

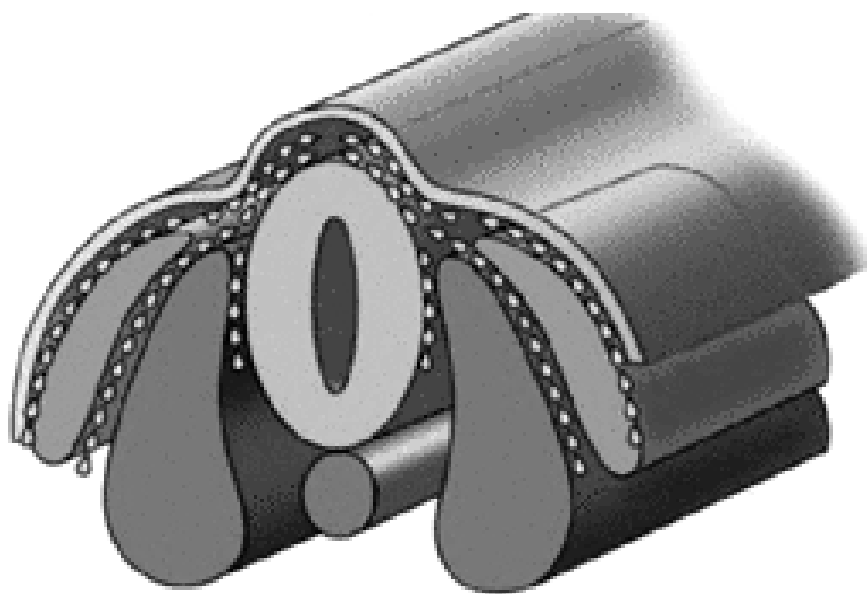
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"GENERAL BIOLOGY"

PART A



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This manual is designed according to syllabus of biology for students of Medical faculty. It will be a guide to action for students during their practical work.

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3.2. Fertilization

3.3. Cleavage

3.4. Gastrulation

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3.6. Extraembryonic membranes

3.7. Human development

Unit 4: Genetics

4.1. Heredity. Mendel G. is foundation of modern genetics.....

4.2. Interaction of genes

4.3. Chromosomes - a unit of heredity

4.4. Sex linkage

4.5. Genetics problems

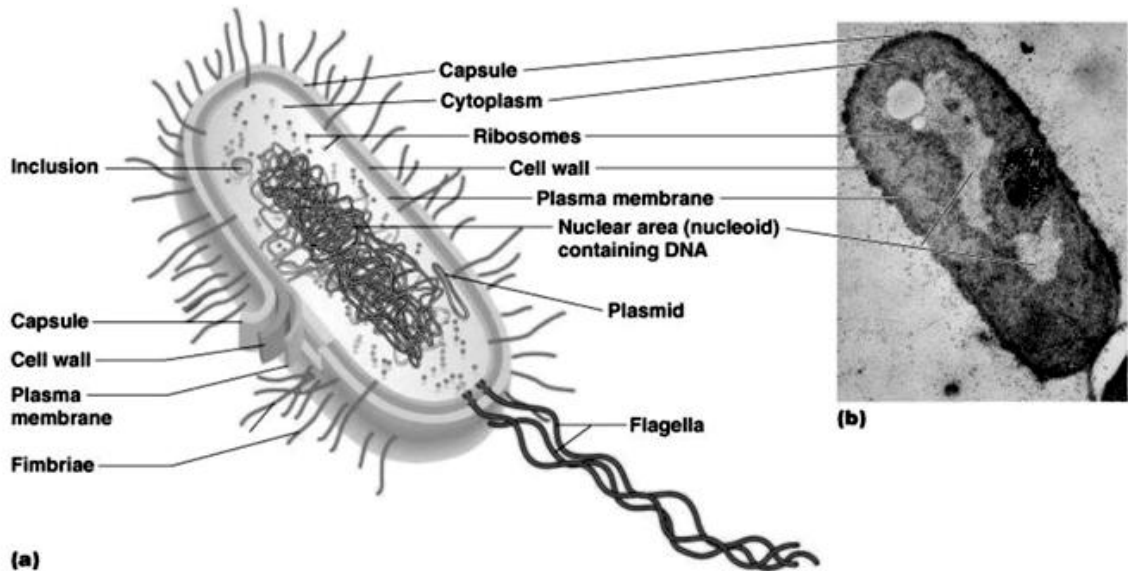
4.6. Variation. Modificational variability

4.7. Mutational variability

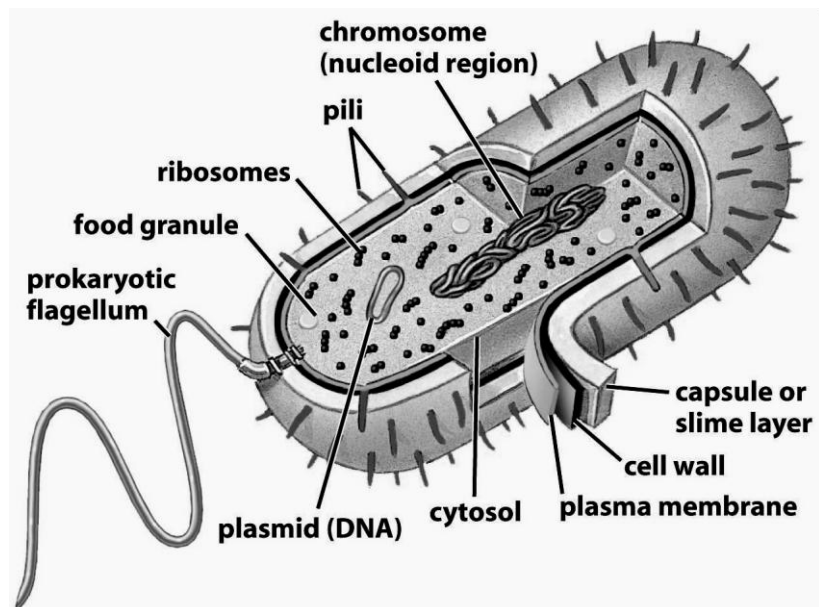
4.8. Combinative variability

UNIT 1: BACTERIAL CELLS

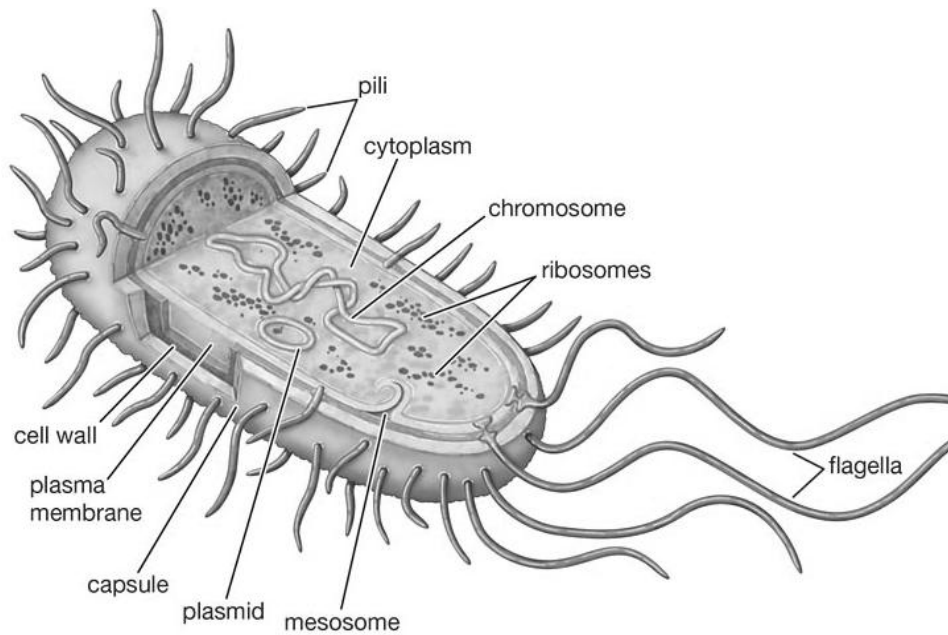
Bacteria are single-celled organisms. They are prokaryotes. Bacterial cells have no nucleus.



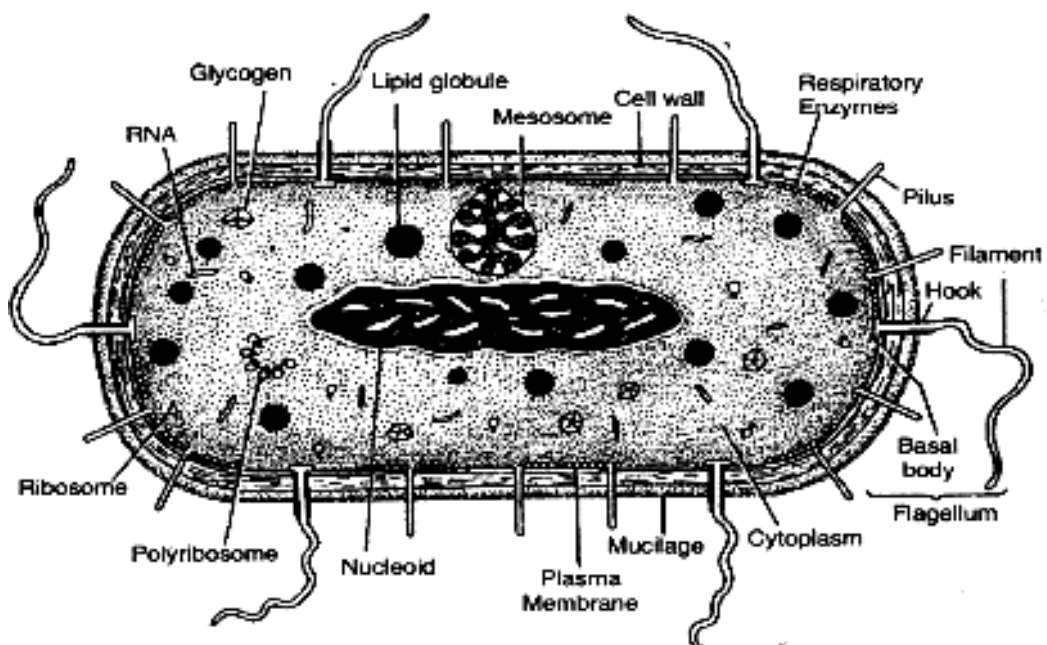
Their circular double-stranded DNA is free located in the cytoplasm. Bacterial DNA forms a complex with the non-histone proteins, forming nucleoid.



Bacterial cells surrounded by an outer membrane. The cell wall is arranged outside the membrane and comprises a polysaccharide murein. Some bacteria may be covered with a mucous capsule.



Bacteria do not have a membrane organelles. Bacteria have no cell center, microtubules, microfilaments. Bacteria have only 70S ribosomes. Some bacteria may have flagellum. The bacterial flagellum has a simple structure. It is a protein strand and unlimited membrane.



THE FORMS OF BACTERIA

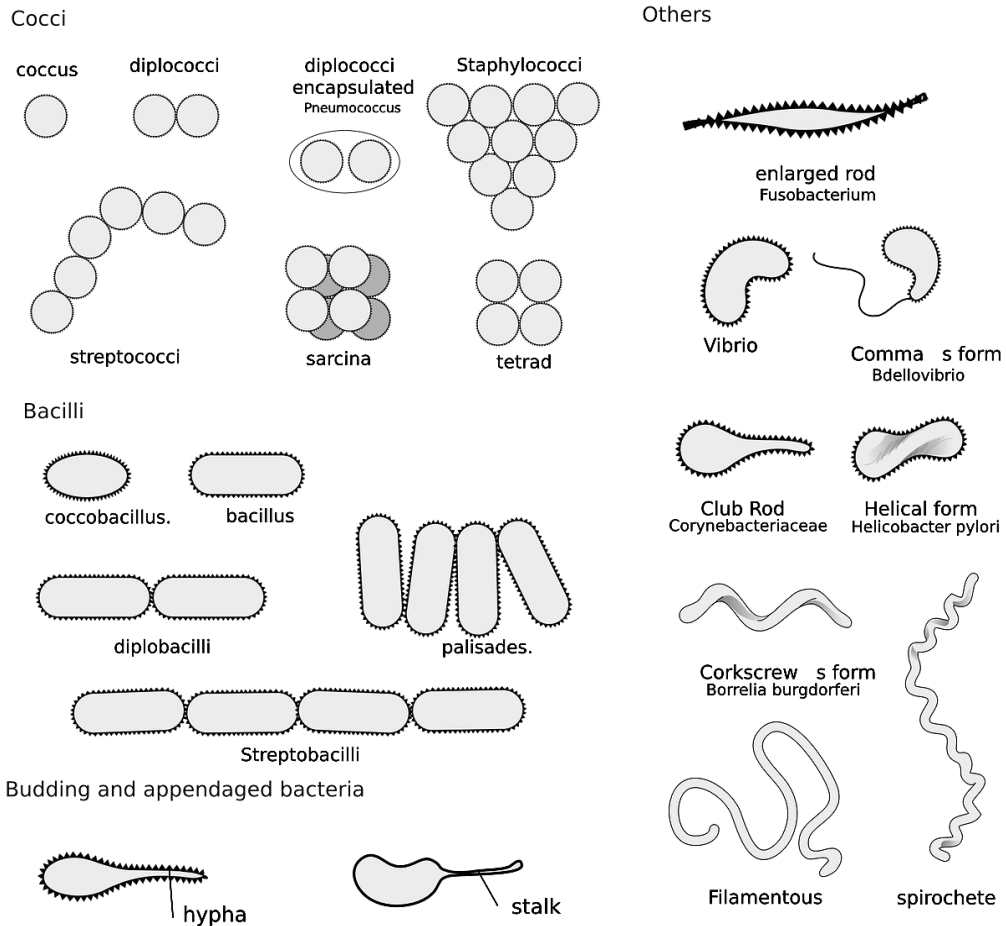
Prokaryotic cells have various shapes; the four basic shapes of bacteria are:

Cocci – spherical

Bacilli – rod-shaped

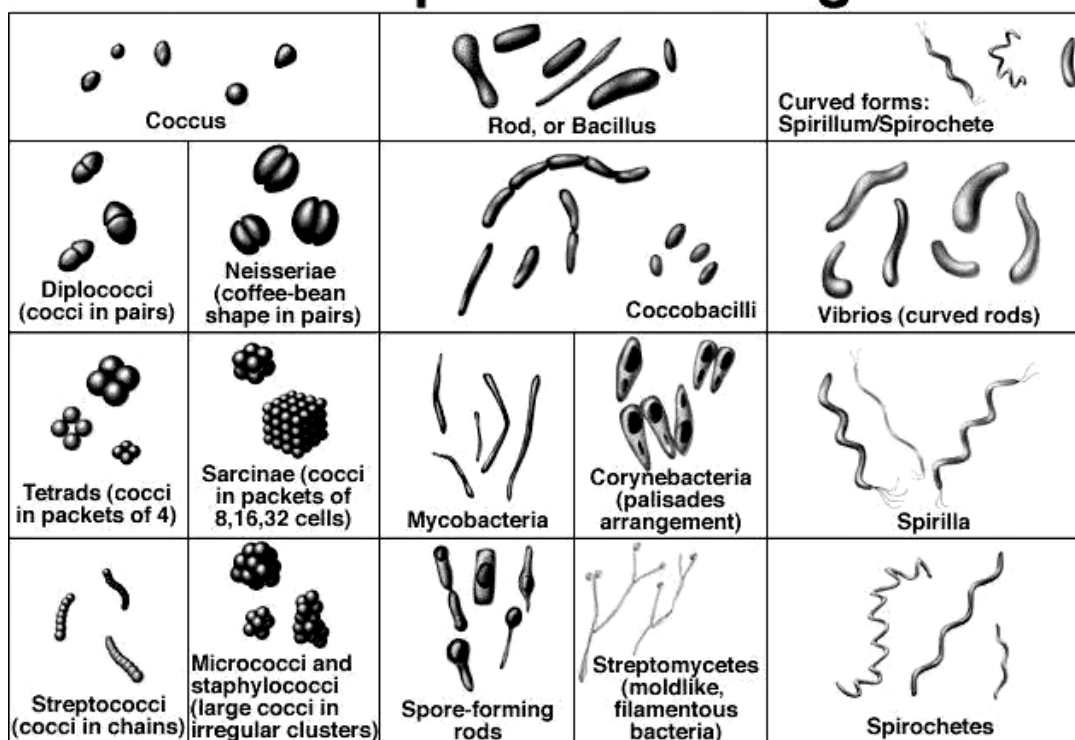
Spirochaete – spiral-shaped

Vibrio – comma-shaped



By way of nutrition bacteria can be divided into autotrophic and heterotrophic organisms.

Bacterial shapes and arrangements



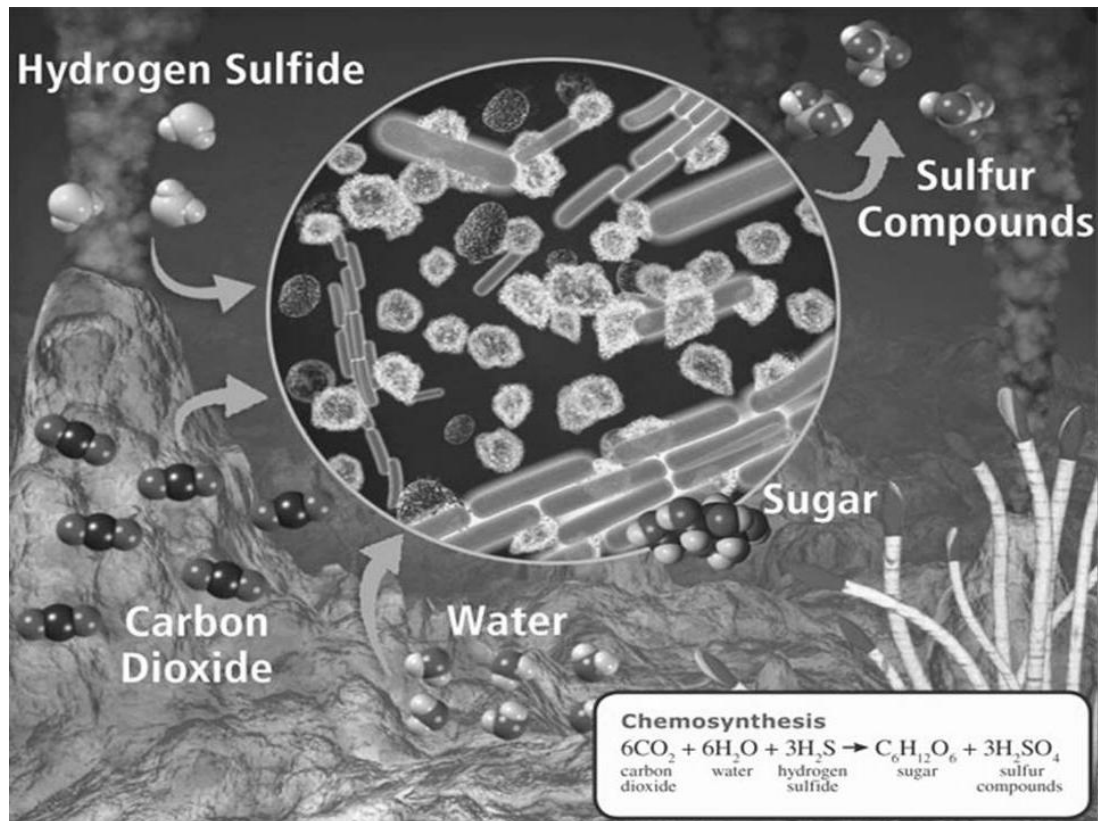
Autotrophic bacteria are divided into photosynthetic bacteria and chemosynthetic bacteria.

Energy-producing oxidation reaction	Type of bacteria
$2\text{H}_2 + \text{O}_2 \rightarrow 2\text{H}_2\text{O}$	Hydrogen bacteria
$2\text{H}_2\text{S} \rightarrow \text{S} \rightarrow \text{S}_2\text{O}_3^{2-} \rightarrow \text{SO}_4^{2-}$	Colorless sulfur bacteria
$\text{Fe}^{2+} \rightarrow \text{Fe}^{3+}$	Iron bacteria
$\text{NH}_3 \rightarrow \text{NO}_2^- \rightarrow \text{NO}_3^-$	Nitrate, nitrite bacteria

Heterotrophic bacteria are divided into the following types:

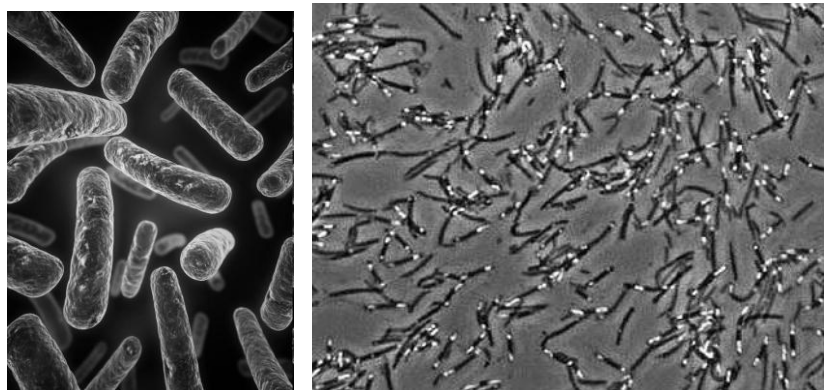
1. Bacteria - parasites. They feed on living organisms and cause various diseases.
2. Bacteria - saprotrophs. They feed on the dead remains. These bacteria convert the organic matter of the dead bodies in minerals. Thus closes the cycle of substances in nature.

3. The bacteria - symbionts. These bacteria form a mutually beneficial relationship with other organisms. For example E. coli in the human colon helps to digest cellulose, creates an acidic environment where harmful bacteria and fungi do not develop, are involved in the formation of some vitamins

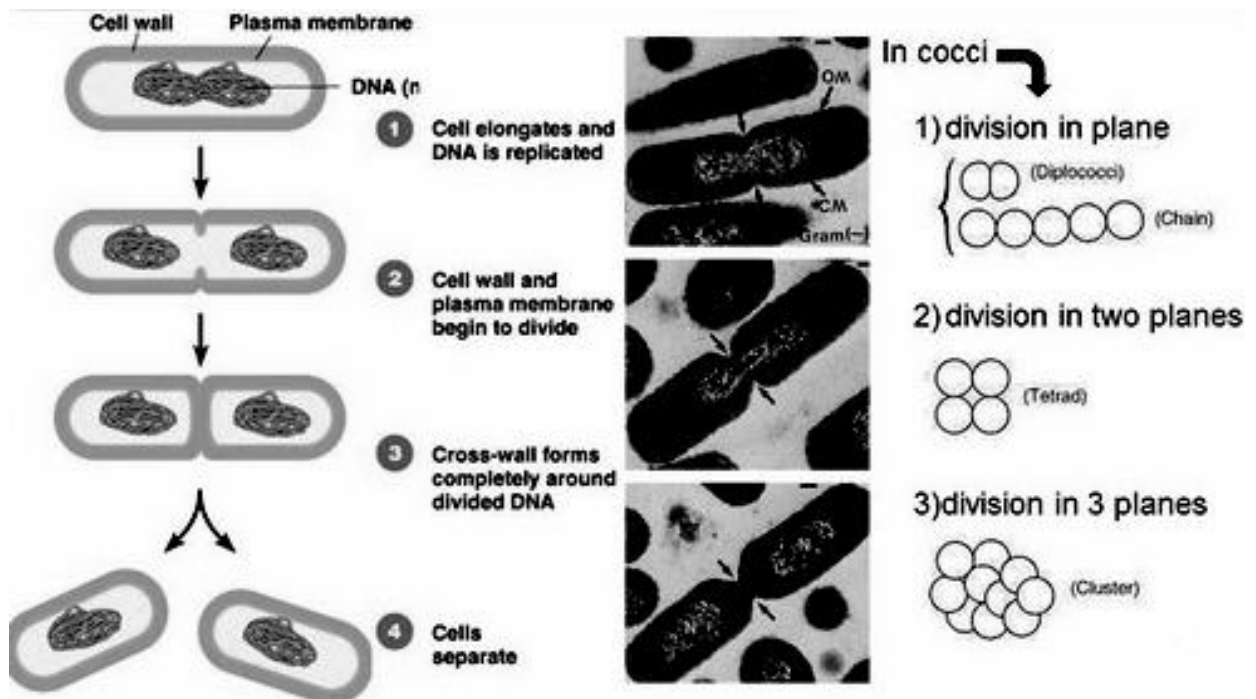


The bacteria can be aerobic and anaerobic respiration by the method.

The bacteria survive in the form of spores in adverse conditions , and for a long time can survive.



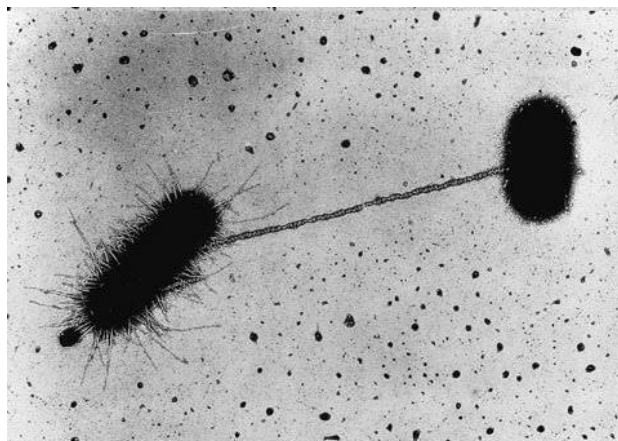
Bacteria reproduce through asexual reproduction, usually by binary fission.



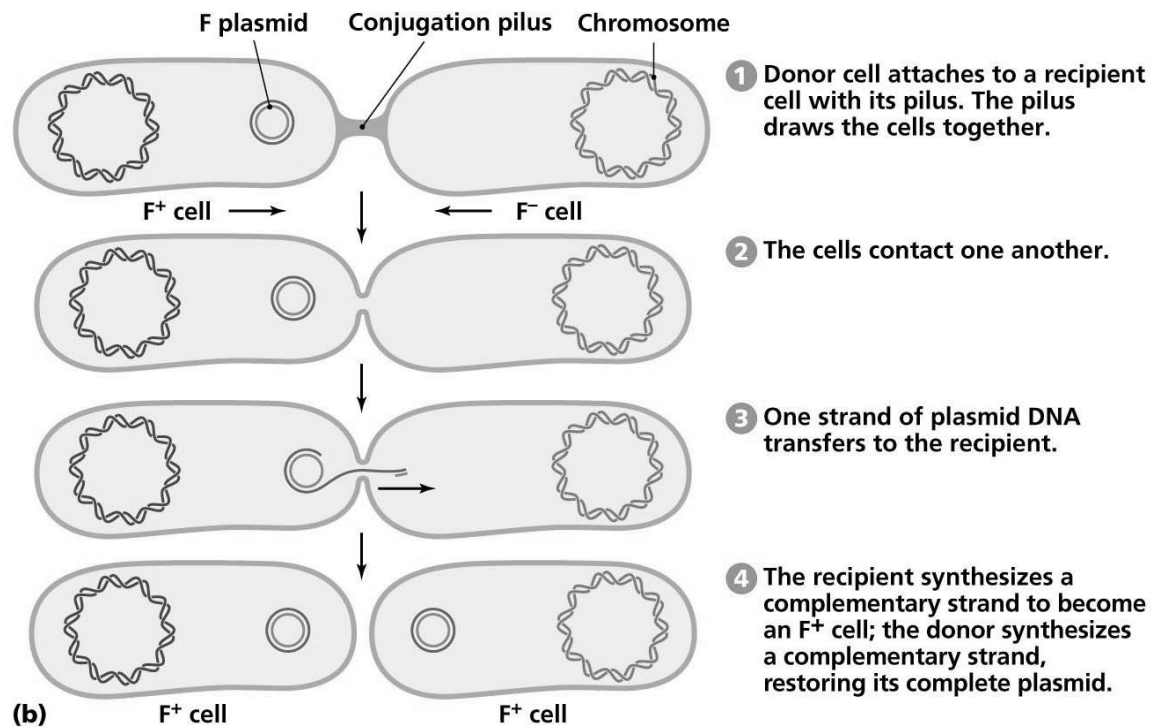
Bacteria do not have a sexual reproduction, but there is sexual recombination. There are three types of sexual recombination: conjugation, transformation, transduction.

Conjugation.

Some bacteria are in addition to the basic DNA (the bacterial chromosome) has additional DNA called plasmids.

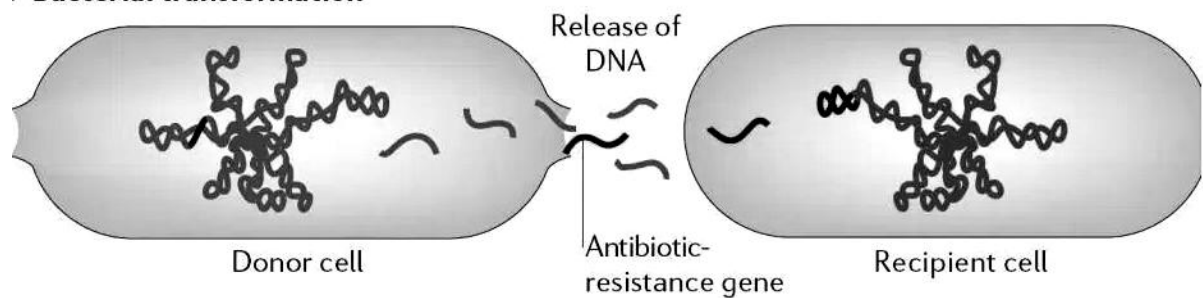


Donor bacterium having the plasmid may form cytoplasmic bridge by using sexual rod with a bacterium which has no plasmid. First, double-stranded plasmid DNA is separated into two chains. One of the chains is transferred to the recipient bacterium. Then, each bacterium completes the missing chain on the principle of complementarity.

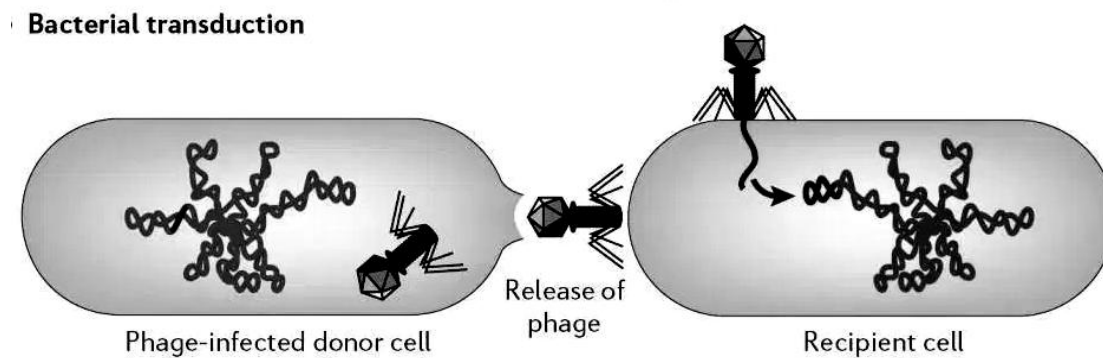


Transformation - the transfer of a fragment of DNA from dead bacteria to live, where the fragment replaces the homologous site.

Bacterial transformation

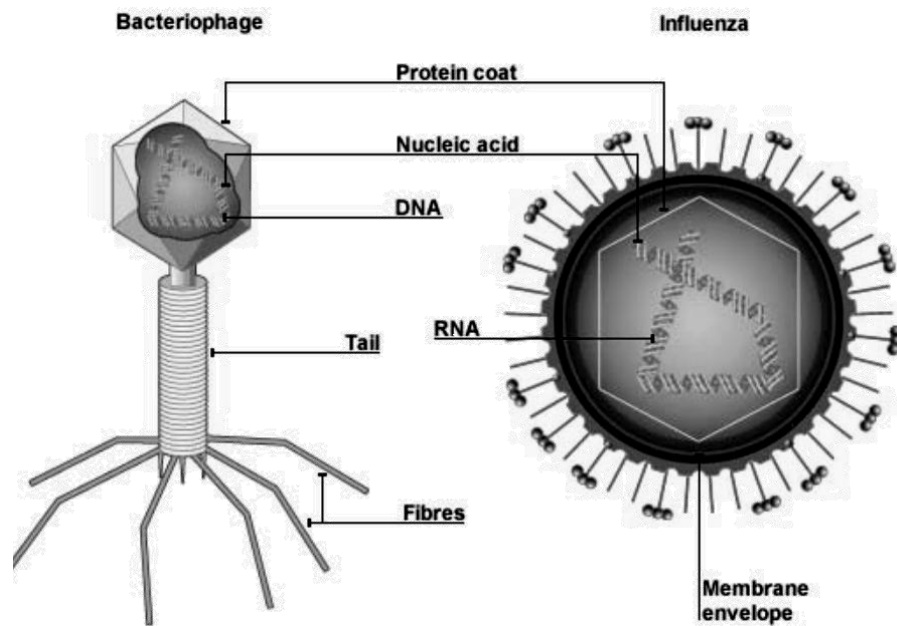


Transduction - the transfer of DNA fragments from one bacterium to another by using viruses - bacteriophages. Penetrating into the bacterial cell the virus multiplies and forms a protein capsid. Sometimes virus capsid randomly encompasses fragments of bacterial DNA. This forms a transducing particle. When such a virus infects a new mutated bacterium, it injects its DNA fragment of the first bacterium and second bacterium acquires new properties.

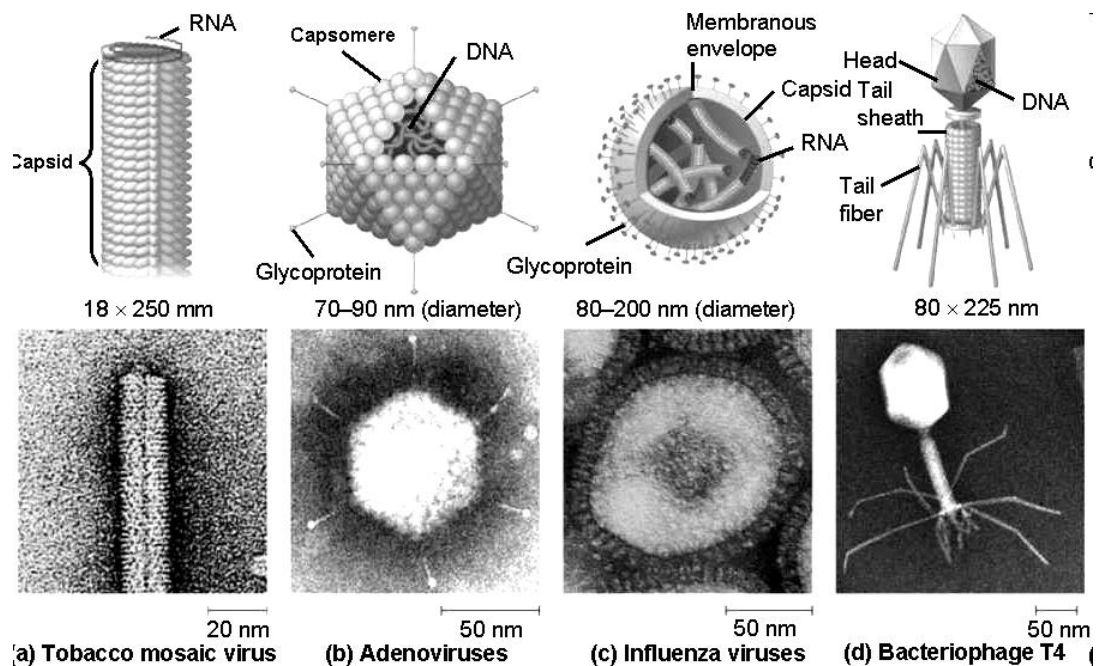


UNIT 2: VIRUSES

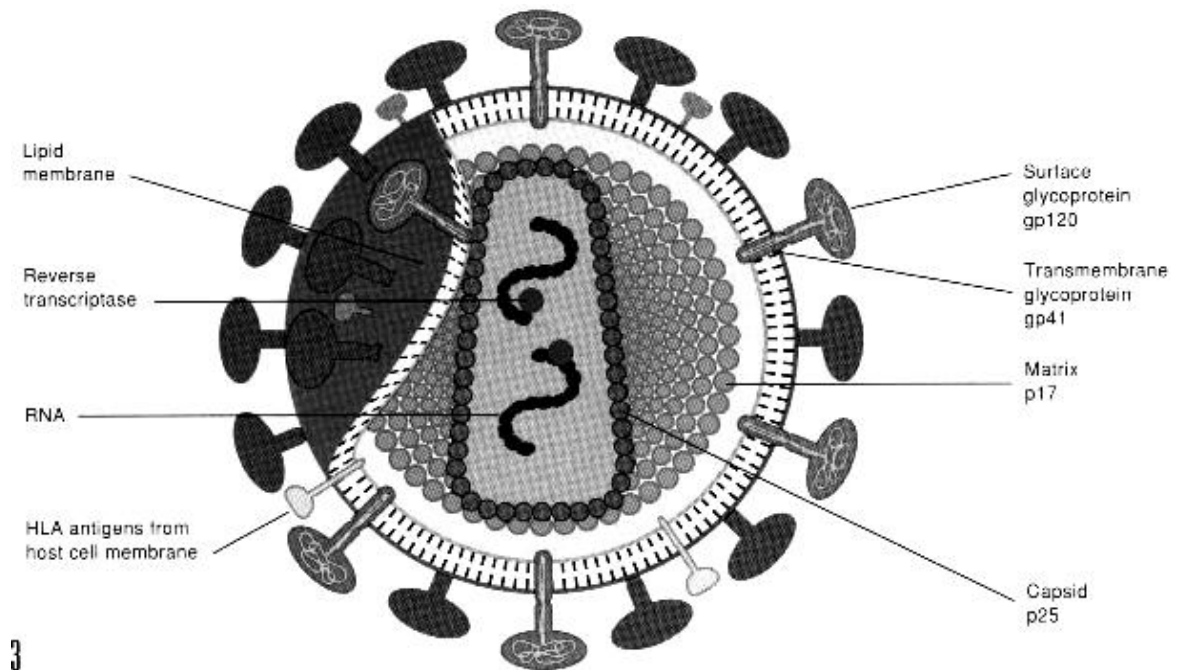
Viruses - organisms having no cellular structure. They consist of nucleic acid (DNA or RNA), which is surrounded by a protein shell called a capsid. Above the capsid may be located lipoprotein envelope, which often is a fragment of the membrane of the host cell from which the virus was released.



Metabolism, growth and development are processes that are not typical for viruses.



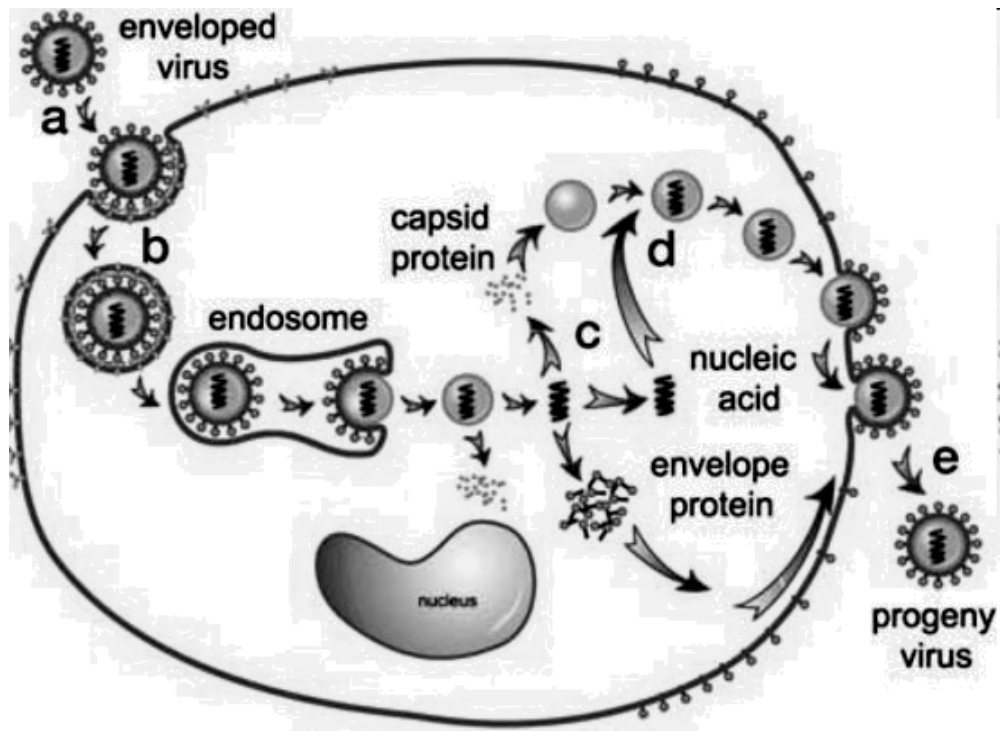
Viruses are permanent parasites that exhibit properties of living within the host cell. Heredity, variation and reproduction are characterized for viruses. Outside the host cell viruses behave like structure of inanimate nature. Therefore, viruses is a bridge between the animate and inanimate nature.



Viruses are very specific. They penetrate into the cells of a specific type only. Viruses have the receptors to specific cells. For example, hepatitis B virus affects the liver cells the AIDS virus affects blood lymphocytes.

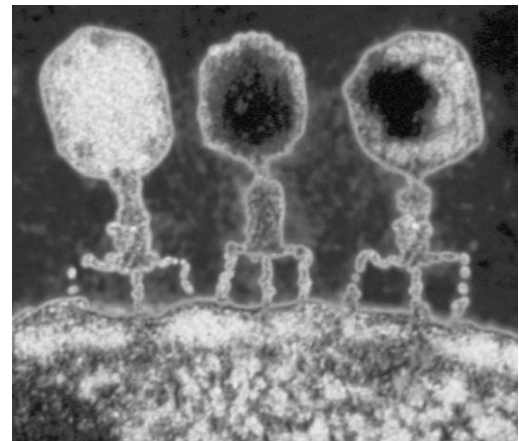
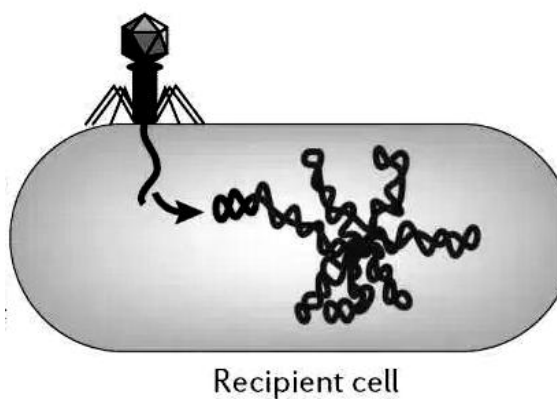
THE BEHAVIOR OF THE VIRUS IN THE CELL

First, the virus recognizes the cell by using receptors. It then enters the cell by one of the methods. Animal viruses enter the cell by phagocytosis.

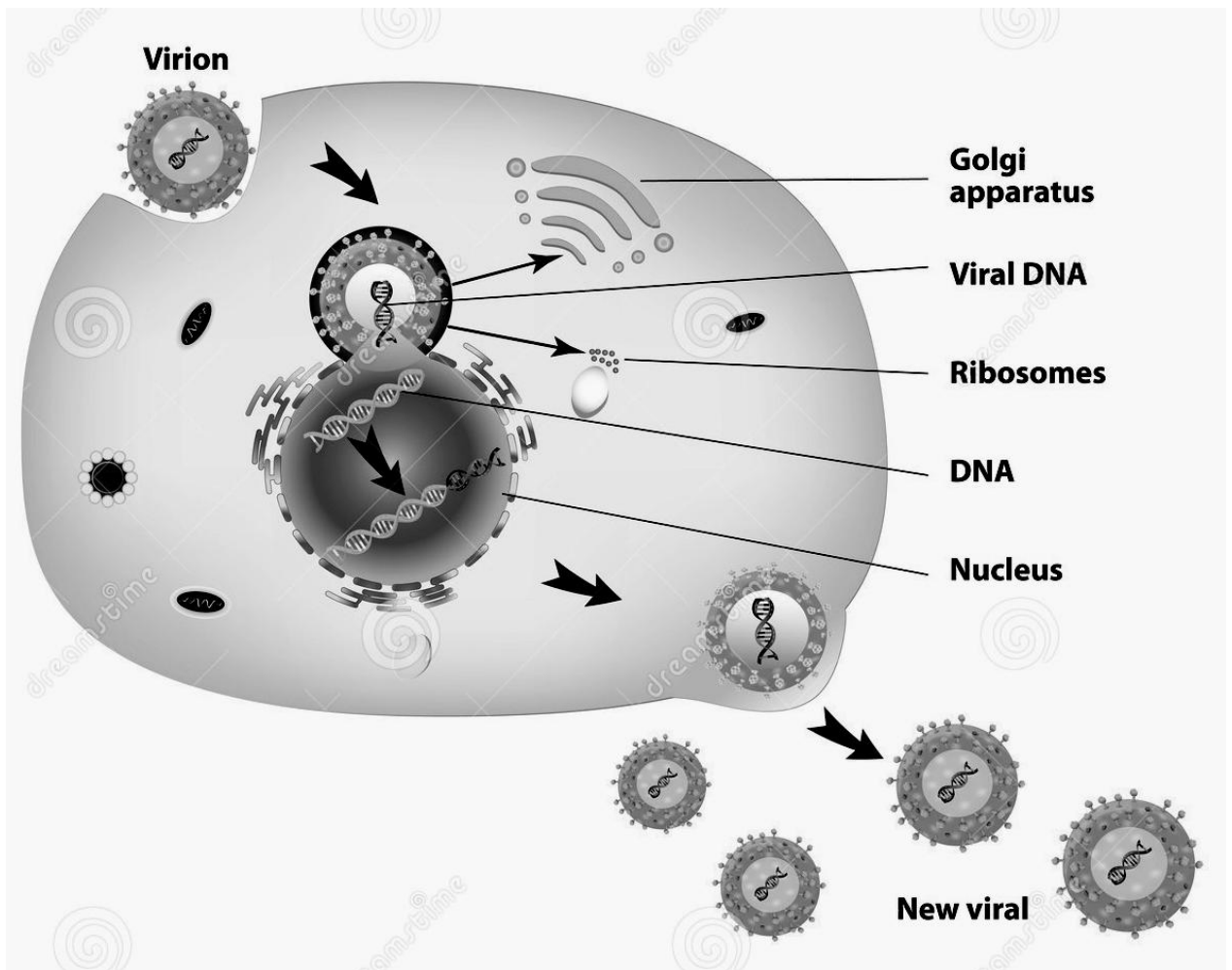


Plant viruses and fungi viruses penetrate into the cell through the damaged cell wall.

Viruses bacteria - bacteriophages - enter the cell by injection.



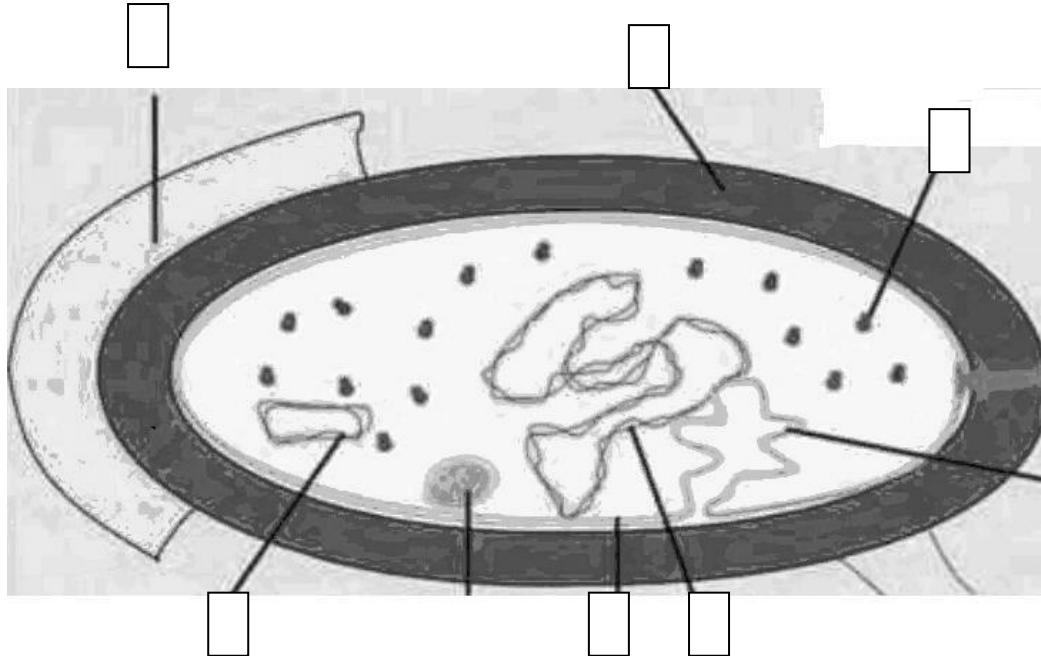
Once in the cytoplasm of the cell, the nucleic acid of the virus is copied many times. Organelles and materials of the host cell are used for this. The virus protein coat (capsid) is formed on the base of the viral nucleic acid.



Testing your knowledge

Bacteria and viruses

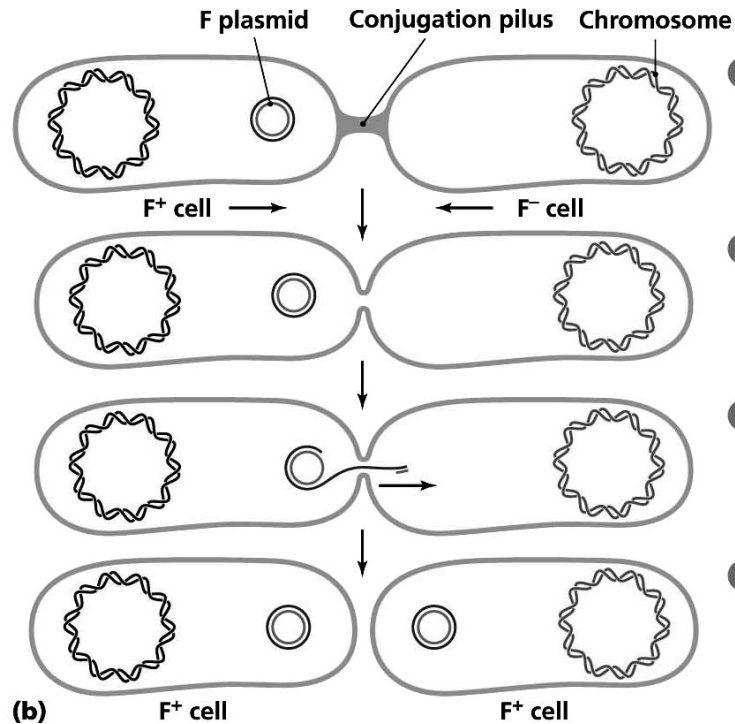
Task 1. Consider the structure of bacteria. Draw the scheme and make a mark. What structures are labeled 1-6?



Task 2. Fill in the table "The difference between prokaryotes from eukaryotes"

Main features	Prokaryotes	Eukaryotes
Polysaccharide which is part of the cell wall		
The presence of the nucleus		
DNA characterization		
The presence of membranous organelles		
The presence of nonmembranous organelles		
The presence of plasmids		
Kinds of reproduction		

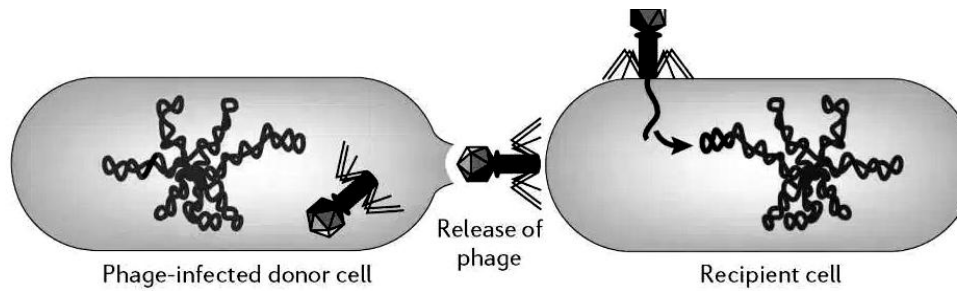
Task 3. Consider the scheme of bacterial conjugation.



Complete the sentences:

1. Some bacteria has additional DNA called _____.
2. Donor bacterium having the plasmid may form _____ by using sexual rod with a bacterium which has no plasmid.
3. First, double-stranded plasmid DNA is separated into two chains. One of the chains is transferred to _____.
4. Then, each bacterium completes the missing chain on the principle of _____.

Task 4. Consider the scheme of bacterial transduction.



Answer the questions in writing:

1. Who carries the fragment of DNA from one bacterium to another during transduction?
2. What to do after the penetration of the virus into a bacterium?
3. What is produced when the virus capsid surrounds the DNA of the bacteria?

Task 5. Read the text, find errors and correct them.

1. Viruses are permanent parasites that exhibit properties of living outside the host cell.
2. Heredity, variation and reproduction are characterized for viruses.
3. Outside the host cell viruses behave like structure of animate nature.
4. The viruses is a bridge between the animate and inanimate nature.
5. Viruses are not specific. They penetrate into the cells of a different type.
6. Viruses have the receptors to specific cells.
7. Animal viruses enter the cell by through the damaged cell wall.
8. Viruses bacteria - bacteriophages - enter the cell by injection.
9. The virus protein coat (capsid) is formed on the base of the nucleic acid of the host cell.
10. Virions leave the cell, either simultaneously or progressively by one.

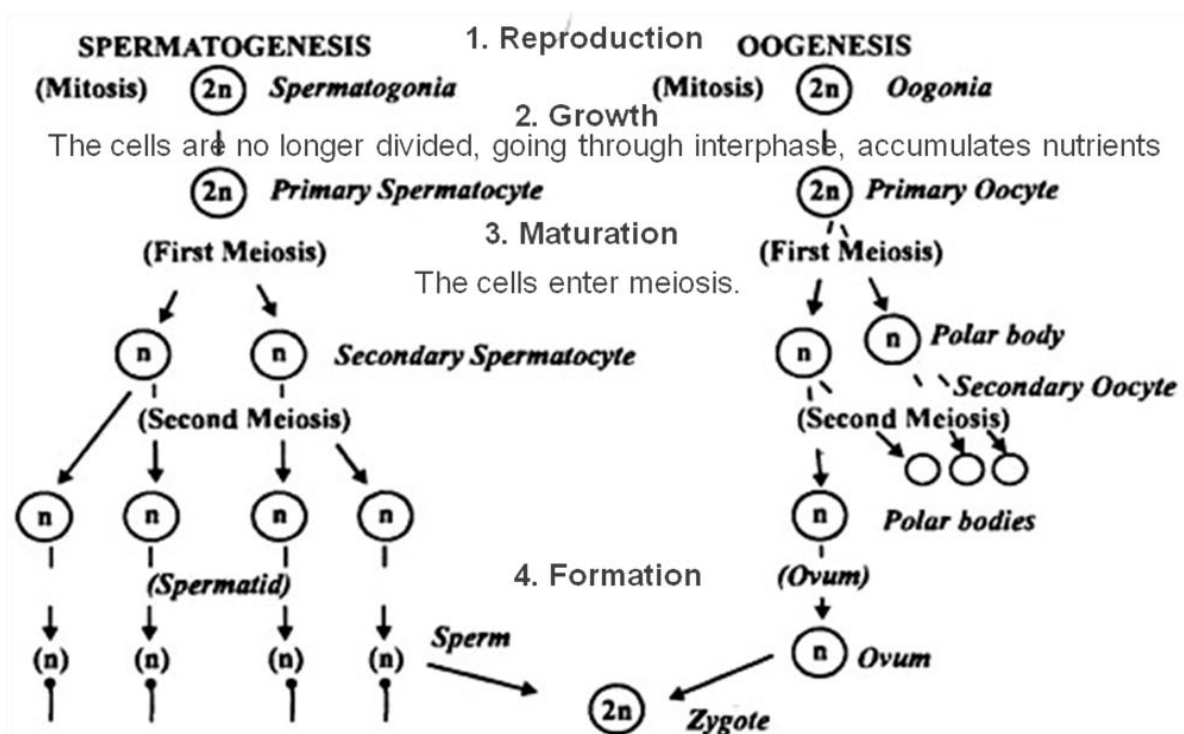
UNIT 3: ONTOGENESIS - THE PROCESS OF INDIVIDUAL DEVELOPMENT OF ORGANISMS

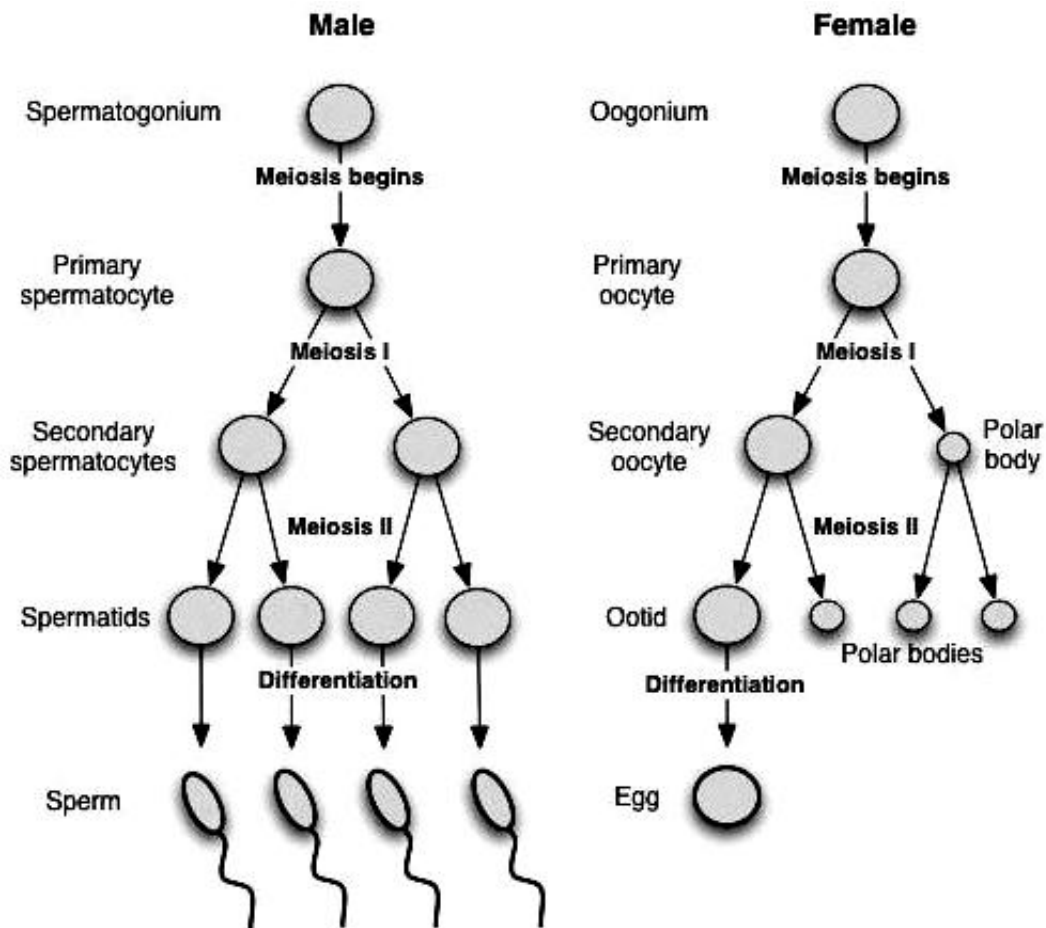
Ontogenesis - the process of individual development of organisms.

Three periods of ontogenesis can be identified.

1. Pre Eembryonic period includes gametogenesis and fertilization.
2. Embryonic period includes cleavage, gastrulation, primary and final organogenesis.
3. Postembrionic period begins with birth or hatching and ends in death.

GAMETOGENESIS





Spermatozoa is the male reproductive cells. They are 60-70 mkm long and have a very specific shape. Each spermatozoon has a head, neck and tail.

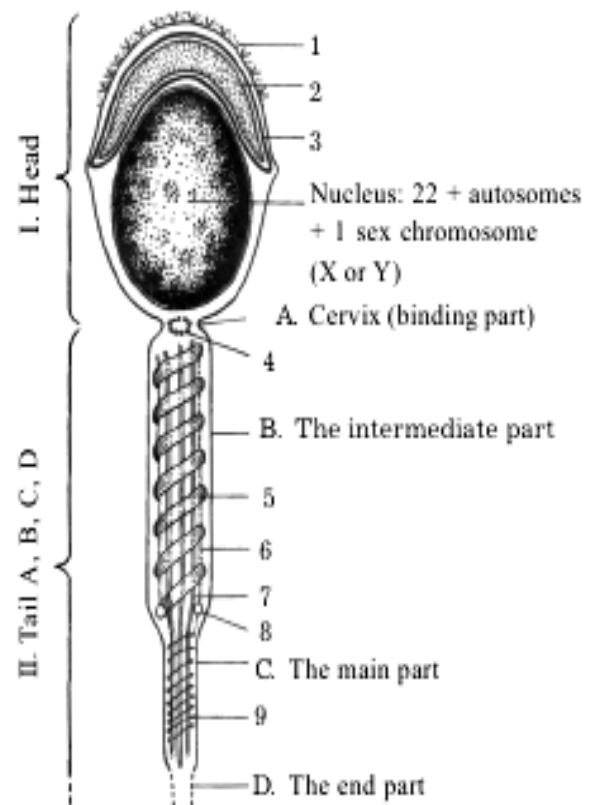
The head of spermatozoon is of oval flattened shape. It is the most suitable shape for passage forward due to the moving of flagella. Most of the sperm's cytoplasm is eliminated during maturation, leaving only certain organelles that are modified for spermatic function. There is a special structure – acrosome (acrosomal vesicle) – at the top of spermatozoon head. It is a specifically changed Golgi apparatus, which contains enzymes hyaluronidase and trypsin.

The neck contain a proximal centriol.

The tail consist of 3 parts; intermediate, main, end (terminal). Axial fibre is the principal component of the all tail. In the intermediate part there are many mitochondria which supply energy to him.

Fig. 1.7. Scheme of structure of human sperm: I - head; II - tail.

1 - receptors of glycocalyx; 2 - acrosome; 3 - outer and inner acrosome membranes; 4 - proximal centrioles, 5 - mitochondrion, 6 - layer of elastic fibrils, 7 - axonema, 8 - distal centrioles; 9 - fibrous vagina (longitudinal pillars and circular fibrils, which consist of keratin filaments).

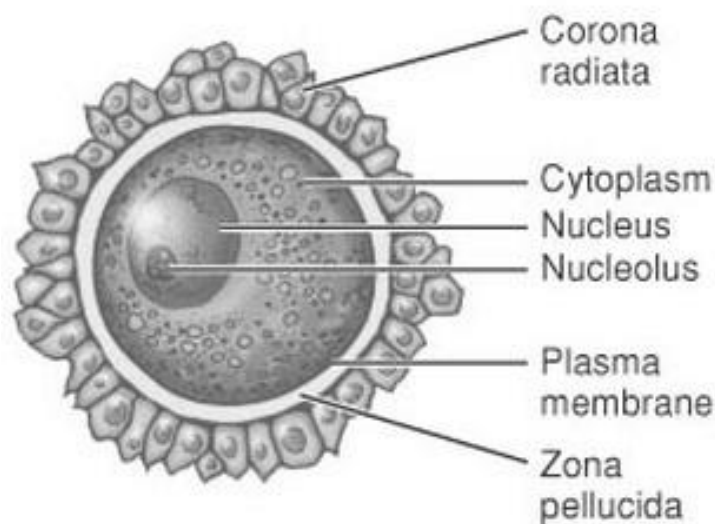


Each spermatozoa is able to travel long distances by whipping its tail. The tail is flagellum. Flagellum is complex structures. The major motor portion of the flagellum is called the axoneme. It is formed by microtubules emanating from the centriole at the base of the sperm nucleus. The core of the axoneme consists of two central microtubules surrounded by a row of nine doublet microtubules. Actually, only one microtubule of each doublet is complete, having 13 protofilaments; the other is C-shaped and has only 11 protofilaments. Although tubulin is the basis for the structure of the flagellum, other proteins are also critical for flagellar function. The force for spermatozoa propulsion is provided by dynein, a protein that is attached to the microtubules. Dynein hydrolyzes molecules of ATP and can convert the released chemical energy into the mechanical energy that propels the spermatozoa.

Biologic significance of spermatozoa in the fertilization.

1. They give 23 “father” chromosomes.
2. The sex of future embryo depends on spermatozoon type. (x or y)
3. Give the centriol, mitochondrial DNA.

Oocyte (ovum) is female germ cell. It is the largest cell (130-150 μm in diameter) in human body which can not move. It is covered with a few tunics. From outside they are: corona radiata (tunica granulosa), zona pellucida and oolemma. The oolemma is the cytolemma. Zona pellucida is a special chemical membrane which contains much glycoproteins and glycosaminoglycans. Glycoproteins presents by three types: Zp1, Zp2, Zp3. Zp3 is receptor for spermatozoa, Zp2, Zp3 prevent polysperm. The outer layer consist of numerous follicular cells which give their processes to the ovum through the previous tunic. Their functions are protection and nutrition of the oocyte and regulation of its maturation.



Nucleus of the ovum excentrically and contains 23 chromosomes: 22 autosomes and the last one – sex chromosome (only x).

There are all the organells of a general meaning in the oocyte cytoplasm. Apparatus Goldgy is especially well developed and produces yolk inclusions.

In the peripheral layer of cytoplasm just under the plasmolemma numerous cortical granules may be seen. They take an active part in the process of fertilization.

Classification of oocytes due to the amount and location of yolk inclusions.

1. *Alecytal oocyte* have no inclusions (insects).

2. ***Oligolecytral oocyte*** contain little yolk inclusions. Oligolecytral oocyte subdivided into two type oocyte: primary isolecytral and secondary isolecytral. In primary isolecytral oocytes nuclei in the center of cells, in secondary isolecytral (human oocyte) nuclei are disposed, yolk inclusions uniformly distributed in cytoplasm.
3. ***Mesolecytral oocyte*** contain the average number of yolk inclusions.
4. Fish and birds have ***polylecytral oocytes*** with great volume of yolk.

Biologic significance of oocyte in the fertilization process.

1. They give half of a necessary amount of chromosomes.
2. Supply the nutrition of embryo.

FERTILIZATION

There are three stages of fertilization

1. Distant interaction of gametes. Sperm and egg cells produce hormones.

Their joint action activates sperm and provides their movement to egg.

Capacitation means special activation of spermatozoa which occurs with them in female reproductive organs. Only after such changes they begin to move forward.

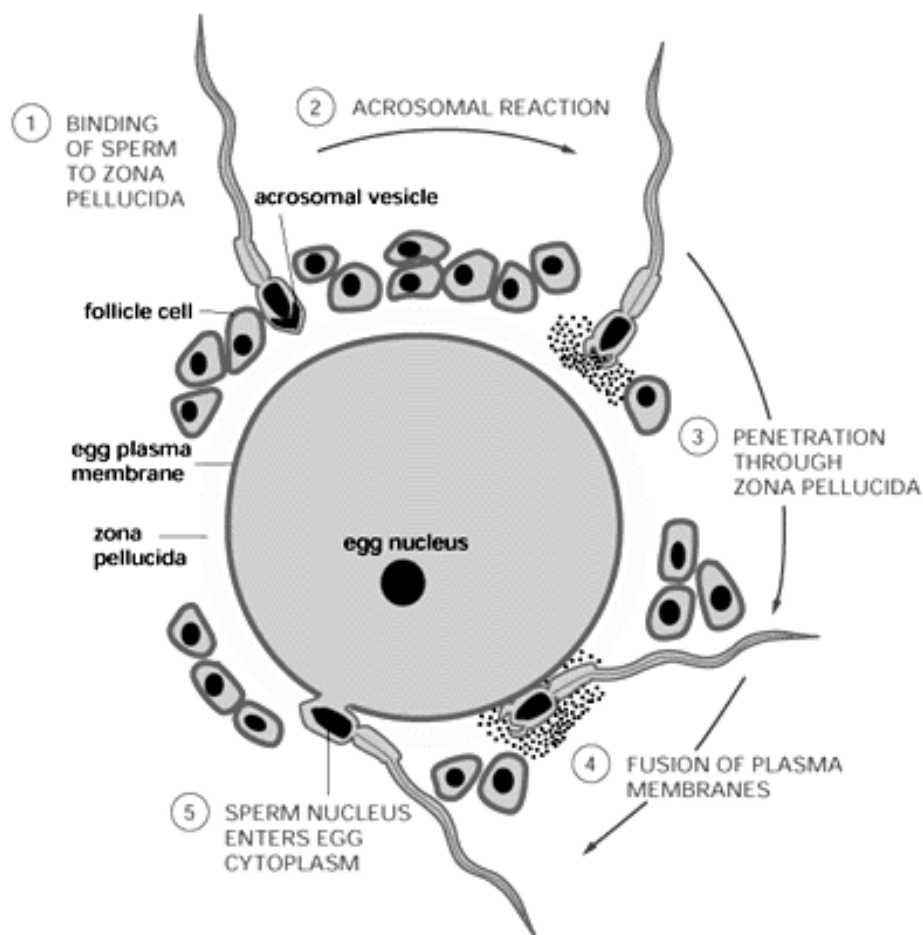
Taxis – active passage of spermatozoa. Chemotaxis (chemotropism) – toward to the oocyte, which produces special chemical substance – gamones. Rheotaxis – against the fluid flow in the uterine tube.

2. Contact interaction of gametes. This stage is characterized by the acrosomal reaction of the sperm and cortical reaction of the egg. As a result, the nucleus of sperm penetrates to the egg and the fertilization envelope which prevents the penetration of other sperm forms around the egg.

Contact stage of fertilization include penetrate of the corona radiata, penetrate of the zona pellucida, penetrate of the ovolemma, syngamy. In female this process takes place in the ampullar portion of the Fallopian (uterine) tube.

Penetrate of the corona radiata. 200-500 spermatozoa surround the egg and join into corona radiata. Due to the fact that spermatozoa tails move, the egg begins to rotate too and the corona radiata cells disperse.

Penetrate of the zona pellucida. Only one spermatozoa can penetrate of the zona pellucida. When this spermatozoa contacts with Zp3 glycoproteins of this zona, acrosomal reaction begins. Acrosomal reaction is release of acrosomal enzymes. This reaction allows spermatozoa to penetrate the zona pellucide and then to coming in contact with the plasma membrane of the oocyte.



Penetrate of the ovolemma. After adhesion, the plasma membranes of the sperm and egg is merge. The spermatozoa core, mitochondria, the part of acronema enter in the ovoplasm, but the spermatozoa plasma membrane is left behind on the

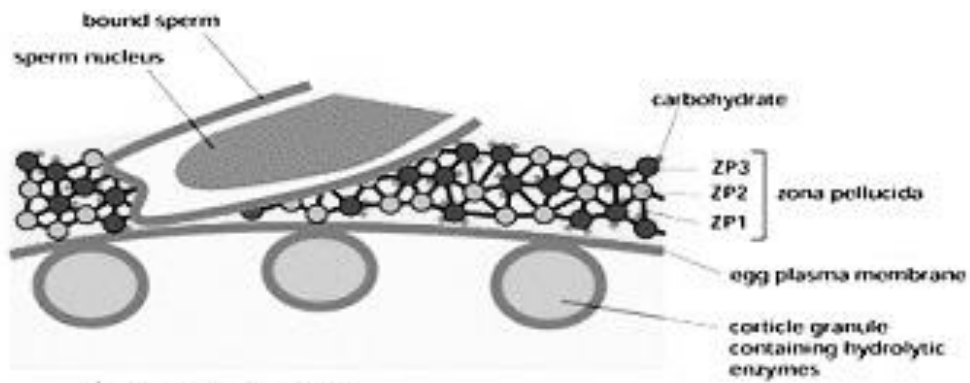
oocyte surface. As soon as the spermatozoa has entered the oocyte, the egg responds by two ways:

1. Cortical reactions. The spermatozoa plasma membrane modifies of cell membrane potential (minus to plus), so that there is releasing of the Ca^{++} from the depot to ovoplasm. Cortical granules are released by exocytosis. As a result of the release of cortical ovocyte granules, which contain lysosomal enzymes, the ovocyte membrane becomes impenetrable to other spermatozoa, and the zona pellucida changes its structure and composition for sperm binding and prevent penetration. These reactions prevent polyspermy (penetration of more than one spermatozoon into the oocyte). The zona pellucide becomes fertilization tunica. The space between the ovolemma and tunica of fertilisation is perivitellin space, in which stored water.

2. Resumption of the second meiotic division. The oocyte finishes its second meiotic division immediately after entry of the spermatozoon. One of the daughter cells, which receives hardly any cytoplasm, is known as the second polar body; the other daughter cell is the definitive oocyte. Its chromosomes ($22 + X$) arrange themselves in a vesicular nucleus known as the female pronucleus.

3. Formation synkaryon (*Syngamy*). The nuclei of the sperm and egg unite and form a diploid zygote.

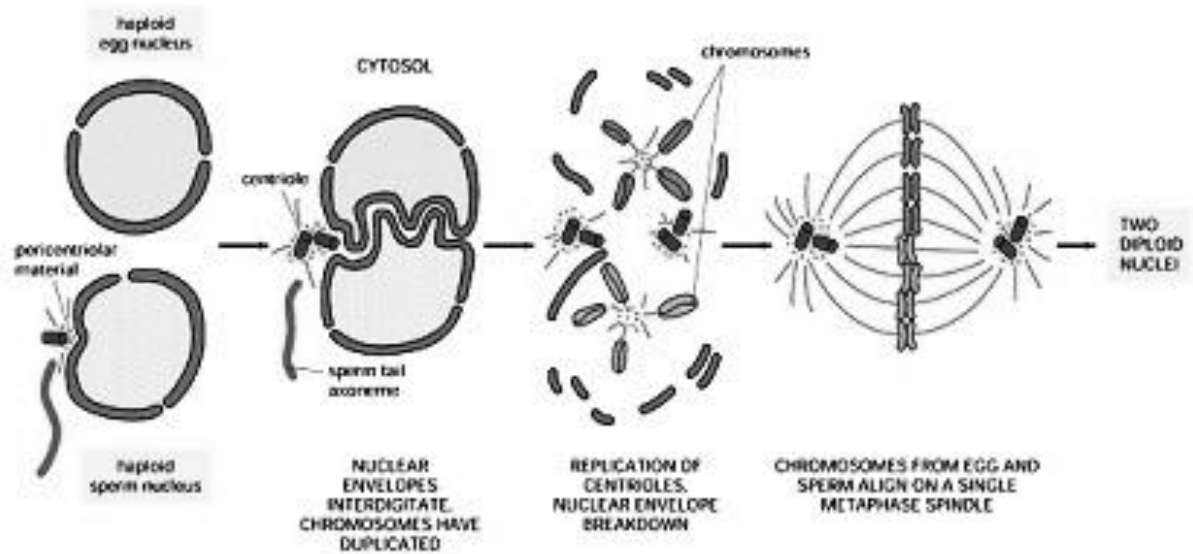
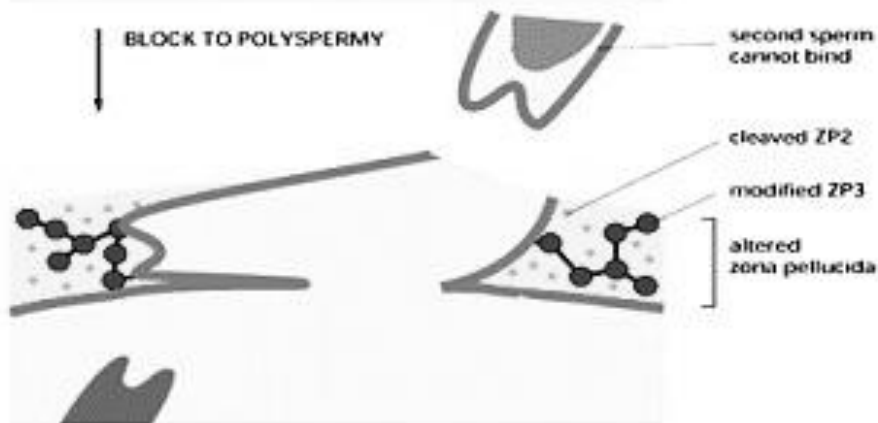
Syngamy – the process of nuclear fusion. The spermatozoa core (male pronucleus) unit with female pronucleus. At the moment of their fusion such unicellular organism is named “synkarion”. As the result a zygote is formed, which consists of 46 chromosomes.



CORTICAL REACTION
(EXOCYTOSIS)



BLOCK TO POLYSPERMY



CLEAVAGE

Cleavage (fissio) is the next process after fertilization – special mitotic division without growth of daughter cells. As a result multicellular organism - “blastula” - is developing. Blastula consists of: a wall - blastoderm consisting of individual cells - blastomeres and the cavity - blastocoel.

The pattern of cleavage is influenced by two factors: the amount of yolk and the formation of mitotic spindles

Cleavage process directly depends on the type of oocyte, his yolk inclusions volume and disposition.

Cleavage can be:

complete (holoblastic)

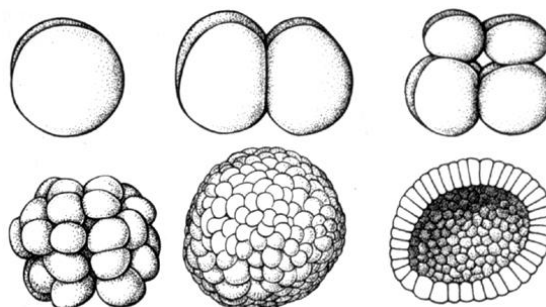
incomplete (meroblastic).

Holoblastic cleavage can be:

- a) uniform (if all blastomeres of equal size) and uneven (blastomeres of different sizes);
- b) synchronous (blastomeres divided at the same time) and asynchronous.

Meroblastic cleavage can be superficial and discoidal.

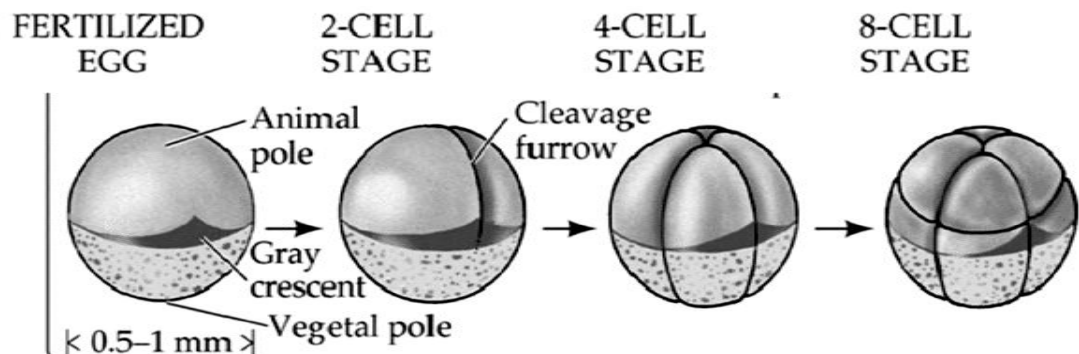
- Yolk is the nutrient material stored in an egg. Yolk impedes the formation of a cleavage furrow.



In embryos with little or no yolk, all daughter cells tend to be of similar size, as in the sea urchin.

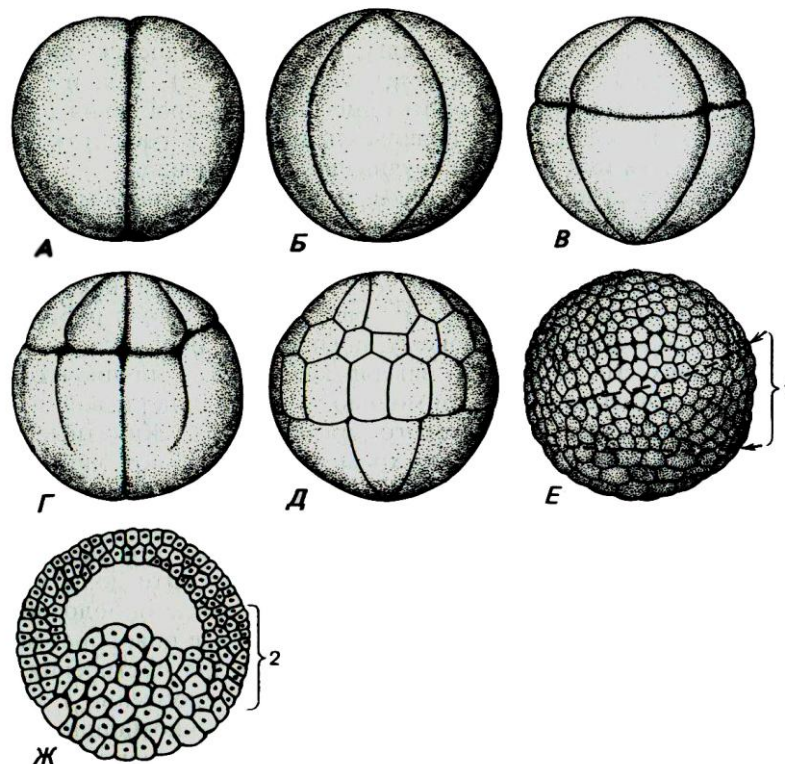
- When yolk quantity is larger, asymmetry of cell size is observed.

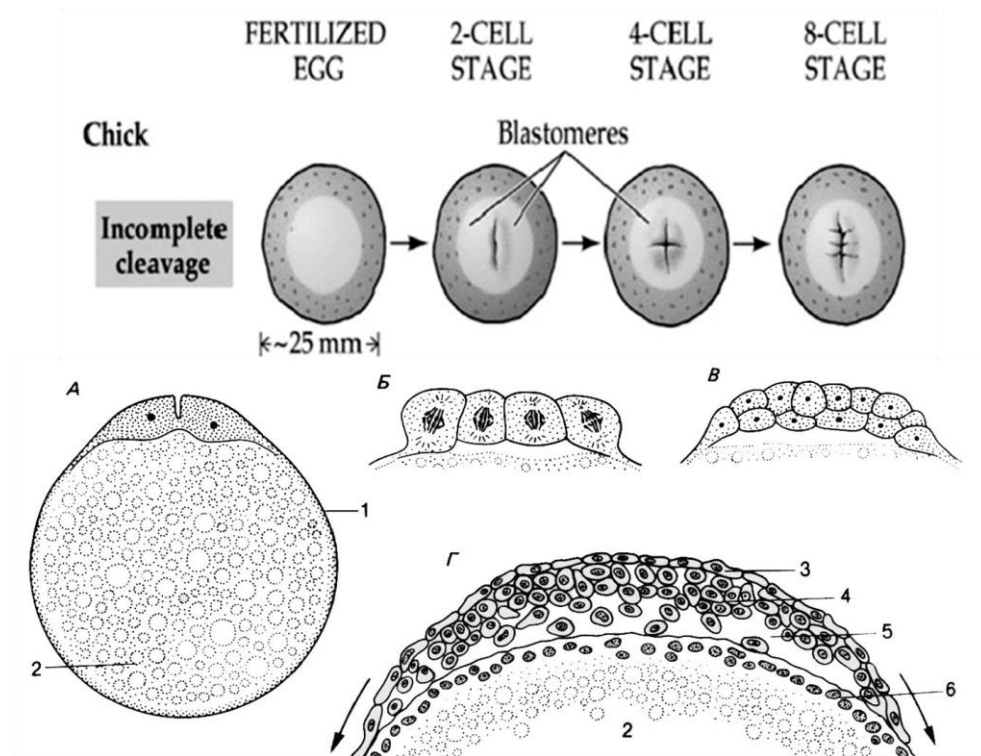
In the frog egg, the vegetal hemisphere ends up with fewer but larger cells than the animal hemisphere. Frogs have complete cleavage.



- In eggs with a lot of yolk, such as the chicken egg, cleavage is incomplete.

The cleavage furrows do not penetrate the yolk. The embryo forms a disc of cells, called the blastodisc, on top of the yolk. This type of incomplete cleavage is called discoidal cleavage and is common in birds, reptiles, and fish.

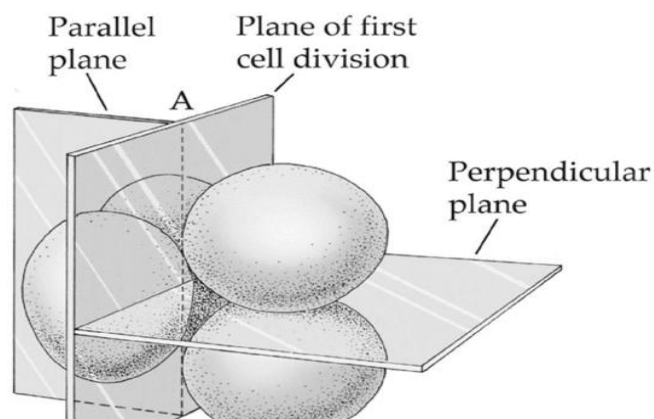




Orientation of the mitotic spindles determine the cleavage planes and arrangements of daughter cells.

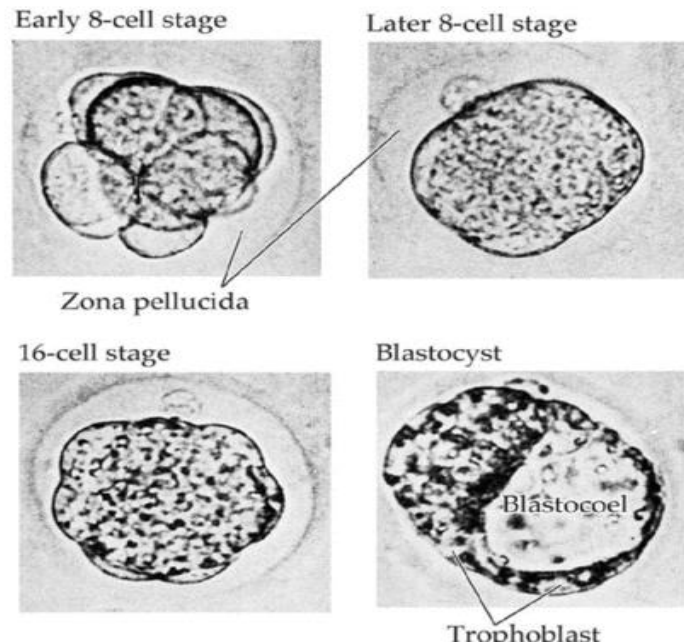
- If the mitotic spindles form at right angles or parallel to the animal-vegetal axis, aradial cleavage pattern results.
- If the mitotic spindles are at oblique angles to the animal-vegetal axis, the pattern has a twist, and is called spiral cleavage.
- In mammals, the first cell division is parallel to the animal-vegetal axis and the second cell division occurs at right angles.

The Mammalian Zygote Becomes a Blastocyst



This pattern of cleaves is referred to as rotational cleavage and is unique to mammals. Cleavage in mammals is slow, with divisions occurring 12 to 24 hours apart. The cell divisions are not synchronous, so the number of cells in the embryo does not follow the regular progression (2, 4, 8, 16, 32, etc.) typical of other species.

The Mammalian Zygote Becomes a Blastocyst



Unlike other animals, gene expression plays a role during mammalian cleavage.

At the 8-cell stage of a mammal embryo, the cells form tight junctions and a compact mass. At the transition from the 16-cell to 32-cell stage, the cells separate into two masses. The inner cell mass develops into the embryo; the outer cells become the trophoblast, which becomes part of the placenta. The trophoblast cells secrete fluid which forms the blastocoel. The embryo is called a blastocyst.

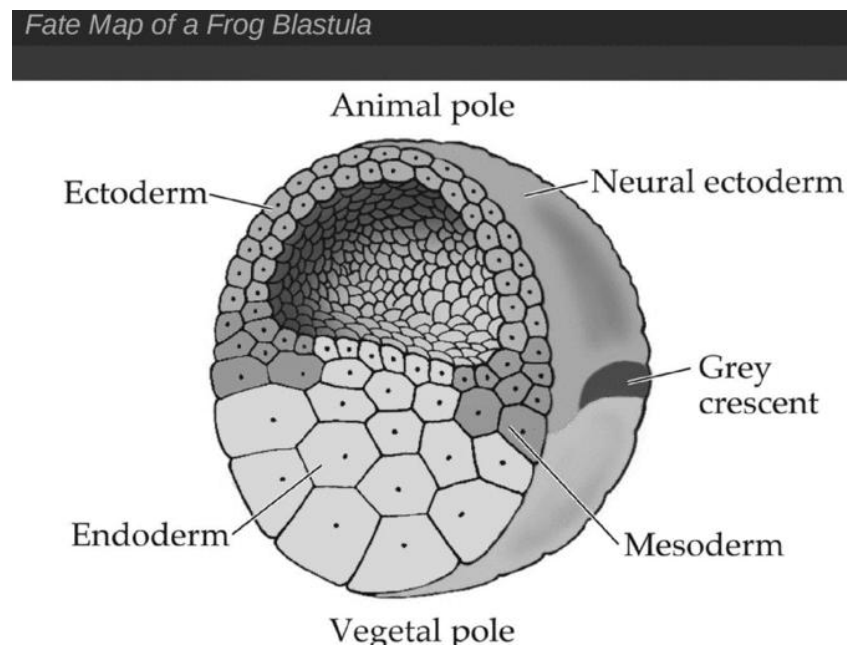
Fertilization in mammals occurs in the upper oviduct; cleavage occurs as the zygote travels down the oviduct.

When the blastocyst arrives in the uterus, the trophoblast adheres to the uterine wall (the endometrium), which begins the process of implantation.

Early implantation in the oviduct wall is prevented by the zona pellucida.

In the uterus, the blastocyst hatches out of the zona pellucida, and implantation can occur.

In all animals, cleavage results in the repackaging of the egg cytoplasm into the cells of the blastula. The cells get different amounts of nutrients and cytoplasmic determinants.



In the next stage, the cells of the blastula begin to move and differentiate.

The cells can be labeled with dyes to determine what tissues and organs develop from each. Fate maps of the blastula are the result.

Blastomeres become determined, or committed to a specific fate, at different times in different animals.

Roundworm and clam blastomeres are already determined at the 8-cell stage.

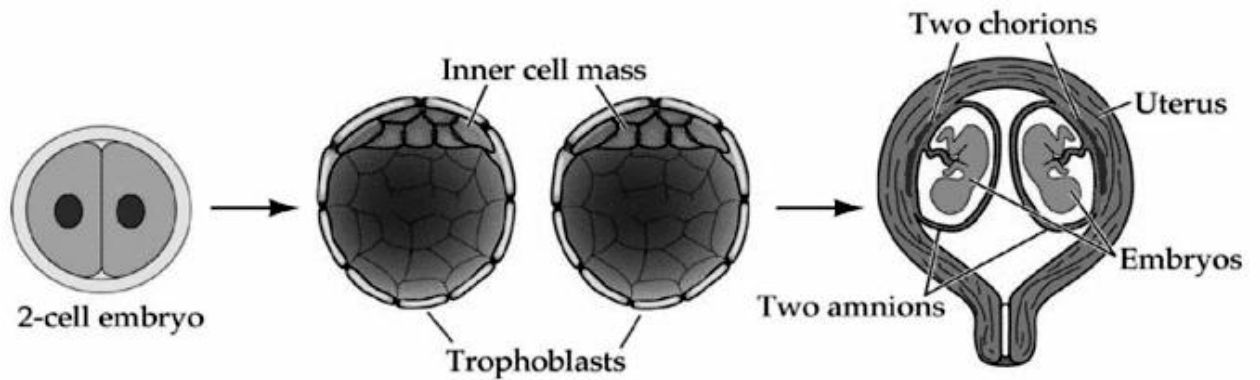
If one cell is removed, a portion of the-embryo fails to develop normally. This is called mosaic development.

Other animals have regulative development. If some cells are lost during cleavage, other cells can compensate.

If blastomeres are separated in an early stage, two embryos can result.

Since the two embryos came from the same zygote, they are monozygotic twins, or genetically identical identical twins.

Non-identical twins are the result of two separate eggs fertilized by two separate sperm and are not genetically identical.



GASTRULATION: PRODUCING THE BODY PLAN

Gastrulation is the process in which a blastula is transformed into an embryo with three tissue layers and body axes.

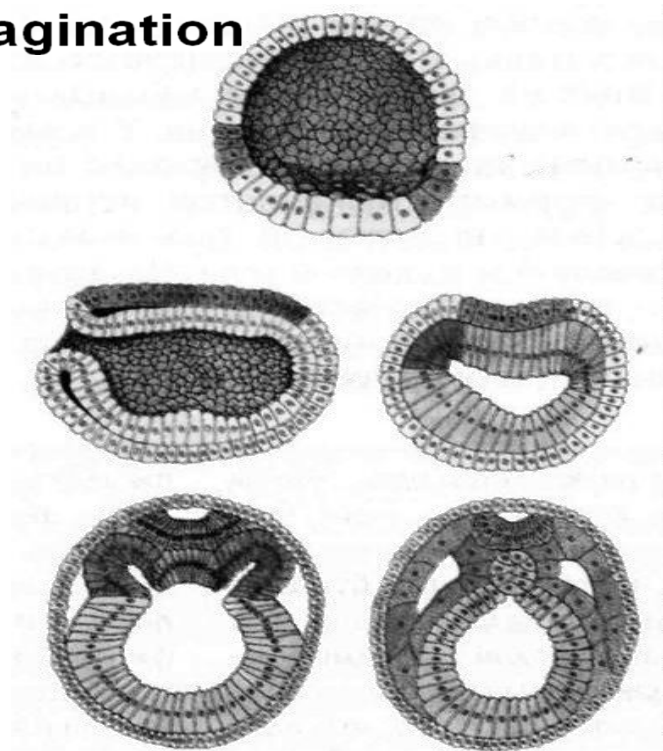
During gastrulation, three germ layers form:

- the inner germ layer is the endoderm and gives rise to the digestive tract, circulatory tract, and respiratory tract.
- the outer germ layer is the ectoderm and gives rise to the epidermis and nervous system.
- the middle germ layer, the mesoderm, contributes to formation of bone, muscle, heart and blood vessels.

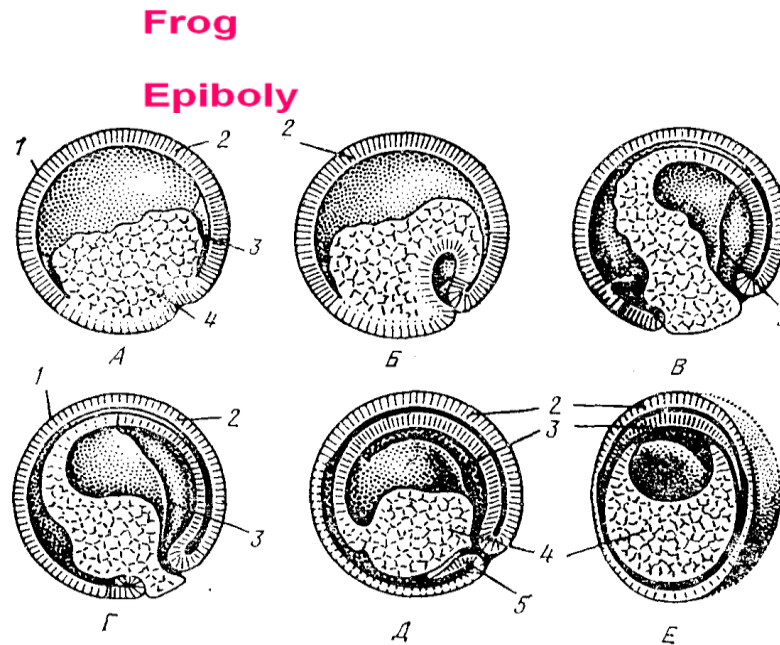
Ways of early gastrulation:

Invagination - blastoderm located on the vegetal pole inside blastocoel and forming the inner layer - the endoderm and the outer layer - the ectoderm.

Invagination



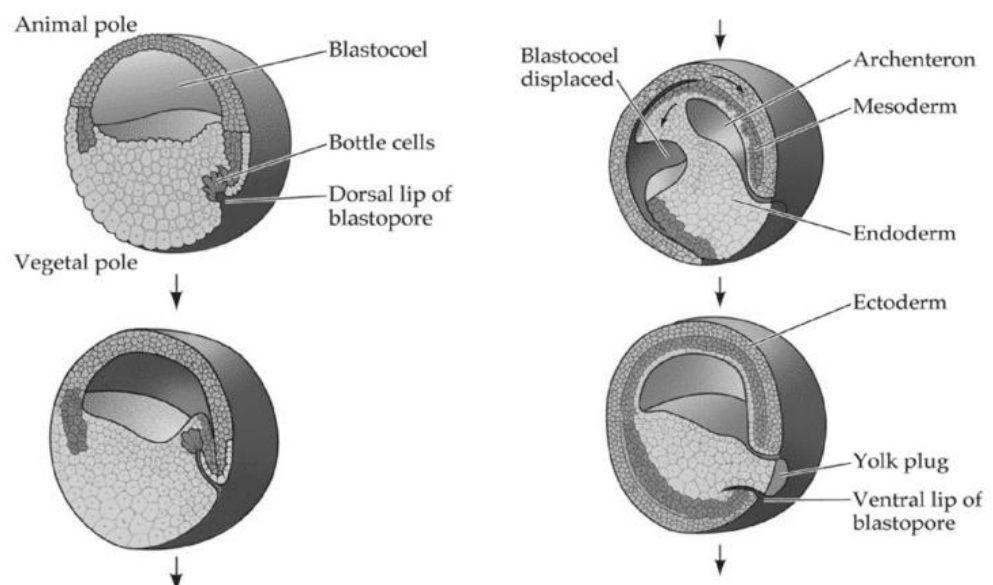
Epiboly - micromeres of animal pole of blastula divide faster macromeres of vegetal pole and overgrown them, forming ectoderm.



Immigration - some blastomeres of the blastula migrate into the cavity and form the endoderm.

Delamination - blastoderm stratified into two pieces: the inner - endoderm and the outer - ectoderm.

Gastrulation in the Frog Embryo



Mammal eggs have no yolk. The inner cell mass of the blastocyst splits into an epiblast and hypoblast with a fluid-filled cavity in between.

The embryo forms from the epiblast; the extraembryonic membranes form from the hypoblast. The epiblast also splits off a layer of cells that form the amnion. The amnion grows around the developing embryo. Gastrulation is similar to that in birds; a primitive groove forms and cells migrate through it to become endoderm and mesoderm.

NEURULATION: INITIATING THE NERVOUS SYSTEM

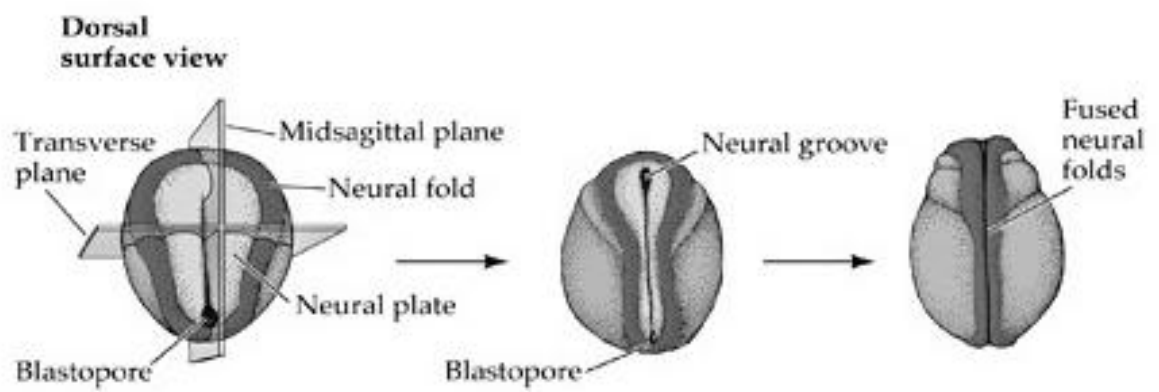
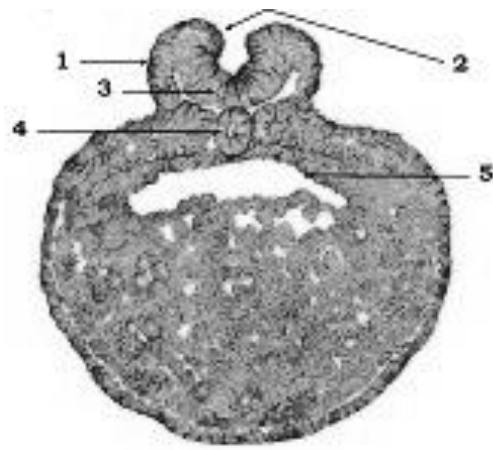
Gastrulation produces an embryo with three germ layers.

Organogenesis occurs next and involves the formation of organs and organ systems.

Neurulation occurs early in organogenesis and begins the formation of the nervous system in vertebrates.

The first cells to pass over the dorsal lip become the endodermal lining of the digestive tract. The second group of cells become mesoderm. The dorsal mesoderm closest to the midline (chordomesoderm) becomes the notochord. The notochord is connective tissue and is eventually replaced by the vertebral column. The chordomesoderm induces the overlying ectoderm to begin forming the nervous system.

Neurulation begins with thickening of the ectoderm above the notochord to form the neural plate. Edges of the neural plate thicken to form ridges. Between the ridges a groove forms and deepens. The ridges fuse, forming a cylinder—the neural tube. The anterior end of the neural tube becomes the brain.



Neurulation in the Frog Embryo

Midsagittal section

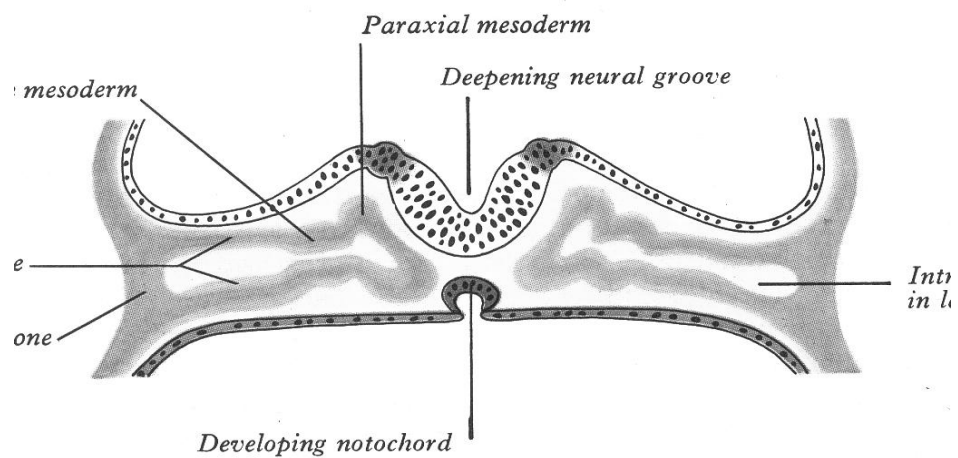
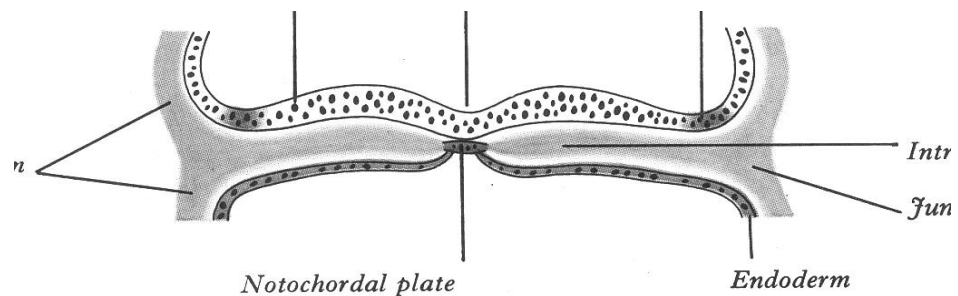
The diagram shows the progression of neurulation in three stages:

- Stage 1:** The neural plate is visible as a flat, dark region. Labels include: Notochord, Neural plate, Blastopore, Ectoderm, Archenteron, and Neural fold.
- Stage 2:** The neural folds are beginning to curve upwards. Labels include: Neural fold and Cavity of gut.
- Stage 3:** The neural tube is fully formed, a hollow tube of neural tissue. Label: Neural tube.

Transverse section

The diagram illustrates the three stages of neurulation in a transverse section:

- Stage 1:** The neural plate is formed from the ectoderm. Labels include: Notochord, Neural plate, Neural fold, Endoderm, Mesoderm, Archenteron, and Ectoderm.
- Stage 2:** The neural plate folds to form a neural groove. Labels include: Neural groove, Neural plate, and Cavity of gut.
- Stage 3:** The neural folds fuse to form the neural tube. Label: Neural tube.

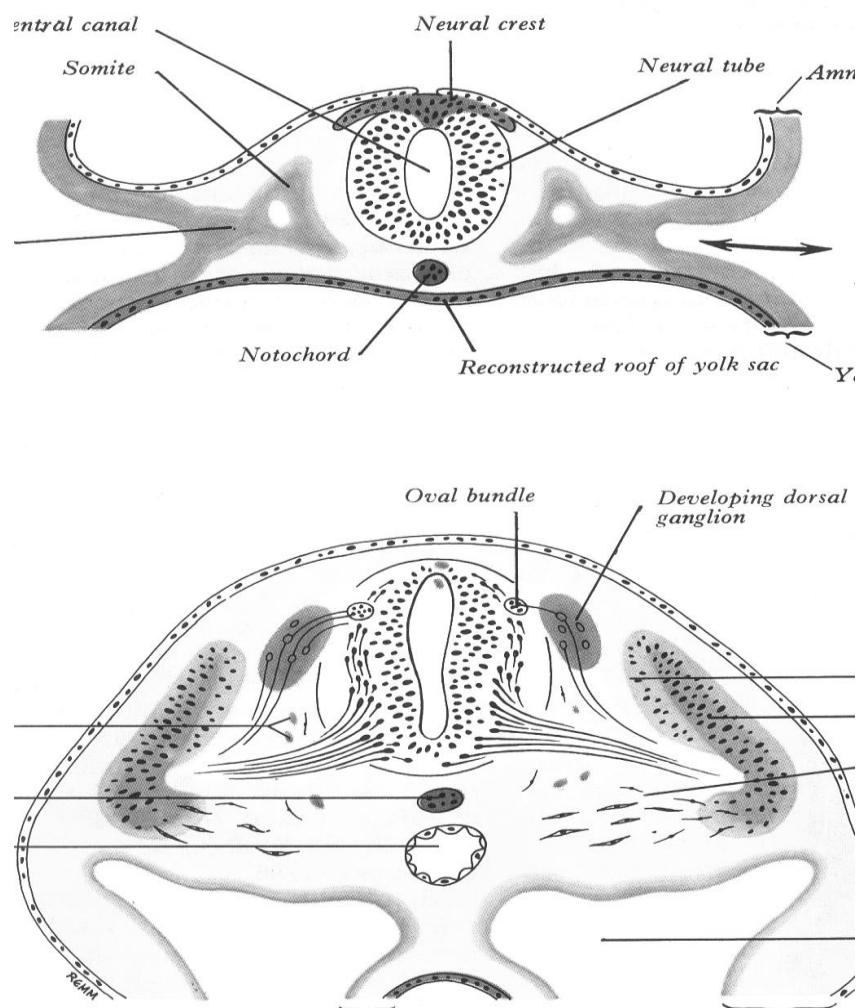


In humans, failure of the neural tube to close completely at the posterior end results in spina bifida. If the tube fails to close at the anterior end, the result is anencephaly, in which the forebrain does not develop. Neural tube defects can be reduced if pregnant women receive adequate folic acid (a B vitamin).

Body segmentation develops during neurulation. Blocks of mesoderm called somites form on both sides of the neural tube. Somites produce cells that form the vertebrae, ribs, and muscles of the trunk and limbs. They also guide the organization of the peripheral nerves.

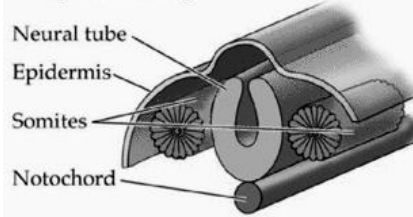
When the neural tube closes, cells called neural crest cells break loose; they migrate inward between the epidermis and the somites and under the somites.

The neural crest cells give rise to many structures and organs of the body.

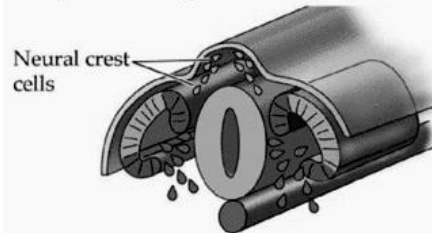


The Development of Body Segmentation

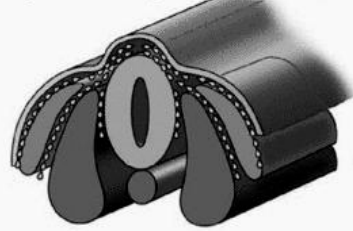
2-Day chick embryo



4-Day chick embryo



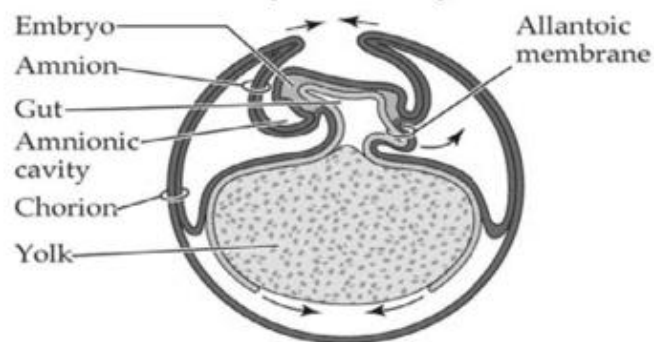
7-Day chick embryo



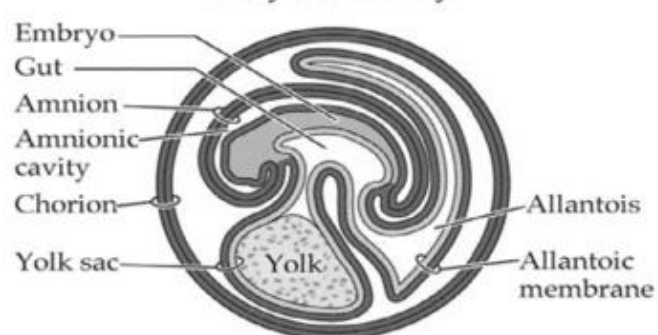
EXTRAEMBRYONIC MEMBRANES

Extraembryonic membranes originate from the germ layers of the embryo and function in nutrition, gas exchange, and waste removal.

5-Day chick embryo



9-Day chick embryo



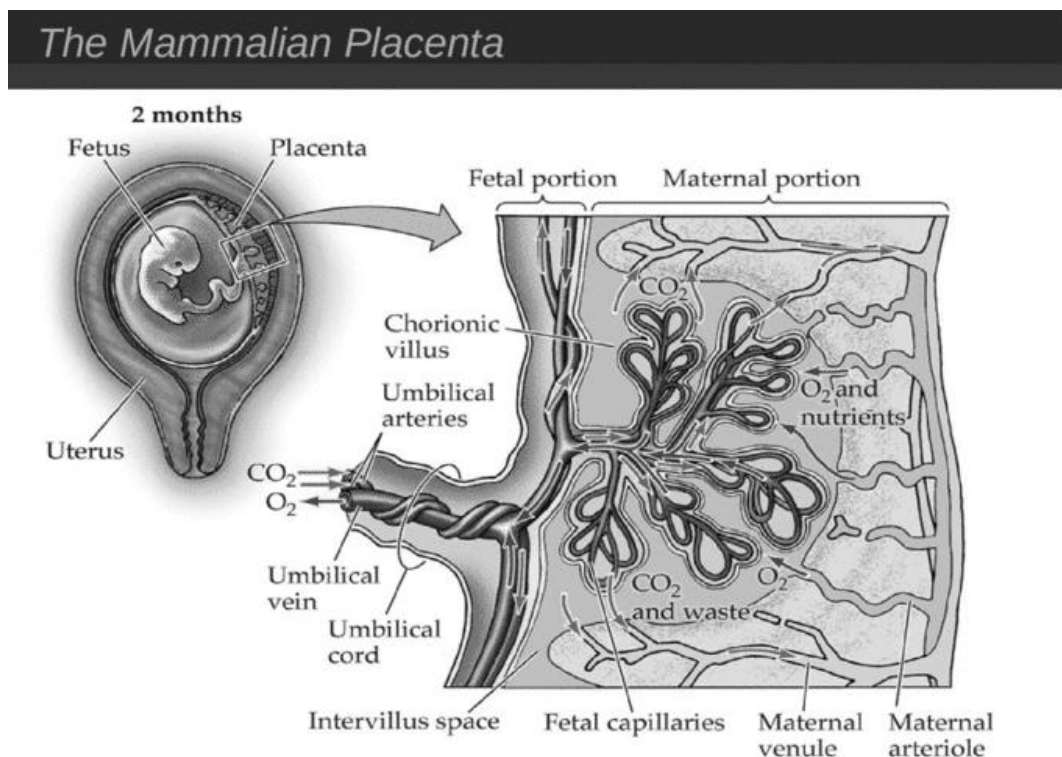
In the chicken, the yolk sac is the first to form, by extension of the endodermal tissue of the hypoblast. It constricts at the top to create a tube that is continuous with the gut of the embryo.

Yolk is digested by the endodermal cells of the yolk sac, and the nutrients are transported through blood vessels lining the outer surface of the yolk sac.

The allantoic membrane, an outgrowth of the extraembryonic ectoderm, forms the allantois, a sac for storage of metabolic wastes.

Ectoderm and mesoderm combine and extend beyond the embryo to form the amnion and the chorion. The amnion surrounds the embryo, forming a cavity. The amnion secretes fluid into the cavity that provides protection for the embryo. The chorion forms a continuous membrane just under the eggshell. It limits water loss.

In mammals, the first extraembryonic membrane to form is the trophoblast. When the blastocyst hatches from the zona pellucida, the trophoblast cells attach to the uterine wall. This is the beginning of implantation. The trophoblast becomes part of the uterine wall, and sends out villi to increase surface area and contact with maternal blood.



The hypoblast cells extend to form the chorion. The chorion and other tissues produce the placenta. The epiblast produces the amnion. Allantoic tissues from the umbilical cord.

Cells from the embryo that are in the amniotic fluid can be sampled and tested for effects. The test is called amniocentesis.

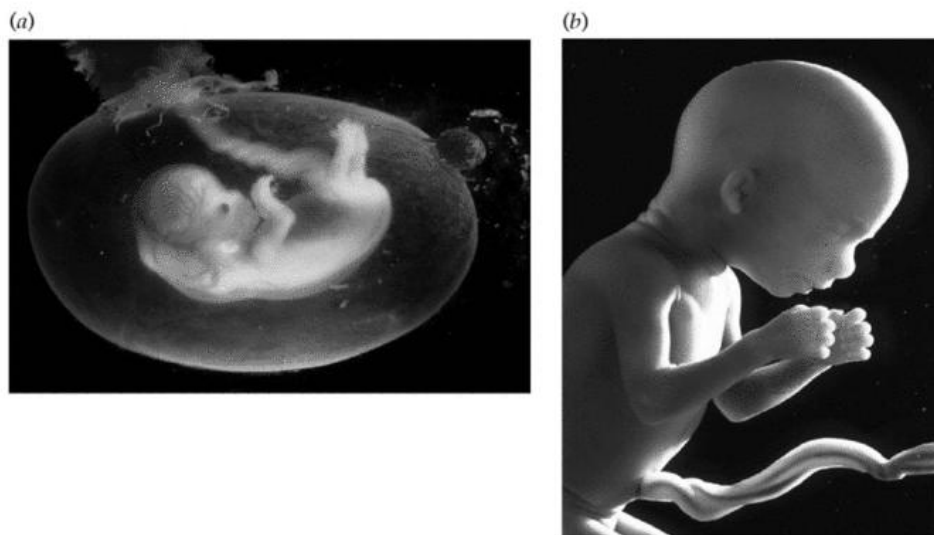
Problems such as Down syndrome, cystic fibrosis, and Tay Sachs disease can be detected using this technique.

A newer technique is chorionic villus sampling which makes earlier detection possible.

HUMAN DEVELOPMENT

The events of human gestation (pregnancy) are divided into three trimesters. The first trimester begins with fertilization. Implantation takes place 6 days later.

Stages of Human Development



Then gastrulation takes place, the placenta forms, and tissues and organs begin to form. The heart first beats at 4 weeks and limbs form at 8 weeks. The embryo is particularly vulnerable to radiation, drugs, and chemicals during the first

trimester. Hormonal changes can cause major responses in the mother, including morning sickness.

During the second trimester the fetus grows rapidly to about 600g. Fingers, toes, and facial features become well formed. Fetal movements are first felt by the mother early in the second trimester. By the end of the second trimester, the fetus may suck its thumb.

The fetus and the mother continue to grow during the third trimester. Throughout the third trimester, the fetus remains susceptible to environmental factors such as malnutrition, alcohol consumption, and cigarette smoking. Kidneys produce urine, the liver stores glycogen, and the brain undergoes cycles of sleep and waking.

Development does not end at birth. The organization of the nervous system exhibits a great deal of plasticity in the early years, as patterns of connections between neurons develop. For example, a child born with misaligned eyes will use mostly one eye. The connections to the brain from this eye will become stronger, while the connections to the other eye will become weak. This can be changed if the alignment is corrected within the first three years. A current area of research into this developmental plasticity in the nervous system examines the role of learning in stimulating the production and differentiation of new neurons in the brain.

Testing your knowledge

ONTOGENESIS

1. During spermatogenesis in stage of growth cells called
 - a) spermatogonia
 - b) primary spermatocytes
 - c) secondary spermatocytes
 - d) spermatids

2. During spermatogenesis in stage of reproduction cells called
 - a) spermatogonia
 - b) primary spermatocytes
 - c) secondary spermatocytes
 - d) spermatids
3. During oogenesis in stage of growth cells called
 - a) oogonia
 - b) primary oocyte
 - c) polar bodies
 - d) secondary oocyte
4. In stage of maturation during first meiosis primary oocyte is divided into two cells:
 - a) oogonia and primary oocyte
 - b) primary oocyte and secondary oocyte
 - c) primary oocyte and polar bodies
 - d) secondary oocyte and polar bodies
5. In stage of maturation during second meiosis secondary spermatocyte is divided into two cells:
 - a) two spermatogonia
 - b) two primary spermatocytes
 - c) two sperm
 - d) two spermatids
6. The acrosomal reaction of the sperm occurs during stage of
 - a) gametogenesis
 - b) contact interaction of gametes
 - c) distant interaction of gametes
 - d) formation synkaryon
7. Each spermatozoon has
 - a) head, body, tail

b) head, body, leg

c) neck, body, tail

d) head, neck, tail

8. Nucleus of the ovum contains 23 chromosomes:

a) 22 autosomes and the last one – sex chromosome (only y).

b) 22 autosomes and the last one – sex chromosome (only x).

c) 21 autosomes and two – sex chromosome (xx).

d) 21 autosomes and two – sex chromosome (xy).

9. Oligolecytral oocyte

a) contain moderate yolk inclusions

b) contain great volume of yolk

c) not contain egg yolk

d) contain a lot of carbohydrates

10. The embryo is formed by the cleavage

a) blastula

b) gastrula

c) neurula

d) zygote

11. The embryo is formed by the gastrulation

a) blastula

b) gastrula

c) neurula

d) zygote

12. The embryo is formed by the fertilization

a) blastula

b) gastrula

c) neurula

d) zygote

13. Blastula consists of:

- a) blastoderm and blastocoel
- b) head, body, tail
- c) blastoderm and blastopore
- d) ectoderm and endoderm

14. Cleavage is special mitotic division of

- a) gastrula without growth of daughter cells
- b) zygote without growth of daughter cells
- c) zygote with growth of daughter cells
- d) neurula without growth of daughter cells

15. Ways of early gastrulation

- a) fertilization, epiboly, immigration, delamination
- b) invagination, epiboly, immigration, delamination
- c) fertilization, epiboly, gametogenesis, delamination
- d) gametogenesis, epiboly, immigration, delamination

16. Micromeres of animal pole of blastula divide faster macromeres of vegetal pole and overgrown them, forming ectoderm during

- a) invagination,
- b) epiboly,
- c) immigration,
- d) delamination

17. Blastoderm stratified into two pieces: the inner - endoderm and the outer – ectoderm during

- a) invagination,
- b) epiboly,
- c) immigration,
- d) delamination

18. Blastoderm located on the vegetal pole inside blastocoel and forming the inner layer - the endoderm and the outer layer - the ectoderm during

- a) invagination,
- b) epiboly,
- c) immigration,
- d) delamination

19. Some blastomers of the blastula migrate into the cavity and form the endoderm during

- a) invagination,
- b) epiboly,
- c) immigration,
- d) delamination

20. Ectoderm leads to the formation of

- a) nervous system,
- b) circulatory system,
- c) digestive system,
- d) muscular system

21. Entoderm leads to the formation of

- a) nervous system,
- b) circulatory system,
- c) digestive system,
- d) muscular system

22. Mesoderm leads to the formation of

- a) nervous system,
- b) respiratory system,
- c) digestive system,
- d) muscular system

23. The chorion is formed from

- a) only ectoderm
- b) ectoderm, mesoderm
- c) mesoderm and entoderm

d) only entoderm

24. The amnion is formed from

a) only ectoderm

b) ectoderm, mesoderm

c) mesoderm and entoderm

d) only entoderm

25. The allantois is formed from

a) only ectoderm

b) ectoderm, mesoderm

c) mesoderm and entoderm

d) only entoderm

UNIT 4. GENETICS

HEREDITY

Genetics - is the scientific study of heredity and variation.

Heredity - is the transmission of traits from one generation to the next.

You must know follow terms:

Gene is a locus (or region) of DNA that encodes a functional RNA or protein product, and is the molecular unit of heredity. We have two copies of each gene that we inherited from our mother and our father.

Locus is the location of a gene on a chromosome.

Allele is one of two or more alternative forms of a gene (A or a).

Dominant allele- an allele that is fully expressed in the phenotype of a heterozygous (A). For example, the allele for brown eyes is dominant, therefore you only need one copy of the 'brown eye'.

Recessive alleles is an allele whose phenotypic effect is not observed in a heterozygous by the presence of a dominant allele. For example, the allele for blue eyes is recessive, therefore to have blue eyes you need to have two copies of the 'blue eye' allele (a).

Homologous chromosomes is a chromosome pairs of the same length, centomere position, and staining pattern that possess genes for the same characters at corresponding loci. One homologous chromosome is inherited from the organisms father. The other from mother.

Homozygous is a individual carrying identical alleles of a gene on both homologous chromosomes (AA , bb).

Heterozygous is a individual carrying two different alleles of a gene on two homologous chromosomes (Aa , Bb). Most human beings are heterozygous for many genes.

Monohybrid cross is a breeding experiment between parental generation (P generation) organisms that differ in one trait ($AA \times aa$).

Dihybrid cross is a breeding experiment between P generation organisms that differ in two traits ($AABB \times aabb$).

Genotype is the genetic makeup of an organism/

Phenotype is the physical and physiological traits of an organisms, which determined by its genetic makeup.

GREGOR MENDEL IS FOUNDATION OF MODERN GENETICS



Mendel was born into a very poor family. To the boy was educated, his father sends him to the monastery. Johann was ordained and takes its name Gregor.

Studied segregation of traits in the garden pea (*Pisum sativum*) beginning in 1854. Published his theory of inheritance in 1865 “Experiments in Plant Hybridization”. Mendel spent his spare time breeding pea plants.

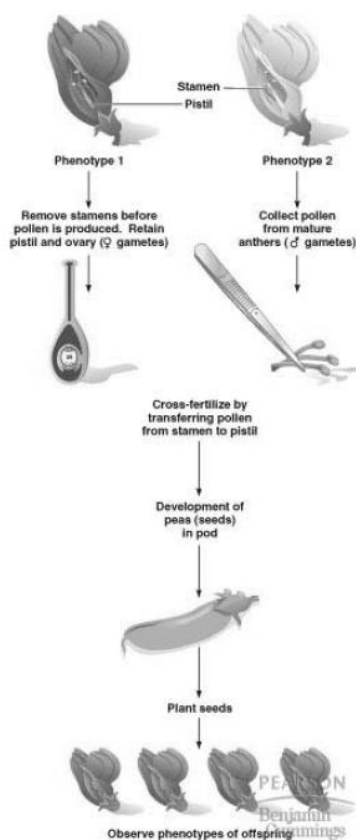
He did this over & over & over again, and noticed patterns to the inheritance of traits, from one set of pea plants to the next. By carefully analyzing his pea plant numbers, he discovered three laws of inheritance.

In his work, the words "[chromosomes](#)" or "genes" are nowhere to be found. The role of these things in relation to inheritance & heredity had not been discovered yet. What makes Mendel's contributions so impressive is that he described the basic patterns of inheritance before the mechanism for inheritance (namely genes) was even discovered

In a typical breeding experiment Mendel mated two contrasting, true-breeding varieties, a process called hybridization

The true-breeding parents are called the P generation. The hybrid offspring of the P generation are called the F1 generation. When F1 individuals self-pollinate the F2 generation is produced. A cross between plants that exhibit differences in one distinct trait (seed color, seed shape, flower color, etc), i.e. plants that produce yellow peas x plants that produce green peas.

Yellow was dominant to green.



Garden pea as experimental organism

General features of model organisms:

- short life cycle
- large number of offspring
- easy (inexpensive) to handle
- sufficient variation
- known genetic history

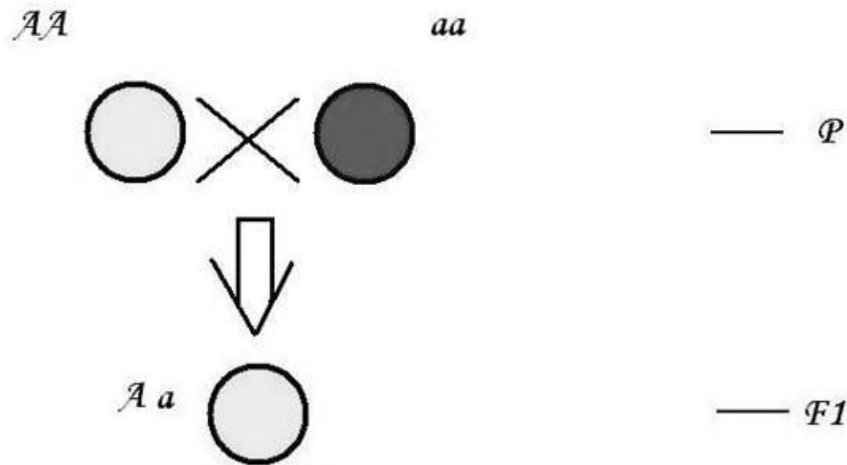


Specific feature of garden pea:

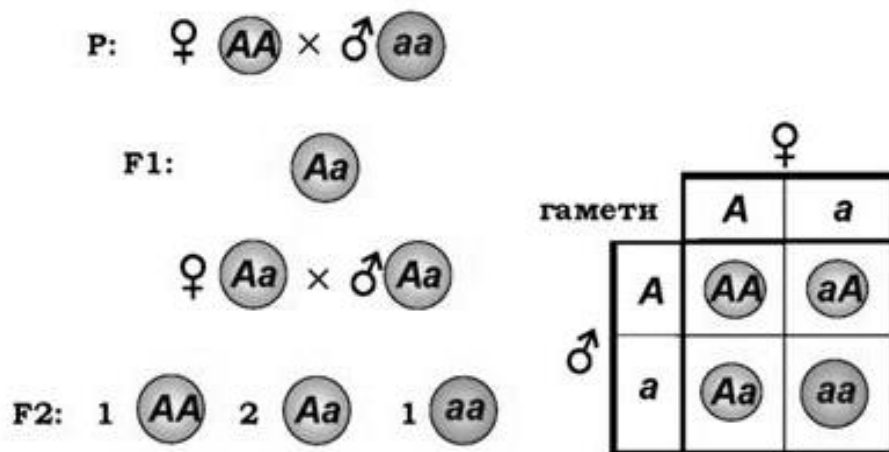
- ❖ can be cross-pollinated (crossed)
- ❖ can be self-pollinated (selfed)

Mendel formulated the **law of dominance (uniformity of hybrids of the first generation)**:

In a cross of parents that are pure for contrasting traits, only one form of the trait will appear in the next generation. Offspring that are hybrid for a trait will have only the dominant trait in the phenotype.



When Mendel crossed the yellow-peas F1 plants, $\frac{3}{4}$ of the plants had yellow peas, but $\frac{1}{4}$ had green peas.



A ratio of about three to one, yellow to green flowers, in the F2 generation.

The **Punnett Square** allows us to visualize a cross by examining the possible combinations of gametes from the parents.

Mendel formulated the **law of segregation**:

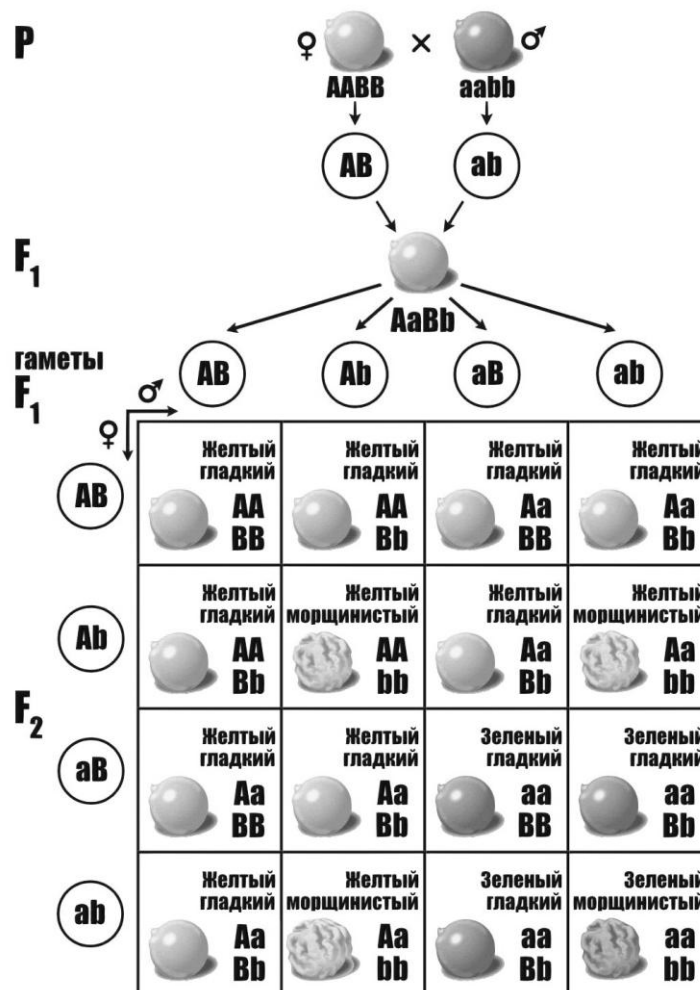
Genes have alternative forms, or alleles. In a diploid organism, the two alleles of a gene segregate (separate) during meiosis and gamete formation; each sperm or egg carries only one allele of each pair.

This law explains the 3:1 ratio of F₂ phenotypes observed when monohybrids self-pollinate.

Mendel also crossed plants that differed in two and more characteristics, such as color and form peas

In a cross between **dihybrids (individuals heterozygous for two genes)**, **the offspring** have four phenotypes in a 9:3:3:1 ratio.

The 9:3:3:1 ratio observed derives from two separate 3:1 phenotypic ratios: the ratio of yellow to green is 12:4 (or 3:1) and of round to wrinkled is 12:4 (or 3:1).

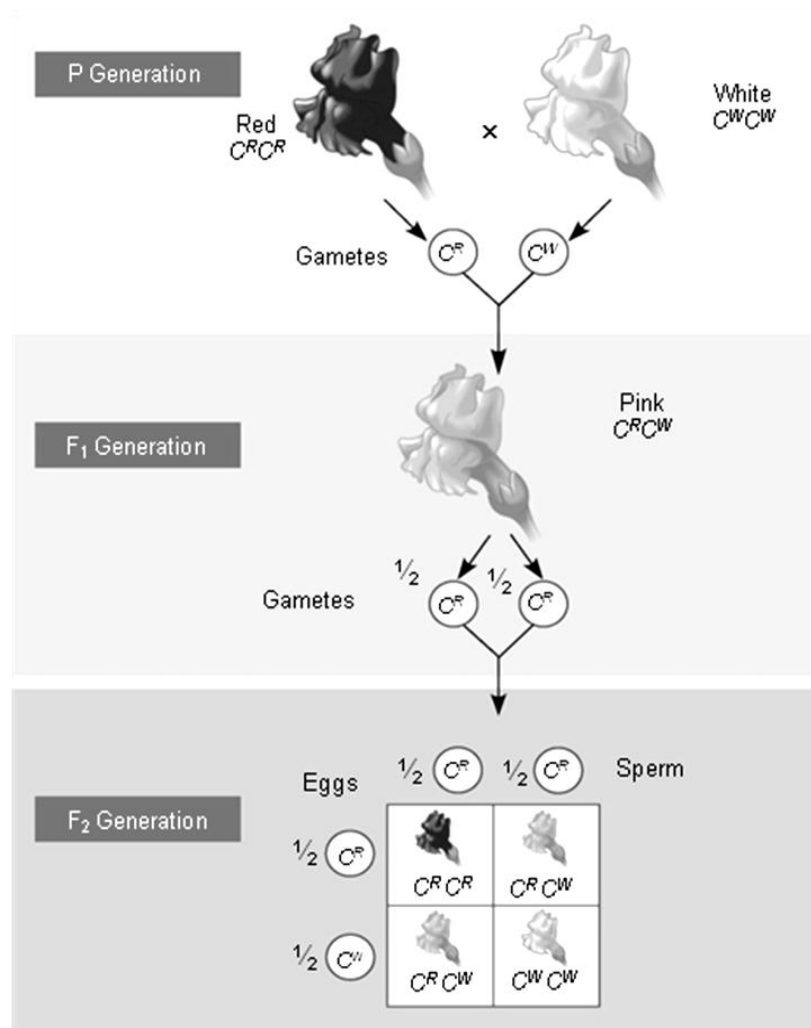


The pair of alleles for a given gene segregates into gametes independently of the pair of alleles for any other gene.

THE INTERACTION OF GENES

Complete dominance. Neither allele is dominant and heterozygous individuals have an dominant phenotype.

Incomplete dominance. Neither allele is dominant and heterozygous individuals have an intermediate phenotype. For example, in Japanese “Four o’clock”, plants with one red allele and one white allele have pink flowers:



Codominant

Neither allele is dominant and both alleles are expressed in heterozygous individuals. Example ABO blood types

ABO Blood groups





–3 alleles

–6 possible ABO genotypes: $I^A I^A$, $I^B I^B$, $I^A I^B$, $I^A i$, $I^B i$, or ii

4 possible phenotypes.

This example demonstrates the manifestation of multiple allelism: A gene can be represented by three alleles.

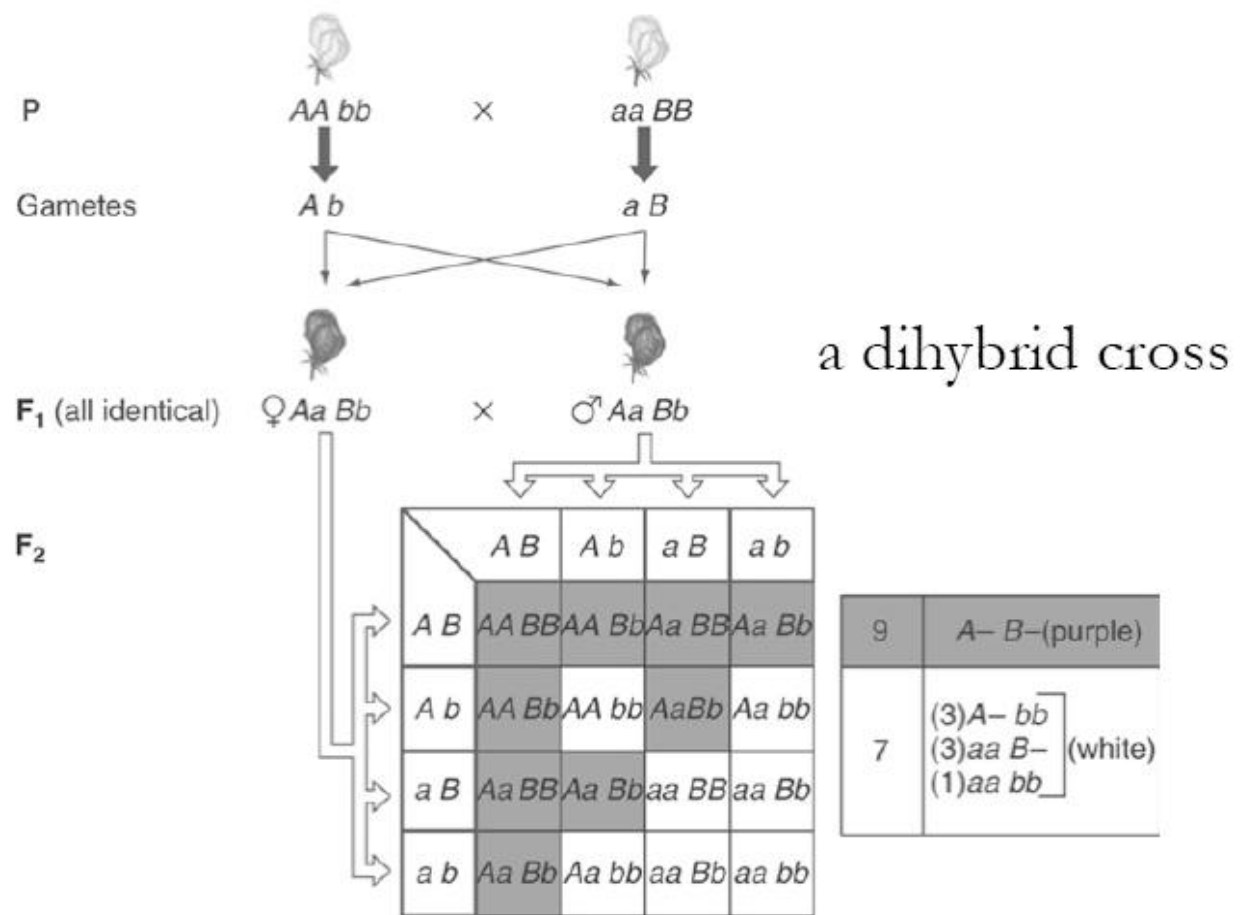
Determination of ABO blood group by multiple alleles

Genotype	Phenotype (Blood Group)	Red Blood Cells
$I^A I^A$ or $I^A i$	A	
$I^B I^B$ or $I^B i$	B	
$I^A I^B$	AB	
ii	O	

Complementation

Two genes work together to produce a phenotype.

- In a dihybrid cross, 9:7 ratio is a phenotypic signature of complementary gene interaction where dominant alleles of two genes act together to produce a trait while other three genotypic classes do not.

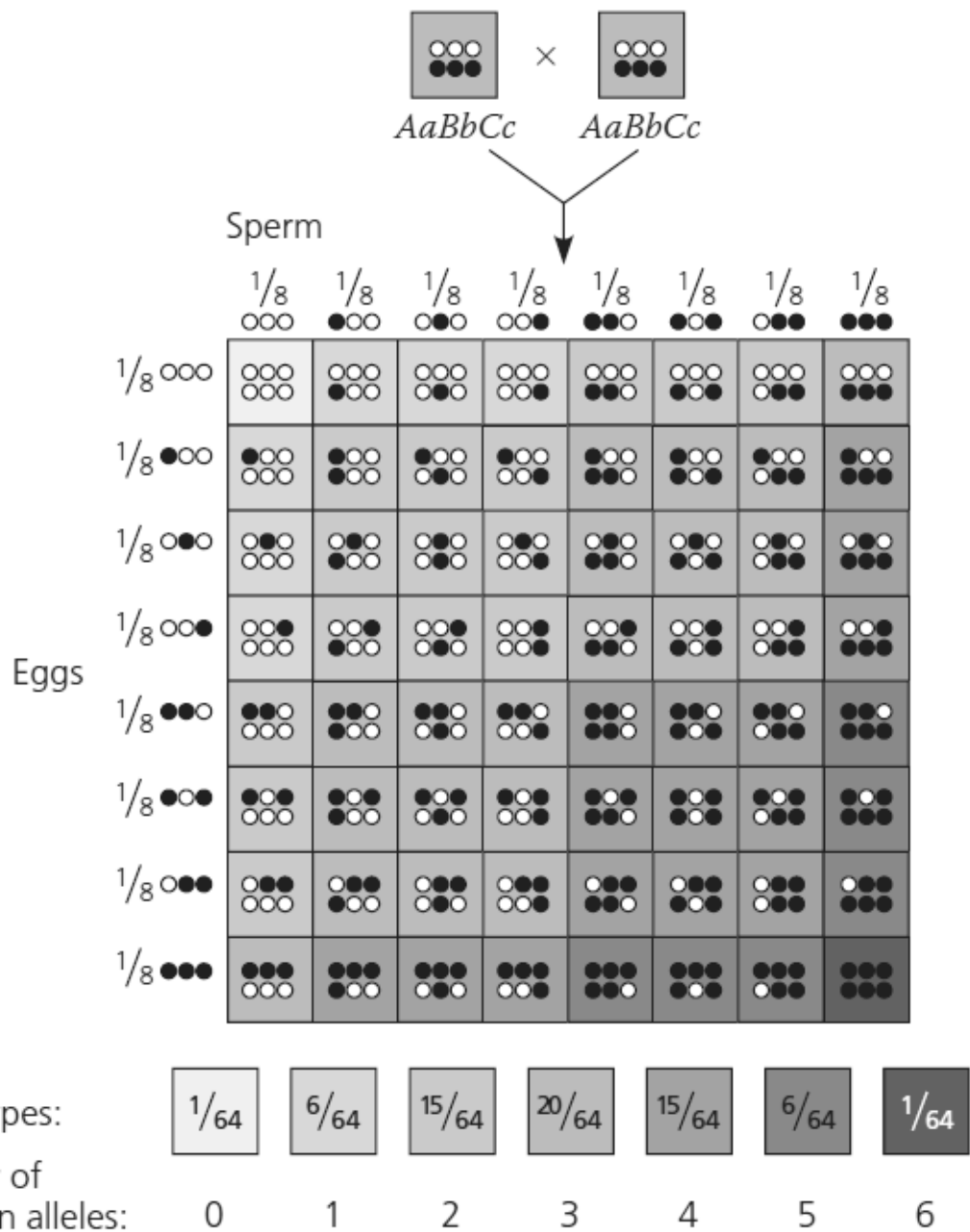


Polygenic Traits

Most traits are not controlled by a single gene locus, but by the combined interaction of many gene loci. These are called polygenic traits.

Polygenic traits often show continuous variation, rather than a few discrete forms.

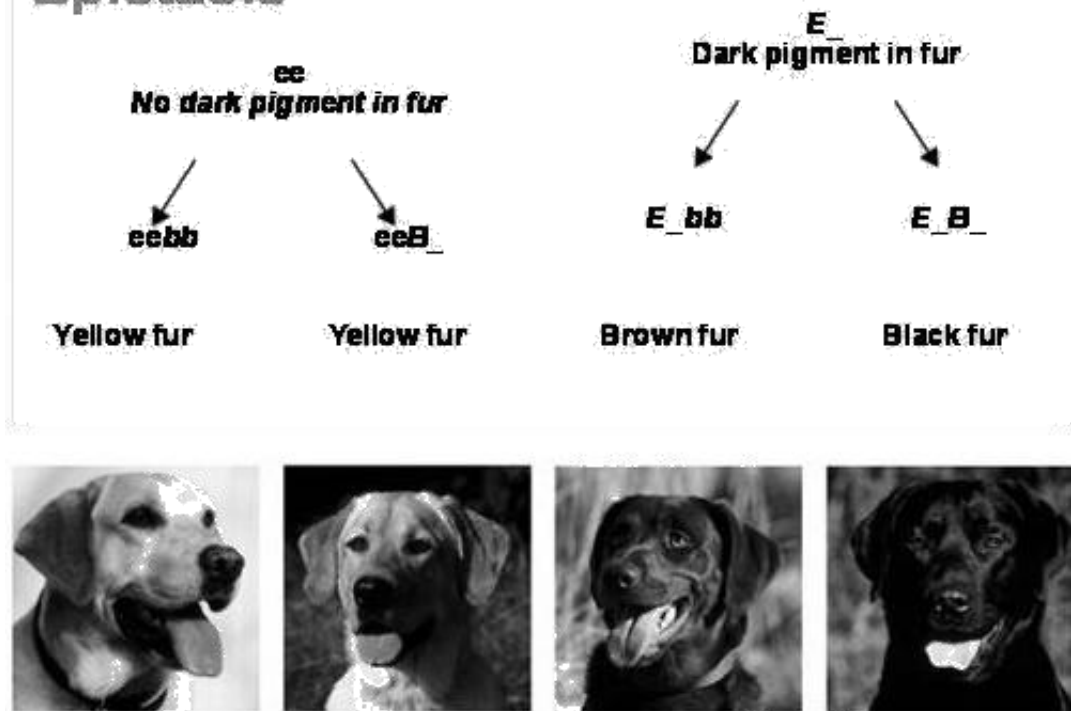
For example? Color of skin man. The heterozygous individuals ($AaBbCc$) represented by the two rectangles at the top of this figure each carry three dark-skin alleles (black circles, which represent A , B , or C) and three light-skin alleles (white circles, which represent a , b , or c).



Epistasis –One gene’s alleles mask the effects of another gene’s alleles

Labrador retriever example – recessive epistasis –Coat color can be black, chocolate brown, or Golden yellow.

Epistasis



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Pleiotropy

This is when a single gene locus affects more than one trait.

For example, in Labrador retrievers the gene locus that controls how dark the pigment in the hair will be also affects the color of the nose, lips, and eye rims. Pleiotropic effects are characteristic of many inherited disorders, such as cystic fibrosis and sickle cell anemia, both discussed later in this chapter. In these disorders, multiple symptoms can be traced back to a single gene defect. In cystic fibrosis, patients exhibit clogged blood vessels, overly sticky mucus, salty sweat, liver and pancreas failure, and a battery of other symptoms. All are pleiotropic effects of a single defect, a mutation in a gene that encodes a chloride ion transmembrane channel. In sickle cell anemia, a defect in the oxygen-carrying hemoglobin molecule causes anemia, heart failure, increased susceptibility to pneumonia, kidney failure, enlargement of the spleen, and many other symptoms. It is usually difficult to deduce the nature of the primary defect from the range of a gene's pleiotropic effects.

CHROMOSOMES - A UNIT OF HEREDITY

Mendel published his results in a scientific journal in 1866. However, his work was not appreciated and forgotten. Independently, Karl Correns, Erich von Tschermak, and Hugo de Vries all found that Mendel had explained the same results 35 years before. It is known that the number of genes is considerably higher than the number of chromosomes. Around 1900, cytologists and geneticists began to see parallels between the behavior of chromosomes and the behavior of Mendel's factors.

In 1909 Johannsen entered the term gene - a unit of heredity.

Thomas Morgan was the first to associate a specific gene with a specific chromosome in the early 20th century.



Like Mendel, Morgan made an insightful choice as an experimental animal, *Drosophila melanogaster*, a fruit fly species that eats fungi on fruit.

Morgan crossed fruit fly, observed followed the inheritance of characters for body color and wing size. The wild-type body color is gray (b^+) and the mutant black (b). The wild-type wing size is normal (vg^+) and the mutant has vestigial wings (vg). The first filial was wild-type with gray body and normal wings (dominance genes). Morgan crossed F_1 heterozygous females (b^+bvg^+vg) with homozygous recessive males ($bbvgvg$). According to independent assortment, this should produce 4 phenotypes in a 1:1:1:1 ratio.

How does linkage between two genes affect inheritance of characters?

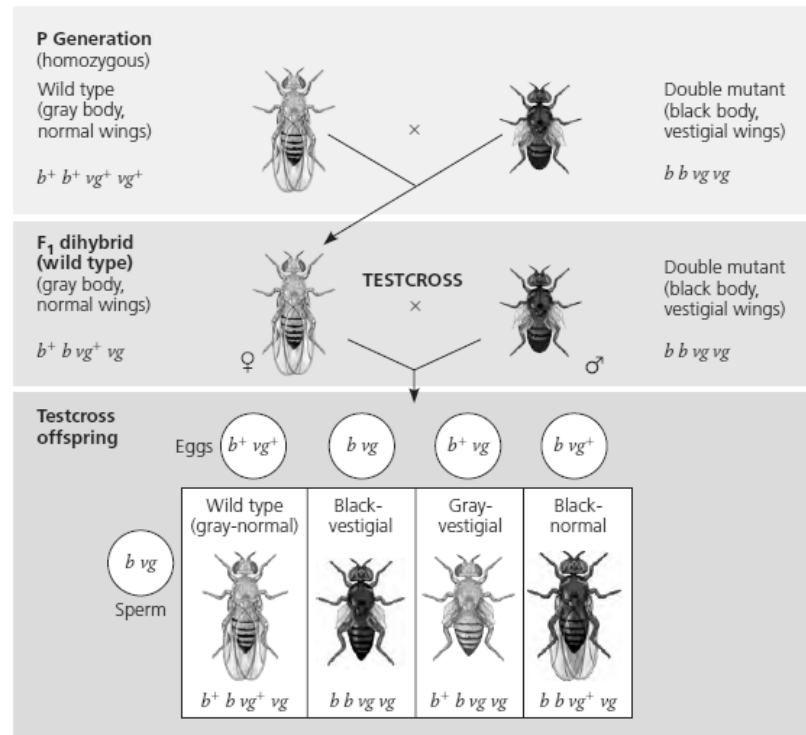
EXPERIMENT Morgan wanted to know whether the genes for body color and wing size were genetically linked, and if so, how this affected their inheritance. The alleles for body color are b^+ (gray) and b (black), and those for wing size are vg^+ (normal) and vg (vestigial).

Morgan mated true-breeding P (parental) generation flies—wild-type flies with black, vestigial-winged flies—to produce heterozygous F_1 dihybrids (b^+bvg^+vg), all of which are wild-type in appearance.

He then mated wild-type F_1 dihybrid females with black, vestigial-winged males. This testcross will reveal the genotype of the eggs made by the dihybrid female.

The male's sperm contributes only recessive alleles, so the phenotype of the offspring reflects the genotype of the female's eggs.

Note: Although only females (with pointed abdomens) are shown, half the offspring in each class would be males (with rounded abdomens).



PREDICTED RATIOS

If genes are located on different chromosomes:

1 : 1 : 1 : 1

If genes are located on the same chromosome and parental alleles are always inherited together:

1 : 1 : 0 : 0

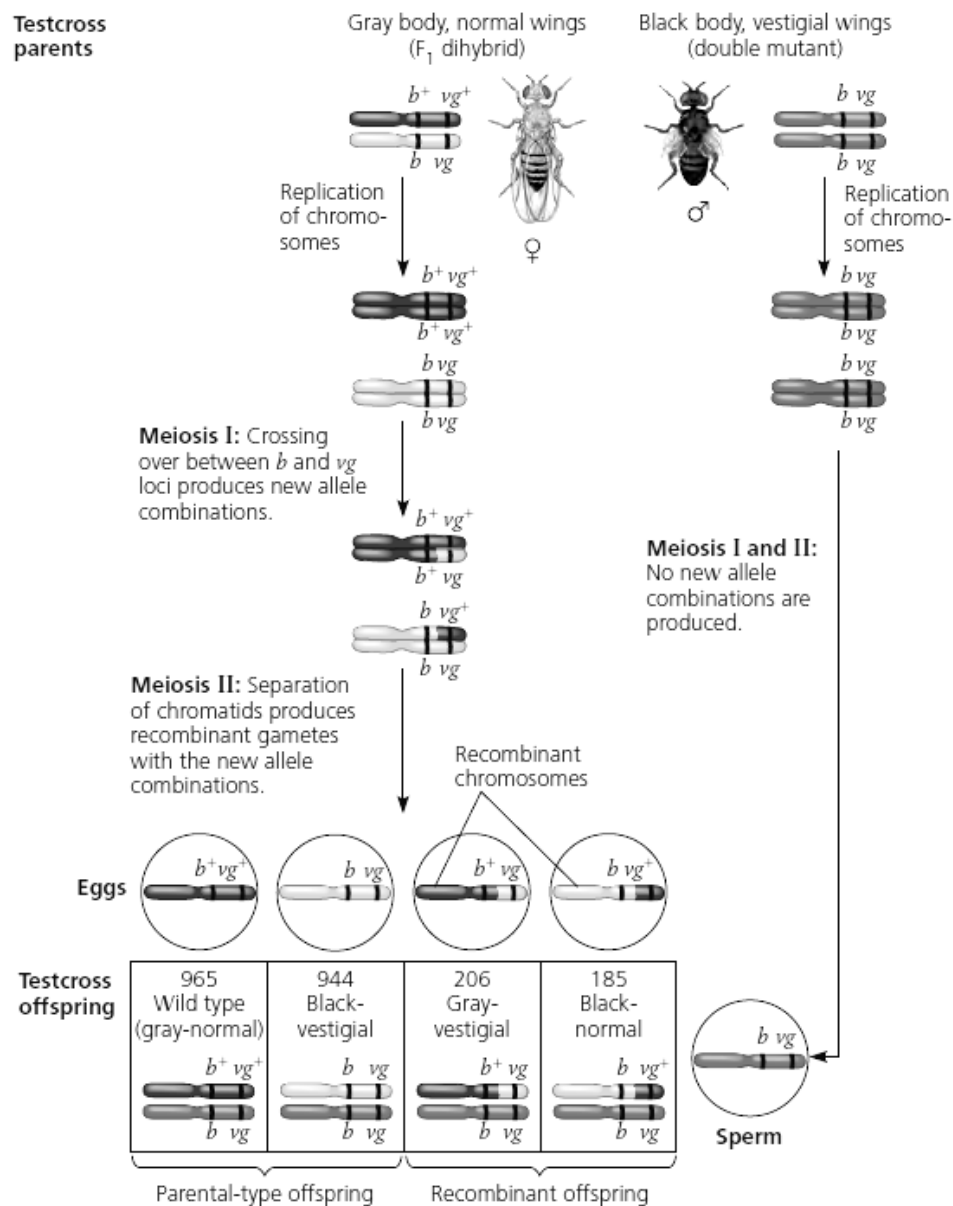
RESULTS

965 : 944 : 206 : 185

CONCLUSION Since most offspring had a parental (P generation) phenotype, Morgan concluded that the genes for body color and wing size are genetically linked on the same chromosome. However, the production of a relatively small number of offspring with nonparental phenotypes indicated that some mechanism occasionally breaks the linkage between specific alleles of genes on the same chromosome.

Surprisingly, Morgan observed a large number of wild-type (gray-normal) and double-mutant (black-vestigial) flies among the offspring. These phenotypes correspond to those of the parents.

Morgan reasoned that body color and wing shape are usually inherited together because their genes are on the same chromosome. The other two phenotypes (gray-vestigial and black-normal) were fewer than expected from independent assortment (and totally unexpected from dependent assortment). These new phenotypic variations must be the result of crossing over.



$$\text{Recombination frequency} = \frac{391 \text{ recombinants}}{2,300 \text{ total offspring}} \times 100 = 17\%$$

*Chromosomal basis for recombination of linked genes. In these diagrams recreating the testcross Morgan's experiment, we track chromosomes as well as genes. The maternal chromosomes are color-coded black and white to distinguish one homolog from the other before any meiotic crossing over has taken place. Because crossing over between the *b* and *vg* loci occurs in some, but not all, egg-producing cells, more eggs with parental-type chromosomes than with recombinant ones are produced in the mating females. Fertilization of the eggs by sperm of genotype *b vg* gives rise to some recombinant offspring. The recombination frequency is the percentage of recombinant flies in the total pool of offspring.*

Morgan formulated a ***chromosome theory of inheritance***:

- Each chromosome has hundreds or thousands of genes.
- Genes located on the same chromosome, linked genes, tend to be inherited together because the chromosome is passed along as a unit.
- Results of crosses with linked genes deviate from those expected according to independent assortment.
- The production of offspring with new combinations of traits inherited from two parents is genetic recombination.
- Genetic recombination can result from independent assortment of genes located on nonhomologous chromosomes or from crossing over of genes located on homologous chromosomes.

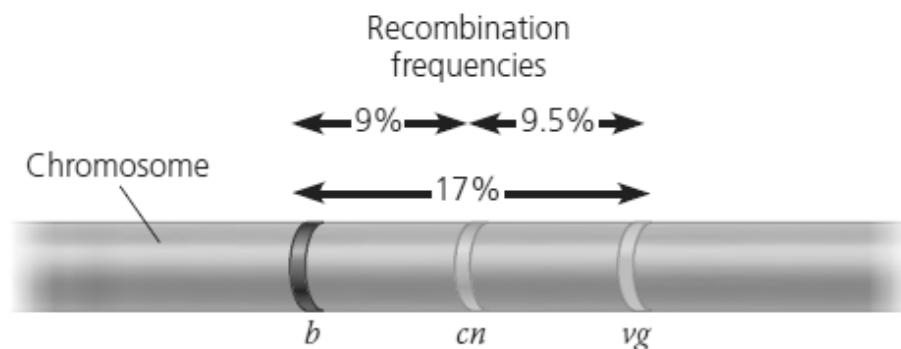
Because crossovers occur along the length of a chromosome at random, the farther apart 2 genes are, the larger the chance a crossover will occur between them, and the higher the frequency of recombinant types.

Therefore, the frequency of recombinant types can be used to construct genetic maps. ***The distance between genes is proportional to the frequency of recombination events.***

APPLICATION: A linkage map shows the relative locations of genes along a chromosome.

TECHNIQUE: A linkage map is based on the assumption that the probability of a crossover between two genetic loci is proportional to the distance separating the loci. The recombination frequencies used to construct a linkage map for a particular chromosome are obtained from Morgan's experimental crosses. The distances between genes are expressed as map units, with one map unit equivalent to a 1% recombination frequency. Genes are arranged on the chromosome in the order that best fits the data.

RESULTS: In this example, the observed recombination frequencies between three *Drosophila* gene pairs (*b*–*cn* 9%, *cn*–*vg* 9.5%, and *b*–*vg* 17%) best fit a linear order in which *cn* is positioned about halfway between the other two genes:



The *b*–*vg* recombination frequency (17%) is slightly less than the sum of the *b*–*cn* and *cn*–*vg* frequencies ($9 + 9.5 = 18.5\%$) because of the few times that one crossover occurs between *b* and *cn* and another crossover occurs between *cn* and *vg*. The second crossover would “cancel out” the first, reducing the observed *b*–*vg* recombination frequency while contributing to the frequency between each of the closer pairs of genes. The value of 18.5% (18.5 map units) is closer to the actual distance between the genes, so a geneticist would add the smaller distances in constructing a map.

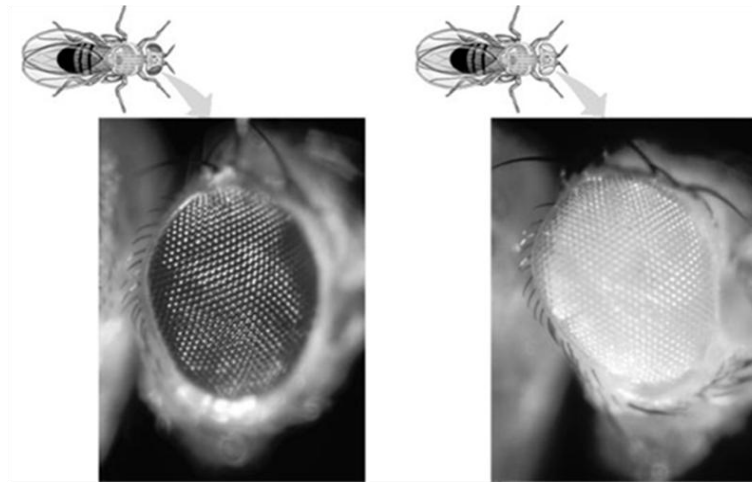
SEX LINKAGE

Sex in humans (and other organisms) is determined by genes located on a pair of chromosomes called the sex chromosomes. All other chromosomes are called autosomes.

The remaining pair, the sex chromosomes, consist of two similar chromosomes in females and two dissimilar chromosomes in males. In humans, females are designated XX and males XY. One of the sex chromosomes in the male (the Y chromosome) is highly condensed and bears few functional genes. Because few genes on the Y chromosome are expressed, recessive alleles on a male's single X chromosome have no active counterpart on the Y chromosome. Some of the active genes the Y chromosome does possess are responsible for the features associated with "maleness" in humans. Consequently, any individual with at least one Y chromosome is a male. Even though the sex chromosomes pair during synapsis, they are not homologous. The larger chromosome is called the X and the smaller is the Y. In humans, XX is female and XY is male.

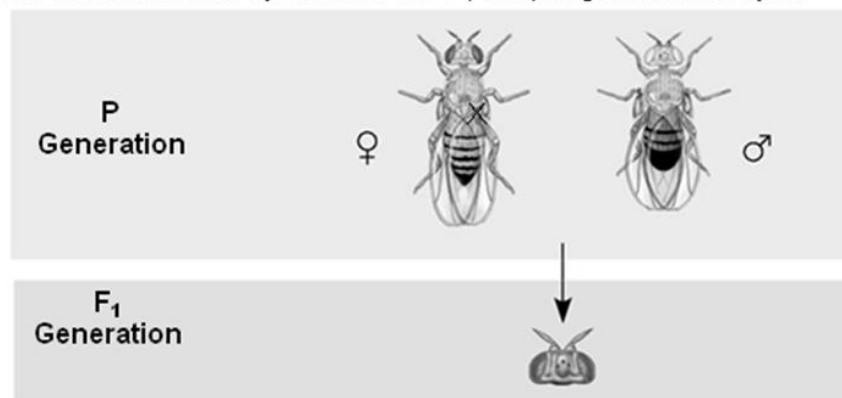
The structure and number of sex chromosomes vary in different organisms. In the fruit fly *Drosophila*, females are XX and males XY, as in humans and most other vertebrates. However, in birds, the male has two Z chromosomes, and the female has a Z and a W chromosome. In some insects, such as grasshoppers, there is no Y chromosome - females are XX and males are characterized as XO (the O indicates the absence of a chromosome).

- Morgan observed and noted wild type (normal), phenotypes that were common in the fly populations.
- Traits alternative to the wild type are called mutant phenotypes.



When Morgan crossed his white-eyed male with a red-eyed female, all the F₁ offspring had red eyes. The red allele appeared dominant to the white allele.

EXPERIMENT Morgan mated a wild-type (red-eyed) female with a mutant white-eyed male. The F₁ offspring all had red eyes.



Morgan then bred an F₁ red-eyed female to an F₁ red-eyed male to produce the F₂ generation.

RESULTS The F₂ generation showed a typical Mendelian 3:1 ratio of red eyes to white eyes. However, no females displayed the white-eye trait; they all had red eyes. Half the males had white eyes, and half had red eyes.



Crosses between the F1 offspring produced the classic 3:1 phenotypic ratio in the F2 offspring.

Surprisingly, the white-eyed trait appeared only in males. All the females and half the males had red eyes. Morgan concluded that a fly's eye color was linked to its sex. Morgan deduced that the gene with the white-eyed mutation is on the X chromosome alone, a sex-linked gene. Females (XX) may have two red-eyed alleles and have red eyes or may be heterozygous and have red eyes. Males (XY) have only a single allele and will be red eyed if they have a red-eyed allele or white-eyed if they have a white-eyed allele.

Other traits, besides sex, are controlled by genes on the sex chromosomes. Traits controlled by the X are X-linked. Traits controlled by the Y are Y-linked. Since most sex-linked traits are controlled by the X, its assumed X-linkage. X-linked traits are an exception to Mendel's laws because females have 2 alleles for each X-linked trait, but males have only 1. In humans, hemophilia is caused by a recessive allele on the X chromosome.

GENETICS PROBLEMS

A. Monohybrid Cross in Corn (*Zea mays*).

Corn kernels are actually the fruit of the corn plant, each containing an individual corn embryo. Around the embryo are a number of structures providing nourishment and protection. Between the endosperm (where starch is stored) and the pericarp (covering of the kernel) is a layer of cells called the aleurone. The color of the aleurone is controlled by several genes. One gene produces a purple aleurone, the other the yellow kernels with which we are most familiar. The purple allele is dominant, the yellow allele being recessive. The dominant purple allele can be symbolized by "R" while "r" represents the recessive yellow allele. Therefore, corn kernels with the homozygous genotype "RR" are purple as are

those kernels who are heterozygous (“Rr”). Only kernels homozygous for the yellow allele (“rr”) will express the recessive phenotype, yellow.

Procedure:

1. Obtain an ear of genetic corn. Notice it contains a mixture of purple and yellow kernels. This ear of corn represents the **F2 generation** from the following cross:

R – purple color

r – yellow color

P: ♀ Rr x ♂ Rr

G: R, r R, r

F₁: RR, 2 Rr, rr

The parents (“Rr” and “Rr”) are the **F1 generation** and were obtained from the **P generation** cross of a homozygous dominant (“RR”) and recessive (“rr”) individual. The offspring (“RR, ‘Rr’, “rr”) are then the **F2 generation**. This cross results in a mixture of phenotypes in the F2 generation. Most of the kernels are purple, a fewer number are yellow. Punnett square diagrams are used to diagram genetic crosses. Using this mating, fill in the Punnett square in Table 1.

Table 1. Monohybrid Cross Punnett Square for Kernel Color in Corn (*Zea mays*)

F1 gametes	R	r
R	genotype _____ phenotype _____	genotype _____ phenotype _____
r	genotype _____ phenotype _____	genotype _____ phenotype _____

Based upon the Punnett square in Table 11.1, about what proportion of the kernels in the ear of corns should be purple? _____

What proportion should be yellow? _____

2. Place a straight pin at the end of the row being counted to serve as a place holder. Count the number of purple kernels and yellow kernels in **one row only**. Enter data in Table 2

Table 2. Kernel Counts in F2 Generation Corn (*Zea mays*) – One Row

# of kernels	Number	Proportion*
purple		
yellow		
total		

*divide the number of kernels of each color by the total number of kernels in that row

Is the proportion of purple to yellow kernels close to the predicted 3:1 (75% purple, 25% yellow) ratio? _____

Phenylthiocarbimide (PTC) Taste Test.

Phenylthiocarbimide is an organic compound which interacts with the taste buds on the tongue and mouth of some people but not in others. Therefore, some people can taste PTC and are considered “tasters.” Other people cannot taste PTC and are considered “non-tasters.” Being able to taste is the dominant condition (“T”), non-tasters the recessive (“t”).

Procedure

1. Obtain a piece of paper treated with a small amount of PTC and place it on the tongue. Tasters will sense a very strong flavor, non-tasters will taste only “paper”

2. Collect results on the number of tasters and non-tasters from the population in class and use that data to fill in Table:

Table. PTC Tasting

phenotypes	Number	Proportion*
taster		
non-taster		
total		

*divide the number of phenotypes by the total class size.

What is the genotype of a taster? _____

What is the genotype of a non-taster? _____

What is the predicted phenotypic ratio for a cross between two heterozygous (Tt) individuals? _____

Were the resultant proportions close to this ratio? _____

If the proportions were not close (and even if they were), provide an explanation as to why they would be (hint: sample size).

B. Inheritance of blood types

Some genes have more than two alleles. Although each organism can only have two copies of a gene, within an entire population there may be several alleles.

Blood types are produced by **multiple alleles**. On the membranes of red blood cells are proteins which can stimulate an immune response. These proteins are called **antigens**. The dominant allele “I^A” produces the A antigen protein, the dominant allele “I^B” produces the B antigen protein. The recessive allele “i” produces no protein. When a genetic trait has more than one dominant allele, a situation of **codominance** may exist. The genetic trait for human ABO blood typing contains three alleles (I^A, I^B, i).

Blood type	Possible genotypes
The first type	i i

The second type	$I^A I^A, I^A i$
The third type	$I^B I^B, I^B i$
The fourth type	$I^A I^B$

Fill in Table with the possible genotypes for the listed phenotypes

Human ABO Blood Typing Genotypes and Phenotypes

Genotype(s)	Phenotype
_____ or _____	A
_____ or _____	B
_____	AB
_____	O

№1. The woman has a first group of blood, a man has a fourth blood group. What blood group can we expect the children of their marriage.

№2. Mother has a second group of blood, the father has a third group of blood. They had a son with the first group of blood. what blood group may be the children of this a married couple.

№3. Mother has a second group of blood and brown eye color, the father has a third group of blood and blue eyes. They had a son with the first group of blood and blue eyes. What kind of blood and eye color may be the children of this a married couple. It is known that brown eye color is dominant over blue.

C: X-Linked Characteristics.

In humans, all somatic cells (typical body cells) contain 23 pairs of chromosomes. Of these, 22 pairs are autosomes, the last pair are the sex

chromosomes. The sex chromosomes are related to the gender of the individual and are called X and Y. Women have two X chromosomes, men have an X and a Y. Haploid sex cells (gametes) produced by women (ova) have only the X chromosome, male gametes (sperm) have either X or Y. Sex of the offspring is determined by the male.

The X chromosome is large and carries many genes such as those for essential muscle proteins and retinal pigments. The Y chromosome, on the other hand, is quite small and carries only a few genes, mostly related to male gender development. A defective gene on the X chromosome will be phenotypically expressed in a male because there is no other X chromosome to compensate. However, a woman with the same defective gene will not express it phenotypically if her other X chromosome is normal. She will be a carrier though in that she has the defective gene but does not express it.

Red-green colorblindness is caused by a mutation in a gene for retinal pigments on the X chromosome. The defective allele is recessive to the normal one so a woman with one normal X chromosome (X) and one colorblind carrying chromosome (X^C) will have normal color vision because the X chromosome is dominant for this trait over the X^C chromosome. Men, however will be colorblind if they possess the X^C chromosome since there is no other X to be dominate over it and will exhibit red-green colorblindness.

Procedure:

1. Obtain a color vision diagnostic chart
2. Test for colorblindness in yourself and among lab partners

What is your gender? _____ (if you don't know, excuse yourself to the restroom to find out)

What is your sex chromosome genotype? _____

Do you exhibit red-green colorblindness? _____

Do any lab partners exhibit colorblindness? _____

If so, what are their sexes and genotypes? _____

D: Dihybrid crosses.

Dihybrid crosses are used to examine the inheritance patterns in more than one gene (*di* = *two*). As an example, consider fur color and texture in guinea pigs. Among these organisms, black fur color is dominant (“B”) over white (“b”) and rough fur coat is dominant (“R”) over smooth (“r”).

Procedure:

B – black; b – white

R – rough; r – smooth

1.

P: ♀ BBRR x ♂ bbrr

G: BR br

F₁: BbRr 100%

F1 gametes	BR
br	genotype _____ phenotype _____

Use a Punnett Square to answer the questions regarding the offspring in the F₁ generation from the P generation crosses indicated

What will the genotype be of all of the offspring from this cross?

What will be the phenotypes of all of the offspring from this cross?

2.

P: ♀ BbRr x ♂ BbRr

G:

F₂:

F2 gametes	BR	Br	bR	br
BR	genotype _____ phenotype _____	genotype _____ phenotype _____	genotype _____ phenotype _____	genotype _____ phenotype _____
Br	genotype _____ phenotype _____	genotype _____ phenotype _____	genotype _____ phenotype _____	genotype _____ phenotype _____
bR	genotype _____ phenotype _____	genotype _____ phenotype _____	genotype _____ phenotype _____	genotype _____ phenotype _____
br	genotype _____ phenotype _____	genotype _____ phenotype _____	genotype _____ phenotype _____	genotype _____ phenotype _____

What proportion of the guinea pigs will be

black, rough? _____

black, smooth? _____

white, rough? _____

white, smooth? _____

3.

P: ♀ BbRr x ♂ bbRr

G:

F₂:

F2 gametes				
	genotype _____ phenotype _____	genotype _____ phenotype _____	genotype _____ phenotype _____	genotype _____ phenotype _____
	genotype _____ phenotype _____	genotype _____ phenotype _____	genotype _____ phenotype _____	genotype _____ phenotype _____

What proportion of the guinea pigs will be

black, rough? _____

black, smooth? _____

white, rough? _____

white, smooth? _____

4.

P: ♀ Bbrr x ♂ bbRr

G:

F₂:

F2 gametes		
	genotype _____ phenotype _____	genotype _____ phenotype _____
	genotype _____ phenotype _____	genotype _____ phenotype _____

What proportion of the guinea pigs will be

black, rough? _____

black, smooth? _____

white, rough? _____

white, smooth? _____

Practice Problems and Review Questions:

1. What are the expected phenotypic and genotypic ratios in a monohybrid cross between two heterozygous individuals?
2. What is the expected phenotypic ratio in a dihybrid cross between two organisms that are heterozygous for both traits?

3. List all the possible types of gametes that can be produced from an organism with the genotype AaBb.

4. Humans as well as many other mammals show a genetic condition known as **albinism**. A recessive allele interferes with the ability to produce the brown pigment **melanin** which colors eyes and hair in addition to protecting the skin from the harmful effects of UV light. People who are homozygous recessive produce little or no melanin and have very pale eyes, white skin, and yellow or white hair. Normal pigmentation is produced by a dominant allele (“A”). The albino allele is recessive (“a”). Use Punnett Squares to calculate genotypic and phenotypic proportions for the parental generation crosses indicated and denote which F1 genotypes (if any) will be carriers .

a. AA x aa;

b. Aa x aa;

c. Aa x Aa;

d. AA x Aa;

e. aa x aa.

5. A woman with normal pigmentation, whose mother was an albino, mates with an albino. What is the probability their first child will be an albino?

6. Normal parents have an albino child. Give the genotypes for the parents and the child.

7. A person is a taster as is their mother. The father is a non-taster. What is this person’s genotype?

8. A non-taster has two taster parents. What is the genotype of this person and their parents?

9. Can two normal vision parents produce a colorblind son? Explain with a diagram.

10. Can two normal vision parents produce a colorblind daughter? Explain with a diagram.

11. Can two colorblind parents, produce a normal vision son? Explain with a diagram.

12. Write down symbols for the alleles. (These may be given in the problem.) When represented by single letters, the dominant allele is uppercase and the recessive is lowercase.

13. Is it possible for a female human to be colorblind? What would her genotype be?

14. Write down the possible genotypes, as determined by the phenotype.

a. If the phenotype is that of the dominant trait (for example, purple flowers), then the genotype is either homozygous dominant or heterozygous (PP or Pp, in this example).

b. If the phenotype is that of the recessive trait, the genotype must be homozygous recessive (for example, pp).

c. If the problem says “true-breeding,” the genotype is homozygous.

15. A normal vision woman, whose father was colorblind, mates with a colorblind man. What proportion of their sons would be colorblind? What proportion of their daughters would be colorblind and what proportion would be carriers? If one of their normal vision sons mates with a homozygous, normal vision woman, would it be possible for them to have a colorblind child?

16. Two pea plants heterozygous for the characters of pod color and pod shape are crossed. Draw a Punnett square to determine the phenotypic ratios of the offspring.

17. In some plants, a true-breeding, red-flowered strain gives all pink flowers when crossed with a white-flowered strain: $C^R C^R$ (red) \times $C^W C^W$ (white) \rightarrow $C^R C^W$ (pink). If flower position (axial or terminal) is inherited as it is in peas, what will be the ratios of genotypes and phenotypes of the F1 generation resulting from the following cross: axial-red (true-breeding) \times terminal-white? What will be the ratios in the F2 generation?

18. A man has six fingers on each hand and six toes on each foot. His wife and their daughter have the normal number of digits. Remember that extra digits is a dominant trait. What fraction of this couple's children would be expected to have extra digits?

19. Parents have brown eyes. They had a child with blue eyes. What is the probability of having a child with blue eyes from this marriage? It is known that the brown eye gene is dominant over blue eye gene.

20. Many animals and plants bear recessive alleles for albinism, a condition in which homozygous individuals lack certain pigments. An albino plant, for example, lacks chlorophyll and is white, and an albino human lacks melanin. If two normally pigmented persons heterozygous for the same albinism allele marry, what proportion of their children would you expect to be albino?

21. A normally pigmented man marries an albino woman. They have three children, one of whom is an albino. What is the genotype of the father?

22. A pea plant heterozygous for inflated pods (Ii) is crossed with a plant homozygous for constricted pods (ii). Draw a Punnett square for this cross. Assume that pollen comes from the ii plant.

23. For ABO blood typing in humans, for which phenotypes is the genotype definitive?

24. A person has blood type O. Their mother is B and father is A. What is the genotype of this person and their parents?

25. Four babies are born in a hospital, and each has a different blood type: A, B, AB, and O. The parents of these babies have the following pairs of blood groups: A and B, O and O, AB and O, and B and B. Which baby belongs to which parents?

26. A woman is married for the second time. Her first husband has blood type A and her child by that marriage has type O. Her new husband has type B blood, and when they have a child its blood type is AB. What is the woman's blood genotype and blood type?

27. A couple with a newborn baby is troubled that the child does not resemble either of them. Suspecting that a mix-up occurred at the hospital, they check the blood type of the infant. It is type O. As the father is type A and the mother type B, they conclude a mix-up must have occurred. Are they correct?

28. A man with type A blood marries a woman with type B blood. Their child has type O blood. What are the genotypes of these three individuals? What genotypes, and in what frequencies, would you expect in future offspring from this marriage?

29. A paternity suit involves a child whose blood type is AB. The mother is blood type B, the alleged father is O. Make a ruling on this case as to whether it is reasonably possible this is the biological father.

30. A guinea pig that is heterozygous for fur color and texture mates with another guinea pig heterozygous for both traits. They produce a total of 96 offspring. Use a Punnett Square to diagram this cross then answer the questions below.

How many of the 96 offspring will phenotypically be:

black with rough fur? _____

black with smooth fur? _____

white with rough fur? _____

white with smooth fur? _____

31. Polydactyly is inherited as a dominant autosomal gene, the normal hand inherited as a recessive autosomal gene. Also analyzed the inheritance of blood groups. Woman with polydactyly and 3 blood group marries a man with polydactyly and 2 blood group. They had a child with the normal hand and 1 blood group. What is the probability of having a child with the normal hand and 3 blood group?

32. Myopia and polydactyly are inherited as autosomal dominant genes. A woman has polydactyly and myopia, but her mother did not have these diseases.

She marries a man who does not have these diseases. What is the probability of the birth of children without anomalies of the couple.

33. Flower position, stem length, and seed shape are three characters that Mendel studied. Each is controlled by an independently assorting gene and has dominant and recessive expression as follows:

Character	Dominant	Recessive
Flower position	Axial (A)	Terminal (a)
Stem length	Tall (T)	Dwarf (t)
Seed shape	Round (R)	Wrinkled (r)

If a plant that is heterozygous for all three characters is allowed to self-fertilize, what proportion of the offspring would you expect to be as follows? (Note: Use the rules of probability instead of a huge Punnett square.):

- (a) homozygous for the three dominant traits
- (b) homozygous for the three recessive traits
- (c) heterozygous for all three characters
- (d) homozygous for axial and tall, heterozygous for seed shape

34. A black guinea pig crossed with an albino guinea pig produces 12 black offspring. When the albino is crossed with a second black one, 7 blacks and 5 albinos are obtained. What is the best explanation for this genetic outcome? Write genotypes for the parents, gametes, and offspring.

35. In sesame plants, the one-pod condition (P) is dominant to the three-pod condition (p), and normal leaf (L) is dominant to wrinkled leaf (l). Pod type and leaf type are inherited independently. Determine the genotypes for the two parents for all possible matings producing the following offspring:

- (a) 318 one-pod, normal leaf and 98 one-pod, wrinkled leaf

- (b) 323 three-pod, normal leaf and 106 three-pod, wrinkled leaf
- (c) 401 one-pod, normal leaf
- (d) 150 one-pod, normal leaf, 147 one-pod, wrinkled leaf, 51 three-pod, normal leaf, and 48 three-pod, wrinkled leaf
- (e) 223 one-pod, normal leaf, 72 one-pod, wrinkled leaf, 76 three-pod, normal leaf, and 27 three-pod, wrinkled leaf

36. Phenylketonuria (PKU) is an inherited disease caused by a recessive allele. If a woman and her husband, who are both carriers, have three children, what is the probability of each of the following?

- (a) All three children are of normal phenotype.
- (b) One or more of the three children have the disease.
- (c) All three children have the disease.
- (d) At least one child is phenotypically normal.

(Note: It will help to remember that the probabilities of all possible outcomes always add up to 1.)

37. The genotype of F1 individuals in a tetrahybrid cross is AaBbCcDd. Assuming independent assortment of these four genes, what are the probabilities that F2 offspring will have the following genotypes?

- (a) aabbccdd
- (b) AaBbCcDd
- (c) AABBCcDD
- (d) AaBBccDd
- (e) AaBBCCdd.

38. What is the probability that each of the following pairs of parents will produce the indicated offspring? (Assume independent assortment of all gene pairs.)

- (a) AABBCc × aabbcc → AaBbCc
- (b) AABbCc × AaBbCc → AAbbCC
- (c) AaBbCc × AaBbCc → AaBbCc

(d) $aaBbCC \times AABbcc \rightarrow AaBbCc$.

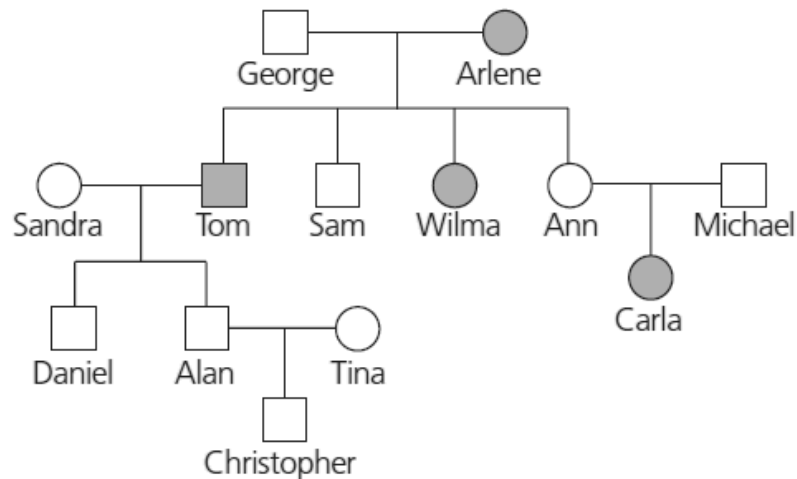
39. In the fly *Drosophila*, the allele for dumpy wings (d) is recessive to the normal long-wing allele (d^+), and the allele for white eye (w) is recessive to the normal red eye allele (w^+). In a cross of $d^+d^+w^+w \times d^+dww$, what proportion of the offspring are expected to be “normal” (long wings, red eyes)? What proportion are expected to have dumpy wings and white eyes?

40. Your instructor presents you with a *Drosophila* with red eyes, as well as a stock of white-eyed flies and another stock of flies homozygous for the red-eye allele. You know that the presence of white eyes in *Drosophila* is caused by homozygosity for a recessive allele. How would you determine whether the single red-eyed fly was heterozygous for the white-eye allele?

41. In tigers, a recessive allele causes an absence of fur pigmentation (a white tiger) and a cross-eyed condition. If two phenotypically normal tigers that are heterozygous at this locus are mated, what percentage of their offspring will be cross-eyed? What percentage of cross-eyed tigers will be white?

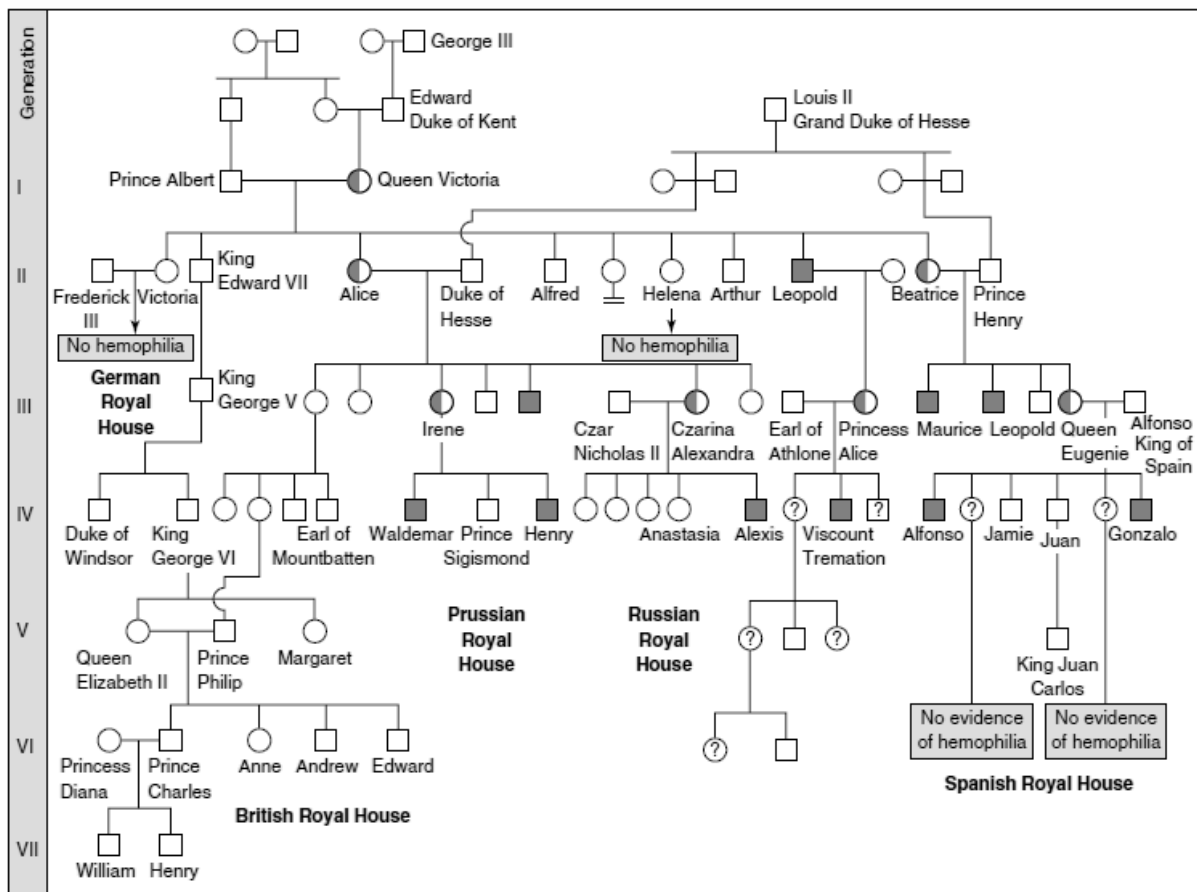
42. In maize (corn) plants, a dominant allele I inhibits kernel color, while the recessive allele i permits color when homozygous. At a different locus, the dominant allele P causes purple kernel color, while the homozygous recessive genotype pp causes red kernels. If plants heterozygous at both loci are crossed, what will be the phenotypic ratio of the offspring?

43. The pedigree below traces the inheritance of alkaptonuria, a biochemical disorder. Affected individuals, indicated here by the colored circles and squares, are unable to metabolize a substance called alkapton, which colors the urine and stains body tissues. Does alkaptonuria appear to be caused by a dominant allele or by a recessive allele? Fill in the genotypes of the individuals whose genotypes can be deduced. What genotypes are possible for each of the other individuals?



44. Hemophilia is a recessive sex-linked human blood disease that leads to failure of blood to clot normally. One form of hemophilia has been traced to the royal family of England, from which it spread throughout the royal families of Europe. For the purposes of this problem, assume that it originated as a mutation either in Prince Albert or in his wife, Queen Victoria.

- a. Prince Albert did not have hemophilia. If the disease is a sex-linked recessive abnormality, how could it have originated in Prince Albert, a male, who would have been expected to exhibit sex-linked recessive traits?
- b. Alexis, the son of Czar Nicholas II of Russia and Empress Alexandra (a granddaughter of Victoria), had hemophilia, but their daughter Anastasia did not. Anastasia died, a victim of the Russian revolution, before she had any children. Can we assume that Anastasia would have been a carrier of the disease? Would your answer be different if the disease had been present in Nicholas II or in Alexandra?



The Royal hemophilia pedigree. Queen Victoria's daughter Alice introduced hemophilia into the Russian and Austrian royal houses, and Victoria's daughter Beatrice introduced it into the Spanish royal house. Victoria's son Leopold, himself a victim, also transmitted the disorder in a third line of descent. Half-shaded symbols represent carriers with one normal allele and one defective allele; fully shaded symbols represent affected individuals.

45. Imagine that you are a genetic counselor, and a couple planning to start a family comes to you for information. Charles was married once before, and he and his first wife had a child with cystic fibrosis. The brother of his current wife, Elaine, died of cystic fibrosis. What is the probability that Charles and Elaine will have a baby with cystic fibrosis? (Neither Charles, Elaine, nor their parents have cystic fibrosis.)

46. In mice, black fur (B) is dominant to white (b). At a different locus, a dominant allele (A) produces a band of yellow just below the tip of each hair in mice with black fur. This gives a frosted appearance known as agouti. Expression

of the recessive allele (a) results in a solid coat color. If mice that are heterozygous at both loci are crossed, what is the expected phenotypic ratio of their offspring?

47. A man with hemophilia (a recessive, sex-linked condition) has a daughter of normal phenotype. She marries a man who is normal for the trait. What is the probability that a daughter of this mating will be a hemophiliac? That a son will be a hemophiliac? If the couple has four sons, what is the probability that all four will be born with hemophilia?

48. Pseudohypertrophic muscular dystrophy is an inherited disorder that causes gradual deterioration of the muscles. It is seen almost exclusively in boys born to apparently normal parents and usually results in death in the early teens. Is this disorder caused by a dominant or a recessive allele? Is its inheritance sex-linked or autosomal? How do you know? Explain why this disorder is almost never seen in girls.

49. A wild-type fruit fly (heterozygous for gray body color and normal wings) is mated with a black fly with vestigial wings. The offspring have the following phenotypic distribution:

wild-type, 778;

black-vestigial, 785;

black-normal, 158;

grayvestigial, 162.

What is the recombination frequency between these genes for body color and wing size?

50. What pattern of inheritance would lead a geneticist to suspect that an inherited disorder of cell metabolism is due to a defective mitochondrial gene?

51. A space probe discovers a planet inhabited by creatures that reproduce with the same hereditary patterns seen in humans. Three phenotypic characters are height (T - tall, t - dwarf), head appendages (A - antennae, a - no antennae), and nose morphology (S - upturned snout, s - downturned snout). Since the creatures are not “intelligent,” Earth scientists are able to do some controlled breeding

experiments using various heterozygotes in testcrosses. For tall heterozygotes with antennae, the offspring are:

- tall-antennae, 46;
- dwarf-antennae, 7;
- dwarf-no antennae, 42;
- tall-no antennae, 5.

For heterozygotes with antennae and an upturned snout, the offspring are:

- antennae-upturned snout, 47;
- antennae-downturned snout, 2;
- no antennae-downturned snout, 48;
- no antennae-upturned snout, 3.

Calculate the recombination frequencies for both experiments.

52. Using the information from problem 51, scientists do a further testcross using a heterozygote for height and nose morphology. The offspring are:

- tall-upturned snout, 40;
- dwarf-upturned snout, 9;
- dwarf-downturned snout, 42;
- tall-downturned snout, 9.

Calculate the recombination frequency from these data; then use your answer from problem 51 to determine the correct sequence of the three linked genes.

53. You collect two individuals of *Drosophila*, one a young male and the other a young, unmated female. Both are normal in appearance, with the red eyes typical of *Drosophila*. You keep the two flies in the same bottle, where they mate. Two weeks later, the offspring they have produced all have red eyes. From among the offspring, you select 100 individuals, some male and some female. You cross each individually with a fly you know to be homozygous for the recessive allele *sepia*, which produces black eyes when homozygous. Examining the results of your 100 crosses, you observe that in about half of the crosses, only red-eyed flies

were produced. In the other half, however, the progeny of each cross consists of about 50% red-eyed flies and 50% black-eyed flies. What were the genotypes of your original two flies?

54. Red-green color blindness is caused by a sex-linked recessive allele. A color-blind man marries a woman with normal vision whose father was color-blind. What is the probability that they will have a color-blind daughter? What is the probability that their first son will be color-blind? (Note the different wording in the two questions.)

55. A wild-type fruit fly (heterozygous for gray body color and red eyes) is mated with a black fruit fly with purple eyes. The offspring are:

wild-type, 721;

black-purple, 751;

gray-purple, 49;

black-red, 45.

What is the recombination frequency between these genes for body color and eye color? Using information from problem 49, what fruit flies (genotypes and phenotypes) would you mate to determine the sequence of the body-color, wing-size, and eye-color genes on the chromosome?

56. A fruit fly that is true-breeding for gray body with vestigial wings (b^+b^+vgvg) is mated with one that is true-breeding for black body with normal wings ($bbvg^+vg^+$).

(a) Draw the chromosomes for the P generation flies, using red for the gray fly and pink for the black one. Show the position of each allele.

(b) Draw the chromosomes and label the alleles of an F1 fly.

(c) Suppose an F1 female is testcrossed. Draw the chromosomes of the resulting offspring in a Punnett square.

(d) Knowing that the distance between these two genes is 17 map units, predict the phenotypic ratios of these offspring.

57. Women born with an extra X chromosome (XXX) are generally healthy and indistinguishable in appearance from normal XX women. What is a likely explanation for this finding? How could you test this explanation?

58. Determine the sequence of genes along a chromosome based on the following recombination frequencies:

A–B, 8 map units;

A–C, 28 map units;

A–D, 25 map units;

B–C, 20 map units;

B–D, 33 map units.

59. Assume that genes A and B are on the same chromosome and are 50 map units apart. An animal heterozygous at both loci is crossed with one that is homozygous recessive at both loci. What percentage of the offspring will show recombinant phenotypes resulting from crossovers? Without knowing these genes are on the same chromosome, how would you interpret the results of this cross?

60. Two genes of a flower, one controlling blue (B) versus white (b) petals and the other controlling round (R) versus oval (r) stamens, are linked and are 10 map units apart. You cross a homozygous blue-oval plant with a homozygous white-round plant. The resulting F1 progeny are crossed with homozygous white-oval plants, and 1,000 F2 progeny are obtained. How many F2 plants of each of the four phenotypes do you expect?

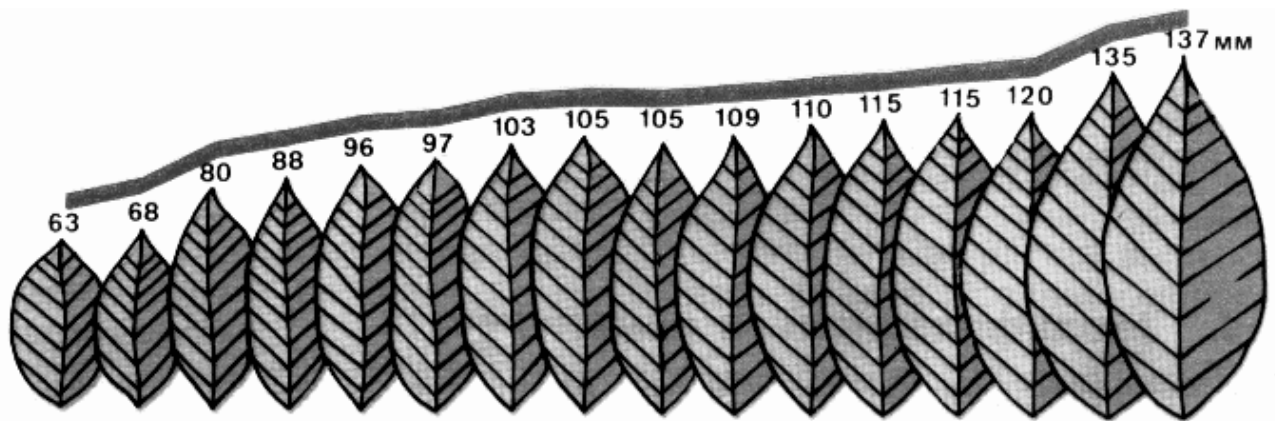
61. Describe the following concepts:

Genetics	Heredity	Gene
Locus	Allele	Dominant allele,
Recessive alleles	Homologous chromosomes	Homozygote
Heterozygote	Monohybrid cross	Dihybrid cross
Genotype,	Phenotype	

4. VARIATION

MODIFICATIONAL VARIABILITY

Modificational variability - variability, affecting only the phenotype and not affecting the genotype.



Features:

1. Do not inherited (as they do not affect the genotype).
2. Wears group character as similar changes occur in a group of individuals.
3. Predictable as the result of factors can predict the actions.
4. Referred to as the changes that occur under the influence of factors that are often adaptive nature.
5. Reversibility arise because changes may be reversible. However, if action is not specific factor or it acts in a critical period of development, then there may be irreversible changes called Morphosis.
6. The boundaries of variability is called the norm of reaction and defined genotype.

Modification variability studied by the variational-statistical method.

MUTATIONAL VARIABILITY

Mutational variability is volatility, which is caused by mutations, ie unpredictable abrupt changes genotype (namely genome chromosomes or genes).

Features:

1. It is inherited, because it affects the genotype.
2. It is individual.
3. It is unpredictable because it is impossible to predict what changes occur in response to factors.
4. The changes occurring in organisms do not usually wear an adaptive character.

Classification of mutations:

1. Mutations associated with changes in the genotype: genetic, chromosomal, genomic.
2. Mutations in different ways affect the viability: the lethal, half-lethal, neutral.
3. Mutations vary in behavior in the heterozygote: dominant and recessive.
4. Mutations vary in relation to the generative ways: somatic (occur in normal cells of the body and are not inherited) and generative (occur in germ cells, therefore inherited).
5. The mutations differ in their localization in the cell: nuclear (occurring in the DNA of the nucleus) and cytoplasmic (occurring in the DNA of mitochondria and plastids).
6. The mutations differ in the causes of mutation: spontaneous (the reason is not clear) and induced (called mutagens).

Mutagens are environmental factors that cause mutation.

By nature, mutagens are:

- Physical (X-rays);
- Chemicals (asbestos, formaldehyde);
- Biological (DNA viruses).

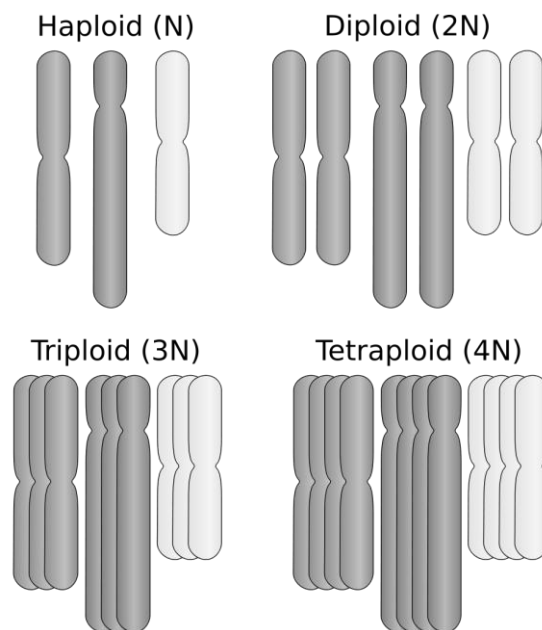
Genome mutations are associated with a change in the number of chromosomes in the genome (haploid set of chromosomes).

There are 2 types:

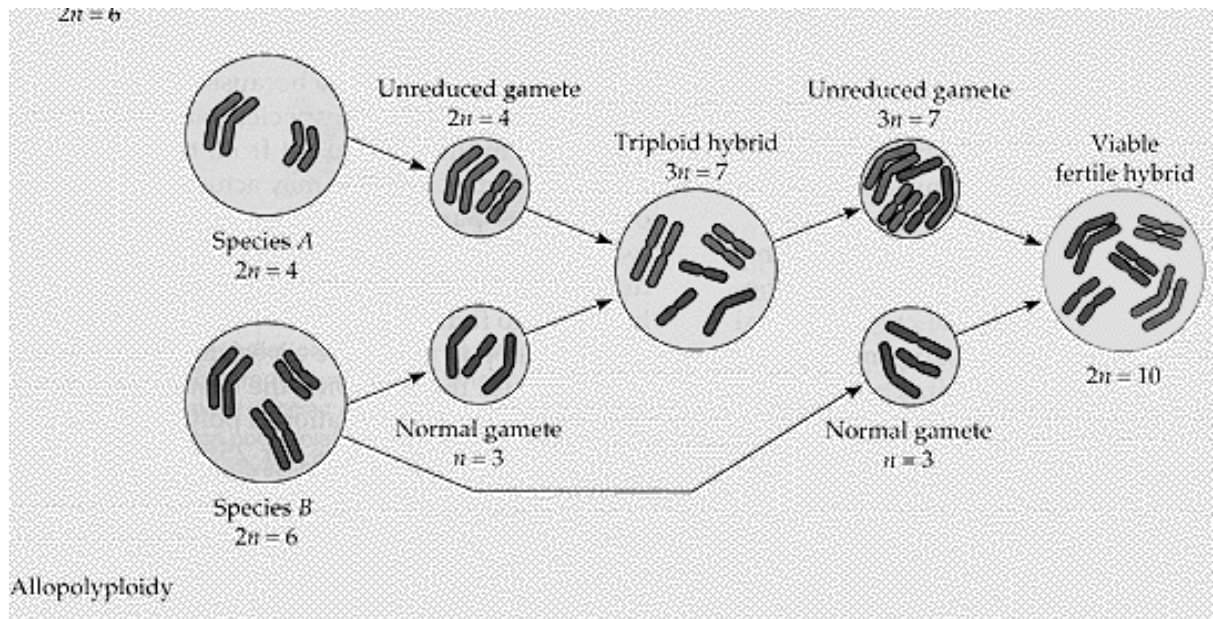
- 1. Polyploidy (euploidiya) is the change in the number of chromosomes, a multiple of the haploid set.

There are autotetraploids and allopolyploid.

- A) autopolyploidy associated with multiple repetition of the same chromosome set. For example, there are types of irises containing 18 chromosomes ($2n$), chromosome 27 ($3n$), chromosome 36 ($4n$) ... 81 chromosomes ($9n$).



B) allopolyploidy repeated many times different chromosome sets. For example, soft wheat - allopolyploid which contains chromosome sets of 6 different wheat species. Allopolyploid in nature is rare. Interspecific hybrids are formed. Polyploidy is needed to interspecific hybrids could produce offspring. So breeder Karpechenko created the prolific hybrid of cabbage and radish.



2. Geteroploidiya (aneuploidy) is the change in the number of chromosomes, not multiple haploid set.

The result is a phenomenon:

- Trisomy ($2n + 1$), for example, trisomy 21 by a pair of chromosomes - Down's syndrome
- Monosomy ($2n-1$);

Geteroploidiya usually accompanied by serious hereditary ano-maliyami often incompatible with life.

Chromosomal mutations are changes in chromosome structure.

There are intrachromosomal and interchromosomal aberration.

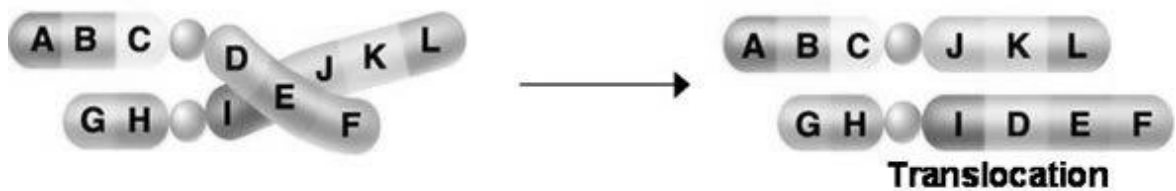
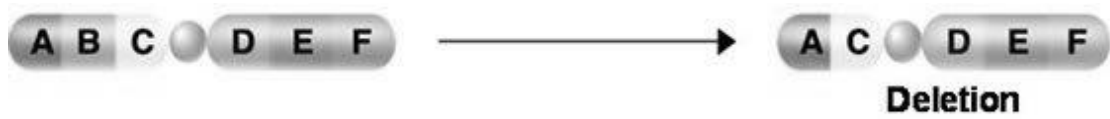
Intrachromosomal adjustment:

1. Duplication - repetition chromosome region.
2. Deletion - loss of chromosome region.
3. Inverse - twist chromosome region 180 degrees.

Interchromosomal adjustment:

1. Translocation - the gap chromosome region and its connection to other non-homologous chromosome.

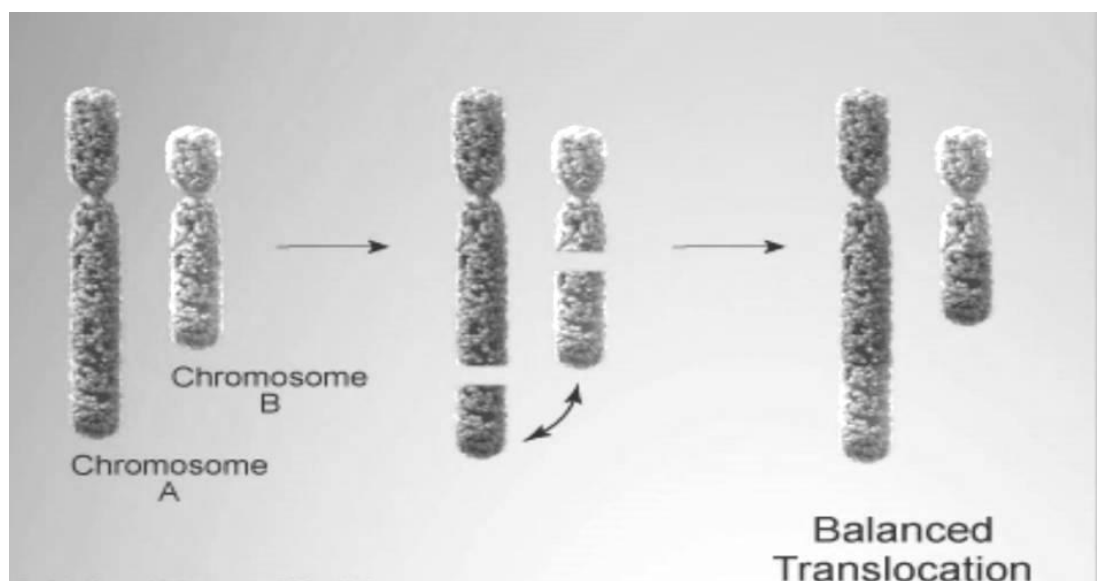
2. Transposition - gap chromosome region and joining it to another location of the same chromosome.



Translocations and Inversions

Translocation:

When a chromosome piece breaks off and reattaches to another, nonhomologous chromosome



Inversion:

Chromosome segment breaks off and then reattaches in reverse orientation to the same chromosome

Gene mutation mutation is associated with changes in the nucleotide sequence of the gene.

There are two mechanisms:

1. The first mechanism involves a change in the number of nucleotides (duplications, deletions). The composition of coding triplets after space mutation changes. This leads to a change in the amino acid composition of the protein and protein synthesis with completely different properties
2. The second mechanism is the substitution of one nucleotide for another. It alters the composition of only one triplet that may to change only a single amino acid in the protein. Substitution of one amino acid is not always essential influence on the change of properties of the protein (particularly if it is close to the properties of the original).

Gene mutations play a large role in evolution. They create a reserve of genetic variation. Since most mutations recessive in the heterozygote state for a long time can not manifest itself. This is very important, since a change in environmental conditions, the mutation may be useful and save the species from extinction.

Significance of Mutations

- Most mutations are neutral (they have little or no effect)

Mutations that cause dramatic changes in protein structure or gene activity can be very harmful

- Mutations are a source of genetic variability in a species

Polyploidy - a mutation where an organism has an extra set of chromosomes

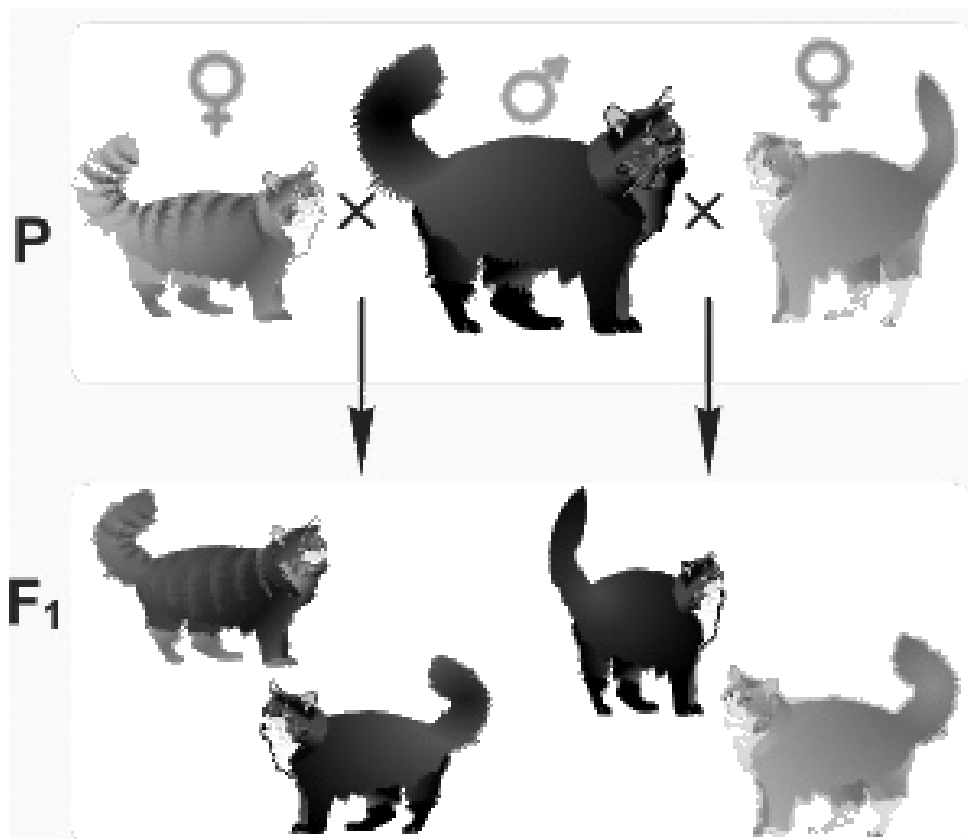
Polyploid plants are often larger and stronger

COMBINATIVE VARIABILITY

Combinative variability is not related to changes in the genes, but only with their recombination in the offspring. It occurs during sexual reproduction.

Causes:

- Crossing over
- Independent divergence of homologous chromosomes in anaphase I of meiosis, which occurs during the formation of gametes.
- The phenomenon of random fertilization.



Value:

- combinative variability increases the genetic diversity of the offspring;
- combinative variability underlies hybridological method in genetics and produces organisms with combinations of traits necessary to man.

Testing your knowledge

Task 1. Emphasize mistakes in the sentences (if any) and correct them:

1. Meiosis is an indirect cell division in which one diploid mother cell forms 4 haploid daughter cells, the genetic material which is the same as the parent cells.
2. Leptotene is stage of prophase I when DNA despiralized and chromosomes become visible in the form of thin fibers.
3. Each bivalent consists of 2 homologous chromosomes (2 chromatids (DNA)).
4. Formation of bivalents occurs at pachytene.
5. Crossing over is the exchange of parts between bivalents.
6. Chiasmata become visible in diplotene.
7. Bivalents line up on the equator of the cell in metaphase II.
8. The microtubules are attached to kinetochores only on one side of each centromere in anaphase I.
9. The independent assortment of chromosomes occurs in anaphase II.
10. The nuclear membrane reforms around each daughter nucleus in prophase.
11. Spindle fibers bind to both sides of the centromeres of chromosomes in telophase II.
12. Cytokinesis occurs in anaphase of mitosis.
13. The nuclear membrane disintegrates in metaphase II of meiosis.
14. You can see the bivalents in the cell in one of the phases of mitosis.
15. A chromosome is divided into two chromatids that move to different poles in metaphase of mitosis.

Task 2. Select only one correct answer:

1. Heritable variability is divided into

- a) mutational and combinative
 - b) mutational and modificational
 - c) phenotypic and **genotypic**
 - d) combinative and nonhereditary
2. Modificational variability is variability, affecting
- a) only the phenotype and not affecting the genotype
 - b) only the genotype and not affecting the phenotype
 - c) genotype and phenotype
 - d) neither genotype nor phenotype
3. Wears group character as similar changes occur in a group of individuals
- a) mutational variability
 - b) modificational variability
 - c) genotypic variability
 - d) combinative variability
4. The boundaries of this variability is called the norm of reaction and defined genotype
- a) mutational variability
 - b) modificational variability
 - c) genotypic variability
 - d) combinative variability
5. This variability is not related to changes in the genes, but only with their recombination in the offspring
- a) mutational variability
 - b) modificational variability
 - c) genotypic variability
 - d) combinative variability
6. The reason of combinative variability is
- a) crossing over
 - b) changes in genes

- c) changes in chromosomes
- d) modification of the phenotype

7. The changes occurring in organisms do not usually wear an adaptive character during

- a) mutational variability
- b) modificational variability
- c) phenotypic variability
- d) nonhereditary variability

8. Mutations associated with changes in the genotype are

- a) lethal, half-lethal, neutral
- b) genetic, chromosomal, genomic
- c) somatic and generative
- d) dominant and recessive

9. Genome mutations are associated

- a) with a change in the structure of gene
- b) with a change in the number of chromosomes in the genome
- c) with a change in the structure of chromosome
- d) with a loss of chromosome plot

10. There are 2 types of genome mutations

- a) aneuploidy and euploidy
- b) deletion and duplication
- c) polyploidy and duplication
- d) inversion and translocation

11. Trisomy refers to mutations

- a) genetic
- b) chromosomal
- c) genomic
- d) somatic

12. Deletion associated with

- a) repetition chromosome region
- b) loss of chromosome region
- c) twist chromosome region 180 degrees
- d) change in the number of chromosomes

13. Translocation refers to mutations

- a) genetic
- b) chromosomal
- c) genomic
- d) somatic

14. Duplication associated with

- a) repetition chromosome region
- b) loss of chromosome region
- c) twist chromosome region 180 degrees
- d) change in the number of chromosomes

15. Inverse associated with

- a) repetition chromosome region
- b) loss of chromosome region
- c) twist chromosome region 180 degrees
- d) change in the number of chromosomes

16. Change in the number of nucleotides is typical for mutation

- a) gene
- b) chromosomal
- c) genomic
- d) polyploidy

17. Somatic mutation occurs in

- a) gametes
- b) normal cells of the body
- c) in sperm
- d) the egg

18. Generative mutation occurs in

- a) gametes
- b) normal cells of the body
- c) brain cells
- d) blood cells

19. The mutations differ in their localization in the cell

- a) nuclear
- b) dominant
- c) neutral
- d) genomic

20. The mutations occurring in the DNA of mitochondria and plastids

- a) nuclear
- b) cytoplasmic
- c) chromosomal
- d) genomic

Task 3. What is the type of mutation, shown in the pictures:



Task 4. Complete the table

Kind of mutation	General characteristics	Importance in nature
Combinative variability		
Mutational variability		
Modification variability		

Task 5. Give the wording of the following terms: polyploidy, modification variability, inversion, translocation, the rate of reaction.

List of recommended literature:

1. Cambell N.A. Biology / N.A. Cambell et al. // Benjamin Cummings, 2013. – p. 1484.
2. Raven P.H. Biology / P.H. Raven, G.B. Johnson, K.A. Mason // MGH, 2002. – p. 1239.