFs	46
CFs	383
cfS	380
Cfs	72
cFS	68
CFS	6
cfs	5

Symbols: coloured= C, colourless= c, full= F, shrunken= f, starchy= S, and waxy= s

i) Are these genes linked? Give reason.
ii) Write the genes in correct order on the chromosome.
iii) What are double crossover, non- crossover and single crossover types?
iv) Write the genotypes involved in the parental and test crosses.
v) Draw a linkage map showing map distances.
vi) Calculate Coefficient of coincidence (CC) and interference (I). Interpret the value.

Question 4

Female F1 maize heterozygous for tall producing round and starchy seed was test-crossed and the following progenies were obtained.

- Tall round starchy :10,
- Dwarf shrunken waxy : 05,
- Tall shrunken waxy : 300,
- Dwarf round starchy : 350,
- Tall round waxy :70,
- Tall shrunken starchy : 135,
- Dwarf round waxy : 70,
- Dwarf shrunken starchy : 60

i. Is there linkage between genes?
ii. Draw a linkage map of the linked genes showing the correct genes order and their map distances.
iii. What are double crossover, non crossover and single crossover types?
iv. Write the genotypes of flies involved in the parental and test crosses.
v. Diagram the cross showing the arrangement of the genetic markers on the chromosomes.
vi. Calculate I and CC and interpret the results.

Question 5

An individual heterozygous for four genes; Aa.Bb.Cc.Dd is test crossed to aabbccdd and 1000 progeny were classified by the gametic contribution of the heterozygous parent as follows:

1. Which genes are linked ?
2. If two pure breeding lines had been crossed to produce the heterozygous individual, what would their genotypes have been ?
3. Draw a linkage map of the linked genes showing the correct genes order and their map distances.
4. Calculate CC and I.

Progeny type	Numbers
aBCD	42
Abcd	43
ABCd	140
abcD	145
aBcD	6
AbCd	9
ABcd	305
abCD	310

Practical 11: Solution of numerical problems related to sex determination and linkage

Question 1

What is the sex designation for each of the following individuals in human ?

i. AAXXX
ii. AAXXXYYY
iii. AAXO
iv. AAXXXXXY
v. AAXYYY
vi. AAXXY

Question 2

In poultry, sex linked allele, B produces barred feather pattern (dominant to b – non-barred). Autosomal allele R produces rose comb and its recessive allele form, r produces single comb in homozygous recessive state. What is the F_1 phenotype obtained by crossing barred female homozygous for rose comb to non-barred single comb male ? Write the genotypes of the parents and F_1.

Question 3

A sex-linked recessive allele c produces a red-green color blindness in humans. A normal woman whose father was color blind marries a color-blind man.

- What genotypes are possible for the mother of the color blind man ?
- What are the chances that the first child from this marriage will be a color-blind boy ?
- Of the girls produced by these parents, what proportion can be expected to be color blind ?
- Of all the children (sex unspecified) of these parents, what proportion can be expected to have normal color vision ?

Question 4

The recessive gene e, produces color blind in human, which is sex linked gene. A carrier woman of this gene married a color blind man and produced first child, a normal vision klinefelter but in the second time this couple gave birth a color blind daughter. How such klinefelter and daughter may be produced? Show with the help of clean diagram.

Question 5

In maize, recessive gene ba produces barren cob and ts gene converts male flower into female flower. Ba and Ts are the dominant genes give the normal cob and male flowers, respectively. A geneticist working at IAAS crossed a plant having genotype Baba Tsts with other plant having genotype BaBa Tsts. Show this cross and determine the sex expression in the progeny.

Practical 12: Microscopic study of DNA and RNA

Practical 13: Field demonstration of segration in crops

Practical 14: Field demonstration of cytoplasmic/genetic male sterility