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List of practical skills for the discipline

Discipline: "Genes and Heredity" (Medical Genetics)

Discipline code: GN 1204

EP: 6B10115 "Medicine"

Volume of study hours\ credits: 120 hours/4 credits

Course and semester of study: 1/2

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Protocol no. __18_ of «_13__» ___06_____ 2023 y.

Topic №1. Fundamentals of medical genetics

I. Draw up a pedigree according to the given family description. Analyse the pedigree. Determine:

1. whether the given trait is hereditary (by its manifestation in relatives),
2. type and character of inheritance (autosomal dominant, autosomal recessive, dominant X-linked, recessive X-linked, Y-linked),
3. the zygosity of individuals in the pedigree (homo- or heterozygosity),
4. probability of giving birth to a child with hereditary pathology (genetic risk).

1. Proband has a white curl in the hair above the forehead Proband's brother has no curl. No anomaly noted on the line of Proband's father. Proband's mother has a white curl. She has three sisters. Two sisters with a curl, one without a curl. One of the proband's aunts on her mother's side has a son with a curl and a daughter without a curl. The second has a son and daughter with a curl and a daughter without a curl. The third aunt of the proband on the mother's side without a curl has two sons and one daughter without a curl. Proband's grandfather on his mother's side and two of his brothers had white curls and two others were curlless. The great-grandfather and great-great-grandfather also had a white curl above the forehead.

Determine the probability of having children with a white curl above the forehead if Proband marries his cousin who has this curl.

2. The newlyweds have normal right handedness. A woman's family had two sisters who are normally right-handed and three brothers who are left-handed. The woman's mother is right-handed and her father is left-handed. The father has a left-handed sister and brother, and a right-handed sister and two brothers. The grandfather on the father's side is right-handed and the grandmother is left-handed. The woman's mother has two brothers and a sister, all right-handed. The husband's mother is right-handed and the father is left-handed. The grandparents on the mother's side and the husband's father's side are normally right-handed.

Determine the probability of having left-handed children in this family.

3. Proband, a healthy woman, has two healthy brothers and two brothers with alkaptonuria. Proband's mother is healthy and has two healthy brothers. Proband's father has alkaptonuria and is his wife's great uncle. His grandmother on his father's side was sick and was married to his healthy cousin. Proband's maternal grandparents are healthy, grandfather's father and mother are also healthy, and grandfather's mother is a sibling of proband's grandfather on his father's side.

Determine the probability of having children with alkaptonuria in the proband's family if she marries a healthy man whose mother had alkaptonuria.

4. Proband, a normal woman, has five sisters, two of whom are identical twins and two of whom are identical twins. All sisters have six fingers on their hands. The proband's mother is normal, the father is six-toed. On the mother's side, all ancestors are normal. The father has two brothers and four sisters, all five-toed. The grandmother on the father's side is six-toed. She had two six-toed sisters and one five-toed sister. The grandfather on the father's side and all his relatives are normally five-toed. Determine the probability of the proband having six-toed children in her family if she marries a normal man.

5. Proband is a healthy woman. Her sister is also healthy and her two brothers are color blind. Proband's mother and father are healthy. Four sisters of the proband's mother are healthy, their husbands are also healthy, About cousins on the side of the proband's mother we know: in one family one sick brother,

two sisters and a brother are healthy; in two other families one sick brother and one healthy sister each; in the fourth family one healthy sister. The grandmother of the proband's mother's side is healthy, the grandfather suffered from color blindness. On the side of the proband's father there are no patients with color blindness.

Determine the probability of the proband having children with color blindness if she marries a healthy man.

6. Proband suffers from a mild form of sickle cell anemia. His spouse is healthy. She has a daughter also with a mild form of anemia. Proband's mother and grandmother suffered from the same form of sickle cell anemia, the other sibs of the mother and her father are healthy. The proband's wife has a sister suffering from a mild form of anemia, the other sister died of anemia. The mother and father of the proband's wife suffered from anemia, in addition, it is known that the father had two brothers and a sister with a mild form of anemia and that in the family of the father's sister two children died of sickle cell anemia.

Determine the probability of having children with severe anemia in the family of the proband's daughter if she marries a man like her father.

7. Proband and his five brothers are healthy. Proband's mother and father are deaf-mute. Two uncles and an aunt on the father's side are also deaf-mute, on the mother's side four aunts and an uncle are healthy and one aunt and one uncle is deaf-mute. The maternal grandparents are healthy. The paternal grandparents are deaf-mute. Paternal grandmother has a deaf-mute brother and two deaf-mute sisters. Paternal grandfather has two brothers, one of whom is healthy and one of whom is deaf-mute, and five sisters, two of whom are deaf-mute. Grandfather's mother and father on his father's side are healthy, and grandmother's mother and father on his father's side is deaf-mute.

Determine the probability of deaf-mute children being born in the proband's family if he marries a woman who is normal with respect to deaf-muteism and comes from a family that is healthy with respect to deaf-muteism.

8. Proband is healthy. The proband's father has epidermolysis bullnose. The mother and her relatives are healthy. Two sisters of the proband are healthy, one brother is ill. Three uncles on the father's side and their children are healthy, and three uncles and one aunt are sick. One sick uncle from the first marriage has a sick son and a healthy daughter, and one sick uncle from the second marriage has a sick daughter and son. A second sick uncle has two healthy daughters and a sick son. The third sick uncle has two sick sons and two sick daughters. The paternal grandmother is sick and the grandfather is healthy, three sisters and two brothers of the grandmother were healthy.

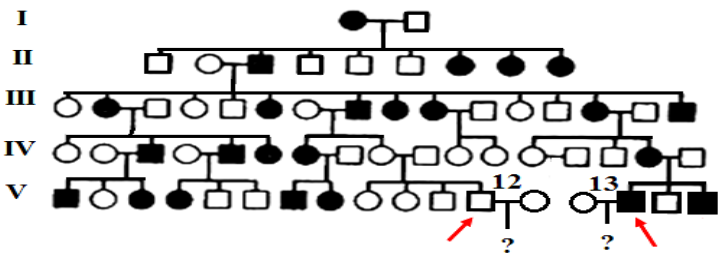
Determine the probability of having sick children in the proband's family if he marries a healthy woman

9. Proband has teeth that are normal in colour. His sister has brown teeth. Proband's mother's teeth are brown and his father's teeth are normal in colour. Seven of the proband's mother's sisters have brown teeth and four brothers have normal teeth. One aunt of the proband's mother's side, who has brown teeth, is married to a man with normal teeth. They have three children: a daughter and son with brown teeth and a daughter with normal teeth. Two uncles of the proband on the mother's line are married to women without abnormality in tooth colouration. One of them has two sons and a daughter, the other has two daughters and a son. All of them with normal teeth. The proband's grandfather on his mother's side had brown teeth, and his grandmother on his mother's side had normal teeth. Two brothers of the grandfather on the maternal side with normal coloured teeth. The great-grandmother (the mother of the grandfather on his mother's side) and

the great-great-grandmother (the mother of this great-grandmother.) had brown teeth, and their husbands had normal-coloured teeth.
Determine what kind of children the proband might have if he marries a woman who is heterozygous for this trait.
10. Proband is a woman with cerebellar ataxia. Her spouse is healthy. They have six sons and three daughters. One son and one daughter have cerebellar ataxia, the other children are healthy. Proband has a healthy sister and three sick brothers. The healthy sister is married to a healthy man and has a healthy daughter. Proband's three sick brothers are married to healthy women. One brother's family has two healthy sons and one healthy daughter, the second brother's family has a healthy son and a sick daughter, and the third brother's family has two sons and three daughters, all healthy. The father of the proband is sick and the mother is healthy.
What is the probability of having sick children in the sick Daughter of the proband if she marries a healthy man?
11. Proband is sick with congenital cataract. He is married to a healthy woman and has a sick daughter and a healthy son. Proband's father has cataracts and his mother is healthy. The proband's mother has a healthy sister and healthy parents. The grandfather on the father's side is sick and the grandmother is healthy. Proband has a healthy aunt and uncle on her father's side. The uncle is married to a healthy woman. Their three sons (Proband's cousins on his father's side) are healthy.
What is the probability of the proband's daughter having sick grandchildren if she marries a man heterozygous for this type of cataract?
12. Proband suffers from a nail and patella defect, while his brother is normal. Proband's father had the syndrome and his mother was healthy. Proband's grandfather on his father's side has the syndrome and his grandmother is healthy. The proband's father has three brothers and four sisters, of whom two brothers and two sisters have nail and patella defect syndrome. The sick uncle on his father's side is married to a healthy woman and has two daughters and a son. All of them are healthy. Determine the probability of having children with the disease in the proband's family if his spouse does not have nail and patella defects.

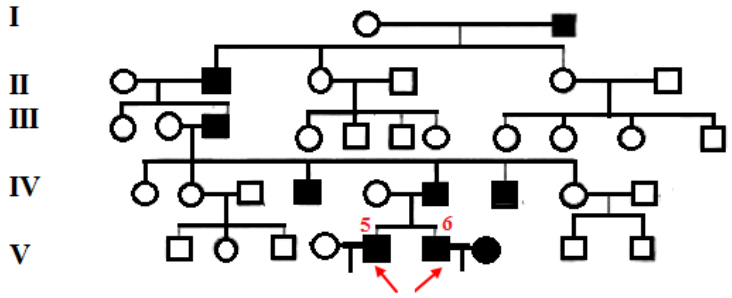
II.Carry out the analysis of family trees.
Determine:
1. whether the given trait is hereditary (by its manifestation in relatives),
2. arguing, type and character of inheritance (autosomal dominant, autosomal recessive, dominant X-linked, recessive X-linked, Y-linked),
3. zygosity of individuals in the pedigree (homo- or heterozygosity),
4. probability of giving birth to a child with an inherited pathology (genetic risk).
Give a full description of the type of inheritance established for your pedigree
Calculate the genetic risk of having children with this abnormality in the family of proband 12 from generation V with a healthy woman; in the family of proband 13 from the same generation with a healthy woman.

II.1 Analyse the family trees. A) Determine:



1. whether the trait is inherited (by its manifestation in relatives),
2. arguing the type and nature of inheritance (autosomal dominant, autosomal recessive, dominant X-linked, recessive X-linked, Y-linked),
3. zygosity of individuals in the pedigree (homo- or heterozygosity),
4. probability of giving birth to a child with hereditary pathology (genetic risk).

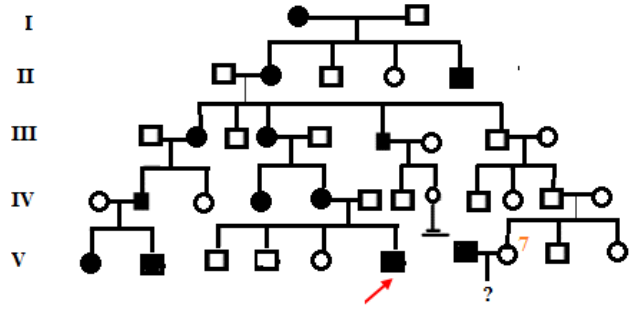
B) Give a full description of the type of inheritance established for your lineage



Calculate the genetic risk of children with this anomaly in the family of proband 5 from V generation with a healthy woman; in the family of proband 6 from the same generation with a sick woman.

II.2 Analyse the pedigrees.
Determine:
1. whether the given trait is hereditary (by its manifestation in relatives),
2. arguing, type and character of inheritance (autosomal dominant, autosomal recessive, dominant X-linked, recessive X-linked, Y-linked),
3. zygosity of individuals in the pedigree (homo- or heterozygosity),
4. probability of giving birth to a child with hereditary pathology (genetic risk).

A) Give a full description of the type of inheritance established for your lineage

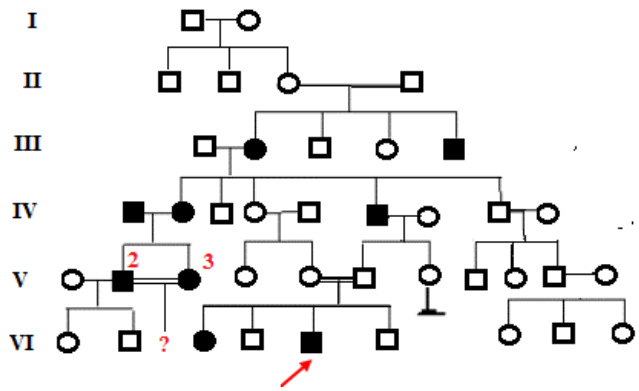


Calculate the genetic risk of having children with this anomaly in the family of proband 7's sibs from generation V with a sick male.

II.3 Analyse the pedigrees.

Determine:
1. whether the given trait is hereditary (by its manifestation in relatives),
2. arguing, type and character of inheritance (autosomal dominant, autosomal recessive, dominant X-linked, recessive X-linked, Y-linked),
3. zygosity of individuals in the pedigree (homo- or heterozygosity),
4. probability of giving birth to a child with hereditary pathology (genetic risk).

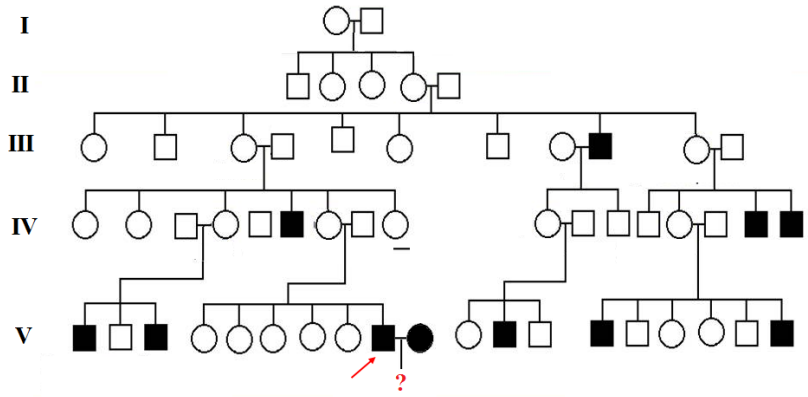
A) Give a full description of the type of inheritance established for your pedigree



Calculate the genetic risk of children with this anomaly in the family of sibs 2 and 3 from V generation.

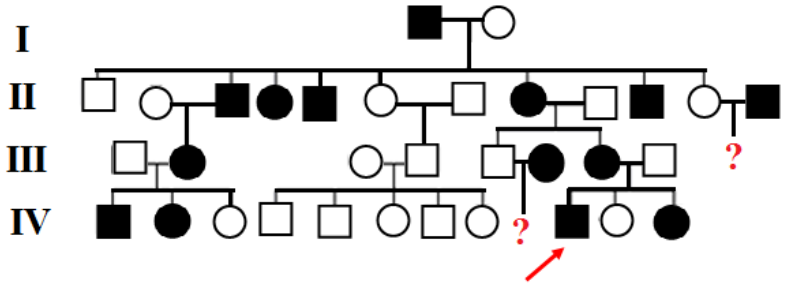
II.4 Analyse the pedigrees.
Determine:
1. whether this trait is hereditary (by its manifestation in relatives),
2. arguing, type and character of inheritance (autosomal dominant, autosomal recessive, dominant X-linked, recessive X-linked, Y-linked),
3. zygosity of individuals in the pedigree (homo- or heterozygosity),
4. probability of giving birth to a child with hereditary pathology (genetic risk).

A) Give a full description of the type of inheritance established for your pedigree



Calculate the genetic risk of children with this anomaly in the family of the proband from the V generation with a sick woman.

II.5 Analyse the pedigrees.
Determine:
1. whether this trait is hereditary (by its manifestation in relatives),
2. arguing, type and character of inheritance (autosomal dominant, autosomal recessive, dominant X-linked, recessive X-linked, Y-linked),
3. zygosity of individuals in the pedigree (homo- or heterozygosity),
4. probability of giving birth to a child with hereditary pathology (genetic risk).
A) Give a full description of the type of inheritance established for your pedigree



Calculate the genetic risk of having children with this anomaly in a family of a second-generation daughter with a male patient and a third-generation son with a female patient.

Topic №2. Fundamentals of general genetics.
Inheritance of mendelating traits. Mono- and dihybrid crossing
I. Monohybrid crossing:
1. In a human, brown-eyedness is a dominant trait and blue-eyedness is recessive. One spouse is heterozygous for brown-eyedness and the other is blue-eyed. What offspring can be expected from this marriage
.2. In a human being, provent-handedness is dominant over left-handedness. Both the spouses are right handed heterozygotes. Can we expect a left-handed child to be born from this marriage
3. Polydactyly hexapalatia in humans is a dominant trait and normal hand structure pentapalatia is a recessive trait. From the marriage of a heterozygous six-toed man with a woman who has normal hand structure, two children were born: a five-toed child and a six-toed child, What is the genotype of these children?
4. Hemifacial microsomia is accompanied by unilateral anomaly of the auricle with underdevelopment of the mandible on the same side. It is determined by an autosomal dominant gene. What offspring can be expected from the marriage of spouses heterozygous for this pathology?
5. Tooth and nail syndrome is manifested by two features at the same time - disorder of nail and teeth development. Inheritance is of the autosomal dominant type. A man suffering from this disease and

homozygous for the genotype married a healthy woman. One child was born from this marriage. What is his phenotype and genotype?

6. In humans, the dominant gene

causes an abnormality of skeletal development expressed in changes in the bones of the skull and reduction of the clavicles. A woman with normal skeletal structure married a man with this anomaly. The child from this marriage had a normal skeleton. Can the phenotype of the child be used to determine the genotype of the father?

7. Eye-bone and skin syndrome is accompanied by shortening of fingers brachydactyly, lack of teeth adontia, lack of hair growth hypotrichosis, light skin due to lack of pigment - melanin albinism - is inherited by autosomal recessive type. A child with this disease was born to healthy spouses. Determine the genotypes of the parents.

8. Familial myoplegia recurrent limb paralysis is due to an autosomal dominant gene. A husband and wife are heterozygous for the myoplegia gene. Determine the probability of having a sick child.

9. Absence of small molars is inherited as a dominant autosomal trait. What is the probability of birth of children with this anomaly, if one of the spouses in the family suffers from the absence of small molars, it is known that his mother and brother are healthy. The other spouse is healthy.

10. Ptosis - drooping of the upper eyelid - is inherited by recessive type. A child with ptosis is born to healthy parents in a family. Determine the probability of the next child being born with the condition.

11. Galactosemia inability to digest milk sugar is inherited by autosomal recessive type. What is the probability of giving birth to sick children in a family where one of the spouses suffers from the analysed disease, the other is healthy, and his parents, brothers and sisters were healthy?

12. Achondroplasia dwarfism, a severe shortening of the limb skeleton, is transmitted as a dominant autosomal trait. In a family where both spouses have achondroplasia, a healthy child is born. What is the probability that the next child will also be healthy?

13. Parahaemophilia, a tendency to skin and nose bleeding, is inherited as an autosomal recessive trait. What is the probability of having sick children in a family where both spouses have parahaemophilia?

14. Myoplegia recurrent paralyses associated with loss of potassium by muscle cells. The disease manifests itself at the age of 20-40 years, is inherited by autosomal dominant type. Determine the probability of having children with anomalies in a family where the father is heterozygous and the mother does not suffer from myoplegia.

15. Galactosemia - inability to use galactose due to reduced enzyme activity - is expressed by a complex of signs: jaundice, emaciation, liver cirrhosis, cataracts, dementia, etc. It is inherited as an autosomal recessive trait. What's the probability of having sick children in a family where one of the spouses one of the spouses is homozygous for the galactosemia gene, but the development of the disease is prevented by diet, and the other is heterozygous for galactosemia.

16. Deafblindness is inherited in an autosomal recessive manner. A deaf-mute child was born from the marriage of a deaf-mute woman to a normal man. Determine the genotypes of the parents.

17. Albinism is inherited as an autosomal recessive trait. In a family where one spouse is albino and the other is normal, identical twins are born, one of whom is normal for the trait being analysed and the other is albino. What is the probability of the next child being born an albino?

18. Hepato-cerebral dystrophy, or Wilson's disease, is associated with impaired synthesis of the protein ceruloplasmin. Causes cirrhosis of the liver changes in brain tissue, impaired transport of substances in the renal tubules. Wilson's disease develops at the age of 10-15 years, inherited by autosomal recessive type. What is the probability of having sick children in a family where one spouse has the disease and the other is healthy and has healthy parents?

II. Dihybrid crossbreeding

1. In humans, right-handedness dominates over left-handedness. A blue-eyed right-handed person married a brown-eyed right-handed person. Two children were born to them: a brown-eyed left-handed person and a blue-eyed right-handed person. Determine the probability of this family having blue-eyed children who are predominantly left-handed.

2. A family with brown-eyed parents has four children. Two blue-eyed children have blood types I and IV, while two brown-eyed children have blood types II and III. Determine the probability of the next child being born brown-eyed with blood group I, if it is known that brown eye colour is dominant over blue and is due to an autosomal gene.

3. In a family where the parents heard well and had one smooth hair and the other curly hair, a deaf child with smooth hair was born. Their second child heard well and had curly hair. What is the probability of further deaf children with curly hair in the family if it is known that the curly hair gene is dominant over the smooth hair gene, deafness is a recessive trait, and both pairs of genes are on different chromosomes.

4. Adult glaucoma is inherited in several ways. One form is determined by a dominant autosomal gene, the other is determined by a recessive autosomal unlinked gene. What is the probability of giving birth to a child with an abnormality if both parents are heterozygous for both pairs of abnormal genes?

5. Shoulder - scapular - facial myopathy muscle atrophy and myopia are inherited as autosomal dominant traits. The mother and father are heterozygous for both traits. What is the probability of having healthy children in this family?

6. A man with normal vision and freckles, his father not having freckles and his mother having freckles, marries a woman without freckles but suffering from myopia. She has several generations of relatives suffering from myopia. What children should be expected to be born to this couple?

7. Hirschprung's disease is inherited autosomal recessively and is characterised by impaired innervation and functioning of the large intestine. Cystic fibrosis is inherited autosomal recessively and is characterised by a deficiency in the functioning of the pancreas. Determine the probability of having a healthy child and a child with both pathologies in a family where the wife has Hirschsprung's disease and is heterozygous for cystic fibrosis and the husband has cystic fibrosis, his father is healthy and his mother has Hirschsprung's disease.

8. Galactosemia (inability to digest milk sugar) is inherited as an autosomal dominant trait, absence of small molars as a dominant autosomal trait. The wife was treated for galactosaemia and was phenotypically healthy, but she was missing small molars.
The husband is healthy. A child was born to them with galactosaemia; there is no dental developmental abnormality. Determine the probability of having a child with two anomalies.

9. Rhesus-positivity is a dominant autosomal trait, while albinism is inherited autosomal recessively. Both parents are rhesus - positive with normal pigmentation. They have an albino child with Rh negative (recessive trait) blood type. Determine the genotypes of the parents and their possible offspring?

10. Albinism (tyrosinase inactivation) and phenylketonuria are inherited autosomal recessively. A healthy woman heterozygous for phenylketonuria, her mother had albinism and her father is heterozygous for

phenylketonuria, marries a man of the same genotype as herself. What children are possible from this marriage?

III. Interaction of allelic and non-allelic genes. Incomplete dominance in mono- and dihybrid crosses

1. In humans, a large nose size is a dominant trait, a small nose is a recessive trait, and a normal medium-sized nose is shown in heterozygotes. One spouse has a large nose and the other has a medium-sized nose. Determine the probability that their children will have: a) a small nose; b) a medium-sized nose; c) a large nose.

2. In humans, curly hair is a dominant trait and straight (smooth) hair is a recessive trait. Heterozygotes have wavy hair. What type of hair can the children have, and with what probability, if both parents have wavy hair?
3. One form of cystinuria (a disorder of metabolism of one of amino acids) is inherited as an autosomal trait with incomplete dominance; heterozygotes have only elevated levels of cystine in the urine, while homozygotes have cystine kidney stones. Identify the possible forms of cystinuria in children in the family:
a) where one spouse suffered from kidney stone disease and the other had only elevated urinary cystine content;
b) where one spouse had renal stone disease and the other spouse was normal with respect to the analysed trait.

4 Thalassaemia is inherited as an incompletely dominant autosomal trait. In homozygotes, the disease is fatal in 90-95% of cases, while in heterozygotes it is relatively mild. What is the probability of having healthy children in a family where both parents have a mild form of thalassaemia?
5. Acatalasia is caused by a rare autosomal recessive gene. Heterozygotes have reduced catalase activity. Determine the probability of having a child in the family without abnormalities if it is known that both parents and their only son have reduced catalase activity.
6. It is known that the gene for long eyelashes is inherited as a dominant autosomal trait, while short eyelashes is recessive. Heterozygotes have normal eyelashes. Brown-eyedness is a dominant autosomal trait, while blue-eyedness is recessive. Determine what offspring can be expected in a family in which the following is known: one parent is brown-eyed and has long eyelashes, and the other parent is blue-eyed and has short eyelashes, and their first child is blue-eyed.
7. Pelger's anomaly of leukocyte nuclear segmentation is inherited as an autosomal incompletely dominant trait. Homozygote for this trait lack nuclear segmentation completely, while heterozygotes have unusual Deafness is an autosomal recessive trait. A deaf child was born to parents with unusual nuclear segmentation. Determine the genotypes of the parents. What kind of offspring can be expected in this family?
8. In humans, the dominant brachydactyly mutation manifests as shortening of the fingers in the heterozygous state, and in the homozygous state it leads to death in the early stages of embryo development. Normal phalangeal length is due to a recessive gene. Normal skin pigmentation is a dominant trait, albinism is recessive. What offspring can be expected in a family where both parents are diheterozygous?

9. Sickle cell anemia is inherited as an incompletely dominant autosomal trait. Homozygous individuals usually die before puberty; heterozygous individuals are viable and their anemia is most often subclinical. Such patients are resistant to malaria because the malarial plasmodium cannot use the hemoglobin of their blood erythrocytes for its nutrition. Polydactyl (six-fingeredness)-autosomal dominant trait, normal hand structure - recessive. A malaria-resistant woman with normal hand structure marries a man who was operated on in childhood for polydactyl and is heterozygous for sickle-cell anemia. What kind of offspring can be expected in this family?

IY. Interaction of non-allelic genes

1. In humans, normal hearing is due to two non-allelic dominant genes that interact on the principle of complementarity. A diheterozygous man married a deaf woman whose deafness is due to the presence of one recessive gene, and she is heterozygous for the other pair of genes. What offspring can be expected from this marriage?
2. Human growth is controlled by several pairs of unlinked genes that interact according to the principle of polymerism. If we ignore environmental factors and conditionally limit ourselves to only three pairs of genes, we can assume that in a population the shortest people have all recessive genes and are 150 cm tall, and the tallest people have all dominant genes and are 180 cm tall.
a) Determine the height of people who are heterozygous for all three pairs of height genes.
b) A short woman married a man of average height. They had four children who were 165 cm, 160 cm, 155 cm, 150 cm tall. Determine the genotypes of the parents and their heights.

3. In humans, differences in skin color are mainly due to two pairs of non-allelic genes B and B1. People with the BBB1B1B1 genotype have black skin, while those with the BBB1B1 genotype have white skin. Different combinations of dominant B and B1 genes provide skin pigmentation of different intensities. What kind of children can there be in a mulatto family?
4. The red coloration of an onion bulb is determined by the dominant gene, yellow by its recessive allele. However, the manifestation of the coloring gene is possible only in the presence of another, unlinked with it dominant gene, recessive allele of which suppresses the coloring and bulbs are white. A red-bulb plant was crossed with a yellow-bulb plant. The offspring produced individuals with red, yellow and white bulbs. Describe and explain this phenomenon.
5. In a pumpkin, the yellow colour of the fruit is determined by a dominant gene, its recessive allele determines the green color of the fruit. Another non-allelic gene suppresses the action of these genes and the fruits have white coloring. When a yellow-fruited pumpkin was crossed with a white pumpkin, all the offspring gave white fruits. When the resulting individuals were crossed with each other, the following offspring were obtained; plants with white fruits -204; with yellow fruits -53; with green fruits - 17. Describe and explain this phenomenon.
6. Interferon synthesis in humans depends on two genes, one on chromosome 2 and the other on chromosome 5. Name the form of interaction between these genes and determine the probability of having a child unable to synthesise interferon in a family where both spouses are heterozygous for these genes.
7. The flowers of the sweet pea can be white or red. When two plants with white flowers were crossed, all the offspring turned out to have red flowers. When the offspring were crossed with each other, the plants with red and white flowers turned out to have a ratio of 9:7. Describe and state the type of interaction between non-allelic genes.

Y. Multiple alleles. Inheritance of blood groups in monohybrid and dihybrid crosses

1. A mother with blood type II has a child with blood type I. Identify the possible blood types of the father.
2. A mother has blood group I and the father has blood group III. Can children inherit their parents' blood types?
3. Two boys were mixed up in a maternity hospital. The parents of one of them have blood groups I and II, the other has blood groups II and IV. An examination shows that the children have blood types I and IV. Determine who is whose son?
4. A woman with blood type II had a child with blood type I. Determine the possible genotype of the father. Can a man with blood type IV be the father of the child?
5. In a family where the wife has ABO blood group I and the husband has IV blood group, a deaf child was born. Determine the probability of birth of a healthy child and its possible blood groups, if it is known that deafness is a recessive trait, and normal hearing is dominant.
6. A family of brown-eyed parents has four children. Two blue-eyed children have blood groups I and IV, two brown-eyed children have blood groups III and II. Determine the probability of the next child being born to a brown-eyed person with blood type I. Brown eye color is dominant over blue eye color. The gene for iris pigmentation is located in the autosomes.

Topic №3. Fundamentals of general genetics. Linked inheritance.

I. Linked inheritance. Inheritance of sex and sex-linked traits

1. Anhydrosis ectodermal dysplasia in humans is transmitted as an X-linked recessive trait. A normal woman marries a man with anhidrosis ectodermal dysplasia. They give birth to a sick girl and a healthy son. Determine the probability of the next child being born without the abnormality.

2. In humans, classical hemophilia is inherited as an X-linked, recessive trait. Albinism is due to an autosomal recessive gene. A couple normal for these traits have a son with both abnormalities. What is the probability that a second son in this family will show these same abnormalities at the same time?

3. A brown-eyed woman with normal vision, whose father had blue eyes and suffered from color blindness, marries a blue-eyed man with normal vision. What offspring can be expected from this couple if it is known that the gene for brown eyes is dominant and located in autosomes, and the gene for color blindness is recessive and linked to the X chromosome?

4. In a family where the wife has ABO blood group I and the husband IV, a child with blood group III color blindness is born. Both parents distinguish colors normally. Determine the probability of giving birth to a healthy son and his possible blood types. Daltonism is inherited as a recessive, X-linked trait.

5. Hypertrichosis is inherited as a U-linked trait that does not manifest itself until age 17. One form of ichthyosis is inherited as a recessive, X-linked trait. In a family where the woman is normal in both features and the husband has only hypertrichosis, a boy with signs of ichthyosis is born, a) determine the probability of hypertrichosis in this boy, b) determine the probability of birth in this family of children without both abnormalities, and what gender will they be?

6. Cataracts and polydactyl in humans are caused by dominant autosomal closely linked (i.e., no crosslinking) genes. However, not necessarily the genes for these anomalies, but also the cataract gene with the gene for normal hand structure and vice versa can be linked. A woman inherits cataract from her mother and polydactyl from her father. Her husband is normal for both traits. What is more likely to be expected in their children: the simultaneous occurrence of cataract and polydactyl, the absence of both traits, and the presence of only one abnormality, cataract or polydactyl?

7. One type of deafness is inherited recessively, X-linked. A deaf boy was born in a family where the husband is sick and has blood group II according to the ABO system (it is known that his mother had blood group I), and the wife is healthy and has blood group IV. Determine the genotypes of the parents and the son. What blood group the boy may have.

8. The genes for ocular albinism and sensor neural deafness are localized on the X chromosome, closely linked, and inherited recessively. The husband is healthy, the wife is also healthy, but she inherited the gene for ocular albinism from her father and the gene for sensor neural deafness from her mother. What children and with what probability can be expected in this family?

9. The genes for hemophilia (h) and color blindness (d) are localized in the X - chromosome at a distance of about 10 morganids. A healthy woman who inherited hemophilia from her mother and color blindness from her father marries a healthy man. What offspring can be expected from this marriage?

10. The genes for color blindness (color blindness - d) and night blindness (s) are linked to the X chromosome and are 50 morganids apart. A girl with normal vision whose father suffered from two forms of blindness gets married. Determine the probability of possible phenotypes in the children in this family if the young man is healthy?

11. How many types of gametes does an organism with the genotype "AaBb" form when:
a) complete coupling of genes?
b) incomplete coupling of genes?

12. Enamel hypoplasia is inherited as an X-linked dominant trait. A son with normal teeth was born to parents with this condition. What kind of offspring can be expected in this family?

13. Hypertrichosis is transmitted through the U chromosome and polydactyl as a dominant autosomal trait. In a family where the father had hypertrichosis and the mother had polydactyl, a daughter normal for both traits was born. What kind of offspring can be expected in this family?

14. Oculo-cerebrorenal syndrome is X-linked recessively inherited, characterized by muscular hypotonia, mental retardation and cataracts. Can children with this syndrome be born in a family where the husband is sick and the wife and all her relatives are healthy?

Topic №5. Congenital malformations of development.

I. Answer the test questions:

1. X-linked hereditary diseases are ....
| Haemophilia, hypertrichosis, ichthyosis.
|Klinefelter's syndrome.
|Down syndrome.
|Shereshevsky-Turner syndrome.
|Haemophilia, haemophilia.
2. Hereditary diseases transmitted with a Y chromosome ....
|hypertrichosis, ichthyosis, tympanic membranes on fingers.
|hypertrichosis, Down syndrome and Patau syndrome.
|Klinefelter's syndrome, Down's syndrome, Patau's syndrome.
|Ichthyosis, Edwards syndrome, Down syndrome.
|Haemophilia, color blindness.
2. Hereditary diseases associated with changes in the number of sex chromosomes are ....
| syndrome, Klinefelter, Schereshevsky-Turner, trisomy-X.
|Haemophilia, Patau syndrome, Down syndrome.
|Edwards Down syndrome, hypertrichosis.
|Patau Edwards Down syndrome.
|Ichthyosis, ichthyosis.
3. Hereditary diseases associated with altered autosome number ....
|Patau syndrome, hemophilia.
|Down syndrome, Patau--
|Klinefelter syndrome, phenylketonuria, albinism.
|Down syndrome, Patau, trisomy X, YYY.
|Tay-Sachs disease, Lejeune's disease, Edwards syndrome, colour blindness.
4. Hereditary diseases associated with disorders of protein metabolism ...
|Haemophilia, ichthyosis, Patau's syndrome.
|Hypertrichosis, alcanthonuria, Klinefelter's.
|Haemophilia, phenylketonuria, glycogenemia.
|Tyrosinosis, Down's syndrome, albinism.
| albinism, phenylketonuria.
5. Deletion of the short arm in the 5th pair of chromosomes in humans results in the formation of syndrome ...
|Cat's Cry.
|Down's.
|Albinism.
|Alkaptonuria.
|Hemophilia.
6. The term "sibs" in pedigrees refers to ....
|siblings.

|The person from whom the pedigree begins.
|A proband.
|The grandparents of the proband.
|The proband's uncles and aunts.

7. A patient with Down syndrome has...chromosomes.
|2п + 2
|2п - 1
|3р + 1р
|1п
|2п + 1

8. The occurrence of Patau syndrome is associated with the ... chromosome.
|15
|21
|13
|8
|18

9. Inbreeding is ...
|hybridisation.
| cross-pollination or fertilisation.
|random crossbreeding, panmixia.
| free crossing, panmixia.
| inbreeding to increase homozygosity.

10. Thalassaemia (a form of anaemia) is inherited in an autosomal dominant pattern and has two forms, mild (Aa) and severe (AA). The occurrence of the mild form of thalassaemia is due to ... gene.
| non-complete dominance+
| over dominance
|complete dominance
|codominance
|pleiotropy

11. The development of Edwards syndrome is associated with trisomy of the ... chromosome.
|17
|3
|21
|18
|14

12. The karyotype of a person with Klinefelter syndrome is ... chromosomes.
|45
|46
|47
|23
|44

13. Monosomy is characteristic of the genotypes of ....
|XO, YO.
|XXX, XXX, XXY.
|XXX, XY.

|XXXO, HYO.
|XXYY, XYYY.
14. The karyotype of Shereshevsky-Turner syndrome has ... chromosomes.
|47
|44
|46
|27
|45
15. An inherited disease of humans resulting from changes in chromosome structure is ....
|Cat's cry syndrome.
|Schereshevsky-Turner syndrome.
|Klinefelter's syndrome.
|Haemophilia.
|X trisomy.
16. In phenylketonuria, there is a disorder of ....
|amino acid metabolism.
|carbohydrate metabolism.
|Lipid metabolism.
| mineral metabolism.
| nucleic acid metabolism.
17. Down syndrome is caused by trisomy ... pair of chromosomes.
|21+
|15
|20
|13
|18
18. The syndrome resulting from deletion of the short arm in the 5th pair of chromosomes in humans is ....
|Cat's cry syndrome.
|Down syndrome.
|Albinism.
|Alkaptonuria.
|Hemophilia.
19. Trisomy on chromosome 13 leads to the development of syndrome ...
|Patau.
|Down's.
|Schereshevsky-Turner.
|Edwards.
|Klinefelter's.

II. Situational tasks

Question.1. In a large family of myopic parents, 12 children were born to myopic parents. Three of them were of normal vision and 6 were myopic. How many myopic children will be homozygous for the myopia gene?

Question.2. Three girls were born from the marriage of a woman suffering from hypertension (autosomal dominant trait) with a healthy man. The first daughter had 4 children with hypertension, the second daughter had 3 children with the same disease, and the third daughter had two healthy children. Determine the highest probability that the third daughter will have children with hypertension.

Question.3. In Transcarpathian region among people in a number of generations there are observed Long-term modifications. This form of variability is: a) similar to genocopy; b) reversible change of genotype and phenotype; c) irreversible change of phenotype; d) reversible change of genotype; e) inherited by the type of cytoplasmic heredity.

Question.4. Determine the probability of occurrence of a disease in a family, about which it is known that all relatives on the mother's line who lived up to
70 years old had the disease, and on the father's side all relatives are healthy.

III.Answer the questions?

1.Definition of chromosomal diseases, mechanisms of their occurrence.
2.Definition of diseases with hereditary predisposition, mechanisms of their occurrence.
3.Types of diseases with hereditary predisposition.

Topic 6. Prenatal diagnostics of hereditary diseases.

I. Questions

1. What is prenatal diagnostics?
2. What diseases are detected by biochemical methods?
3. Indications for biochemical testing?
4. Characterize the main steps of molecular genetic methods?
5. Indications for cytogenetic studies?
6. What mandatory indications are included for prenatal diagnosis?
7. Give a brief characterization of methods of prenatal diagnostics:
   - Analyses of parental pedigree?
   - Genetic analysis for parents?
   - Invasive methods of prenatal diagnosis?
   - Non-invasive methods of prenatal diagnosis?
8. What is "screening" and what is the basis of the medical term "screening tests"?
9. What are the main indications for referring a pregnant woman for prenatal diagnosis?
10. Describe the process of preimplantation diagnosis?
11. Basics of prevention of hereditary diseases.
12. Medical and genetic counseling (MGC): what is the definition, what is the task of MGC?

II. Test questions may contain - 1, - 2, - 3 or 4 correct answers.

1. Amniocentesis is:
A) examination of sex chromatin
B) drawing of an idiogram
C) examination of enzyme activity
D) determination of the type of inheritance
E) examination of fetal cells in the amniotic fluid
2. Direct methods of prenatal diagnosis:

A) determination of alpha-fetoprotein
B) twin
C) amniocentesis
D) cordocentesis
E) chorionbiopsy
3. Timing for amniocentesis:
A) 8-12 weeks of pregnancy
B) 5-7 weeks of pregnancy
C) 15-18 weeks of pregnancy
D) end of 1 week of pregnancy
E) 2-4 weeks of pregnancy
4. Invasive methods of prenatal diagnosis include:
A) ULTRASOUND
B) amniocentesis
C) chorionbiopsy
D) electroencephalography
E) cordocentesis
5. Methods used in the diagnosis of human hereditary diseases:
A) biochemical
B) hybridological
C) genealogical
D) cytostatic
E) molecular genetic
6. Methods used in prenatal diagnosis of chromosomal diseases:
A) twin
B) amniocentesis
C) genealogical
D) cytogenetic
E) biochemical
7. The holandric type of inheritance is characterized by:
A) sons get sick
B) daughters are ill
C) the trait is transmitted from the father to all sons
D) the trait is transmitted from the father to all daughters
E) the trait is transmitted from the carrier mother to all sons.
8. Cordocentesis is performed:
A) 18-22 weeks of pregnancy
B) 15-18 weeks of pregnancy
C) to study blood from the umbilical cord
D) to study amniotic fluid with fetal cells
E) for the purpose of prenatal diagnosis of hereditary diseases
9. Screening programs are divided into:
A) mass
B) direct
C) indirect

D) selective
E) invasive
10. Medical and genetic counseling is indicated for:
A) all women under 35 years of age
B) in consanguineous marriages
C) spouses over 50 years of age
D) heterozygous carriers of recessive diseases
E) women over 35 years of age and men over 40 years of age.
11. Non-invasive methods of prenatal diagnosis include:
A) determination of markers in the mother's blood
B) chorionbiopsy
C) ultrasound scanning
D) amniocentesis
E) cordocentesis
12. Prenatal diagnosis by amniocentesis is carried out:
A) at 18-22 weeks of pregnancy
B) at 15-18 weeks of pregnancy
C) to study blood from the umbilical cord
D) to study amniotic fluid with foetal cells
E) for the purpose of prenatal diagnosis of hereditary diseases
13. Indirect methods of prenatal diagnosis of hereditary diseases include:
A) determination of albumin concentration in the mother's blood
B) determination of maternal blood sugar concentration
C) determination of alpha-fetoprotein concentration in maternal blood
D) determination of maternal estriol in blood
E) determination of maternal blood nitrogen
14. Diseases with hereditary predisposition include:
A) haemophilia
B) haemoglobinopathy
C) diabetes mellitus
D) phenylketonuria
E) schizophrenia
15. The interaction of genetic and harmful environmental factors leads to the development of diseases:
A) schizophrenia
B) chicken blindness
C) coronary heart disease
D) diabetes mellitus
D) haemophilia
16. Medical and genetic counselling is:
A) a method of prevention of hereditary diseases
B) a method of preventing infectious diseases
C) a method of treatment of hereditary diseases
D) a method of early detection of hereditary diseases
E) a method of early detection of parasitic diseases
17. Indications for medical and genetic counselling:

A) birth of a child with CM
B) physical developmental delay or mental retardation in the child
C) the mother's age is under 30 years
D) repeated medical abortions
E) stillbirths
18. Prospective counseling:
A) takes place in a family that already has a sick child
B) takes place in a family at high risk of having a sick child
C) is carried out to determine the repeat risk of having a sick child
D) is carried out after the birth of a sick child
E) is the most effective way to prevent hereditary diseases
19. Medical and genetic counseling consists of stages:
A) diagnosis
B) calculation of genetic risk (prognosis)
C) treatment
D) medical judgment
E) counseling
20. Prenatal diagnostic methods are divided into:
A) Invasive
B) invasive
C) non-invasive
D) non-invasive
E) molecular-genetic
21. A married couple with a sick child with autosomal dominant disease applied to the medical and genetic consultation. The parents are healthy, the wife's age is 25 years, and the husband's age is 45 years. Determine the possible causes of the child's disease:
A) action of teratogens during pregnancy
B) spontaneous mutation in the gametogenesis of a woman
C) spontaneous mutation in the X chromosome of a man
D) spontaneous mutation in the autosome of a man
E) age of the male
22. A young couple in a consanguineous marriage without children has applied to a medical and genetic counselling centre. What will be the nature of medical and genetic counselling and genetic risk assessment in the offspring:
A) retrospective
B) prospective
C) high risk of dominant pathology
D) high risk of recessive pathology
E) no risk of hereditary pathology
23. A woman who has a child with Down syndrome applied to the medical and genetic consultation. The karyotype of the child is 46, t 13/21. What investigations the counsellor recommends:
A) biochemical analysis of blood and urine of the child
B) karyotyping of the woman
C) microbiological tests during the second pregnancy
D) karyotyping of the child's father

E) prenatal diagnosis of Down syndrome

Topic №7. Diseases with non-Mendelian type of inheritance.

I. Answer the test questions:

1. X-linked hereditary diseases are ....
| Haemophilia, hypertrichosis, ichthyosis.
|Klinefelter's syndrome.
|Down syndrome.
|Shereshevsky-Turner syndrome.
|daltonism, haemophilia.
2. Hereditary diseases transmitted with a Y chromosome ....
|hypertrichosis, ichthyosis, tympanic membranes on fingers.
|hypertrichosis, Down syndrome and Patau syndrome.
|Klinefelter's syndrome, Down's syndrome, Patau's syndrome.
|Ichthyosis, Edwards syndrome, Down syndrome.
|Haemophilia, colour blindness.
3. Hereditary diseases associated with changes in the number of sex chromosomes are ....
| syndrome, Klinefelter, Schereshevsky-Turner, trisomy-X.
|Haemophilia, Patau syndrome, Down syndrome.
|Edwards Down syndrome, hypertrichosis.
|Patau Edwards Down syndrome.
|Ichthyosis, ichthyosis.
4. Hereditary diseases associated with altered autosome number ...
|Patau syndrome, haemophilia.
|Down syndrome, Patau--
|Klinefelter syndrome, phenylketonuria, albinism.
|Down syndrome, Patau, trisomy X, YYY.
|Tay-Sachs disease, Lejeune's disease, Edwards syndrome, colour blindness.
5. Hereditary diseases associated with disorders of protein metabolism ...
|Haemophilia, ichthyosis, Patau's syndrome.
|Hypertrichosis, alcanthonuria, Klinefelter's.
|Haemophilia, phenylketonuria, glycogenemia.
|Tyrosinosis, Down syndrome, albinism.
| albinism, phenylketonuria.
6. Deletion of the short arm in the 5th pair of chromosomes in humans results in the formation of syndrome ...
|Cat's Cry.
|Down's.
|Albinism.
|Alkaptonuria.
|Haemophilia.
7. The term "sibs" in pedigrees refers to ....
|siblings.
|The person from whom the pedigree begins.

|A proband.
|The grandparents of the proband.
|The proband's uncles and aunts.
8. A patient with Down syndrome has...chromosomes.
|2п + 2
|2п - 1
|3р + 1р
|1п
|2п + 1
9. The occurrence of Patau syndrome is associated with ... chromosome.
|15
|21
|13
|8
|18
10. Inbreeding is ...
|hybridisation.
| cross-pollination or fertilisation.
|random crossbreeding, panmixia.
| free crossing, panmixia.
|Close inbreeding that increases homozygosity.
11. Thalassaemia (a form of anaemia) is inherited in an autosomal dominant pattern and has two forms, mild (Aa) and severe (AA). The occurrence of the mild form of thalassaemia is due to ... gene.
| non-complete dominance+
| over dominance
|complete dominance
| codominance
|pleiotropy.
12. The development of Edwards syndrome is associated with trisomy of the ... chromosome.
|17
|3
|21
|18
|14
13. The karyotype of a person with Klinefelter syndrome is ... chromosomes.
|45
|46
|47
|23
|44
14. Monosomy is characteristic of the genotypes of ....
|ХО, УО.
|ХХХ, ХХУ.
|ХХ, ХУ.
|ХХО, ХУО.

|ХХУУ, ХУУУ.
15. The karyotype of Shereshevsky-Turner syndrome has ... chromosomes.
|47
|44
|46
|27
|45
16. An inherited disease of humans resulting from changes in chromosome structure is ....
|Cat's cry syndrome.
|Schereshevsky-Turner syndrome.
|Klinefelter's syndrome.
|Haemophilia.
|X trisomy.
17. Phenylketonuria is a disorder of ....
|amino acid metabolism.
|carbohydrate metabolism.
|Lipid metabolism.
| mineral metabolism.
|Nucleic acid metabolism.
18. Down syndrome is caused by trisomy ... pair of chromosomes.
|21+
|15
|20
|13
|18.
19. The syndrome arising from deletion of the short arm in the 5th pair of chromosomes in humans is ....
|Cat's cry syndrome.
|Down syndrome.
|Albinism.
|Alkaptonuria.
|Haemophilia.
20. Trisomy on chromosome 13 leads to the development of syndrome ...
|Patau.
|Down's.
|Schereshevsky-Turner.
|Edwards.
|Klinefelter's.

II. Situational tasks

Question.1. In large families of myopic parents 12 children were born to myopic parents. Three of them were of normal vision and 6 were myopic. How many myopic children will be homozygous for the myopia gene?

Question.2. Three girls were born from the marriage of a woman suffering from hypertension (autosomal dominant trait) with a healthy man. У

The first daughter had 4 children with hypertension, the second daughter had 3 children with the same disease, and the third daughter had two healthy children. Determine the highest probability that the third daughter will have children with hypertension.

Question.3. In Transcarpathia among people in a series of generations there are observed long-term modifications. This form of variability is: a) similar to genocopy; b) reversible change of genotype and phenotype; c) irreversible change of phenotype; d) reversible change of genotype; e) inherited by the type of cytoplasmic heredity.

Question.4. Determine the probability of occurrence of a disease in a family where it is known that all relatives on the mother's side who lived to the age of 70 had the disease, and on the father's side all relatives are healthy.

III. Answer the questions?

1.Definition of chromosomal diseases, mechanisms of their occurrence.
2.Definition of diseases with hereditary predisposition, mechanisms of their occurrence.
3.Types of diseases with hereditary predisposition.

Topic №8. Principles of prevention of human hereditary pathology. Medical and genetic counseling
Topic №9. Monogenic diseases arising due to changes in protein structure.

I. Characterise the autosomal dominant type of monogenic inheritance.
1.
2
3.
4
5.
Characterise the autosomal recessive type of monogenic inheritance.
II.Characterise the X-linked dominant type of monogenic inheritance.
IV.Characterise X-chain recessive type of monogenic inheritance.
V.Characterise the U-chain type of monogenic inheritance.
Vl.Characterise polygenic diseases.
VII.Characterise mitochondrial type of monogenic inheritance.
VIII.Characterise diseases with non-traditional type of inheritance:
- mitochondrial diseases resulting from mitochondrial gene mutations;
- genomic imprinting diseases,
- diseases of expansion of trinucleotide repeats in regulatory or transcribed parts of genes.
- diseases caused by disruption of epigenetic regulation of gene expression.

Select one correct answer.

1. theoretical risk is calculated for:
A. mendelating monogenic diseases
B. polygenic diseases and multifactorial diseases
C. monogenic mitochondrial diseases
D. chromosomal diseases
D. chromonemal diseases
2 Empirical risk is calculated for:
A. infectious diseases

B. polygenic diseases
C. monogenic diseases
D. mendelating diseases
D. autosomal dominant diseases
3. Important features of hereditary diseases are:
A. high fertility of patients
B. delay in mental and physical development
C. predisposition to parasitic diseases
D. acute onset and course of the disease
D. accelerated mental and physical development
4. Autosomal syndromes are characterized by:
A. multiple organ inflammation, accelerated mental development, high risk of disease inheritance
B. multiple pigmentation of the skin, hair, accelerated physical development, no risk of inheriting the disease
C. multiple malformations, mental retardation, low risk of inheriting the disease
D. multiple metabolic abnormalities, mental and physical completeness, inheritance of the disease through a generation
E. no malformations, mental and physical retardation, medium genetic risk
5. Identify the correct list of causes of inherited diseases:
A. mutations, migration, gene drift
B. mutations, replication errors, nucleotide pair substitutions
C. mutations, respiration errors, lipid substitutions
D. mutations, mediations, gene relaxations
E. mutations, gene colour changes, phosphoric acid pair substitutions
6. Identify the correct list of occurrences of monogenic diseases:
A. deficiencies, deletions, translocations of chromosomes
B. duplications, relaxations, trisomies
C. transitions, transversions, reading frame shifts.
D. translocation, monosomy, polysomy
D. chromosome translocation, transposition, transversion, chromosome translocation
7. Mendelinating monogenic diseases include:
A. diseases caused by mutations of mycoplasmas, Golgi apparatus, ribosomes
B. diseases caused by mutations of proteins, lipids, nuclear membranes
C. diseases caused by mutations of the genome, chromosomes, chromonemes
D. diseases caused by mutations of nuclear genes, with autosomal dominant and autosomal recessive inheritance
E. diseases caused by mutations of mitochondrial genes with X- and U-linked inheritance
8. Hereditary diseases can be recognized by the following combinations of clinical features:
A. cough, runny nose, high body temperature
B. obesity, weight loss, sleep disturbance
C. diarrhea, constipation, jaundice
D. congenital malformations, mental and physical retardation
E. congenital toxoplasmosis, accelerated physical and mental development
9. Human hereditary diseases are characterized by the following features:
A. most of them begin in old age, accompanied by high body temperature, end with recovery

B. most of them begin in old age, accompanied by headaches, fatigue
C. most of them begin in adulthood, accompanied by increased tone, mental activity
D. most of them begin in childhood, accompanied by accelerated mental and physical development
E. most of them begin in childhood, accompanied by delayed mental and physical development
10. Monogenic diseases are characterized by:
A. low frequency, early onset, end in death or disability in children
B. low frequency, late onset, effective treatment
C. high incidence, acute onset, recovery.
D. moderate frequency, runny nose, cough.
E. low frequency, acute onset, recovery
11. Hereditary diseases are divided into:
A. infectious, protozoal, prostaglandin-related
B. chromonemal, chromaffin, polyphenic
C. polygenic, polyphenic, multiplex.
D. chromosomal, gene, polygenic
E. gene pool, genetic, genesis, chromoplast.
12. human hereditary diseases are diagnosed by methods of:
A. genealogical, biochemical, cytogenetic
B. descriptive, biological, cytostatic
C. biophysical, laser, anatomical
D. echological, cardiographic, cartographic
D. physical, chemical, biological
13. Identify the correct criteria for recognising monogenic diseases:
A. changes in the number and structure of chromosomes, late onset, acute course of the disease
B. changes in the structure and function of single genes, early onset, chronic course of the disease
C. changes in the structure and function of many genes, early onset, acute course of the disease
D. changes in the number of genomes, individual chromosomes, no impairment of mental and physical development
E. disturbances in the structure and function of all genes, early onset, acute course of the disease
14. Identify the correct combination of inheritance types for monogenic diseases:
A. autogenously, autosomal, genome-linked
B. autosomal, recessive, linked to the U chromosome
C. autosomal, genomic, chromosomal.
D. gonosomal, homologous, autosome-linked
D. genotype-linked, autosomal, homologous, homologous
15. Types of inheritance of monogenic diseases:
A.autosomal recessive, autosomal dominant, recessive, X-linked
B. autosomal regressive, autosomal polypotent, genome-linked.
C. autosomal reciprocal, recessive, regressive.
D. autosomal-chromosomal, genotype-linked, gonosomal-autosomal
E. autosomal-polygenic, autosomal-codominant, genotype-linked.
16. Mendelating diseases (traits) are traits (diseases):
A. caused by mutations of individual chromosomes, extranuclear genes, sex-linked
B. caused by mutations of several genes, separate genomes, cytoplasmic genes
C. caused by polygenes, mutations of genomes, linked to the X-chromosome

D. caused by chromosome divergence errors, loss of chromosomes, linked to sex chromosomes
E. caused by mutations of single, nuclear genes, with autosomal dominant type of inheritance
17. Diseases with a non-traditional type of inheritance include:
A. autosomal dominant, autosomal recessive, polygenic diseases
B. autosomal-honosomal, autosomal-recessive, multifactorial diseases
C. mitochondrial, genomic imprinting, prion diseases
D. sex chromosome-linked, dominant and recessive, hereditary predisposition diseases
E. autosome-linked, polygenic, multifactorial diseases

Topic №10. Polygenic diseases
I. Criteria for describing diseases:
1.Name of the disease
2.General characteristic
3. Symptom of the disease
4. Causes of the disease (genetic mechanisms)
5. Conditions necessary for the development of the disease (action of special environmental factors on the organism)
II. Polygenic diseases
1. Ischemic heart disease
2. Hypertensive disease
3. Gastric and duodenal ulcer.
4. Type I diabetes
5. Bronchial asthma
6. Schizophrenia
7. Epilepsy
8. Manic-depressive psychosis
9. Alcoholism
10. Oligophrenia (undifferentiated
11. Neural tube defects (most common pathology):
* anencephaly (absence of part of the skull brain);
* encephalocele (herniation of the brain);
* cleft spine (herniation of the spine);
* hydrocephalus (brain drops);
* porencephaly (absence of brain tissue anywhere).
12.Cleft lip (cleft lip and cleft palate).
13.Congenital malformations of the heart, gastrointestinal tract and kidneys.
14.Congenital dislocation of the hip and leg.
16. Atherosclerosis
17. Alzheimer's disease
18. Autoimmune diseases of the thyroid gland.
19. Rheumatoid arthritis,
20. Cirrhosis of the liver
21. Psoriasis

Topic №12: Fundamentals of population genetics

I. Questions

1. What does population genetics study?
2. What is a population and gene pool?
3. Give an ecological and genetic characterisation of a population?
4. Give definition of terms like panmixia, Mendelian populations, demes and isolates?
5. Hardy-Weinberg's Law. For which populations is Hardy-Weinberg's law applicable?
6. Name the three main statements (principles) of Hardy-Weinberg's law?
7. What forms of natural selection exist?

II. Test questions may contain - 1, - 2, - 3 or 4 correct answers.

1. Demographic indicators characterising a population:
A) abundance
B) polymorphism
C) sex composition
D) age composition
E) gene pool
2. evolutionary factors that maintain polymorphism (heterogeneity) in a population:
A) isolation
B) migration
C) gene drift
D) inbreeding
E) natural selection
3. Genetic criteria that characterise a human population:
A) abundance
B) polymorphism
C) sex composition
D) age composition
E) gene pool
4. Evolutionary factors that reduce the polymorphism (heterogeneity) of a population:
A) isolation
B) migration
C) gene drift
D) inbreeding
E) natural selection
5. Conditions for maintaining the constancy of gene frequencies in a population:
A) absence of crossing over
B) absence of mating
C) absence of migration
D) absence of natural selection
E) absence of light
6. Selective marriage in which individuals with certain traits form pairs more often than usual is called:
A) incestuous marriage
B) assortative marriage
C) outbreeding

D) inbreeding
E) panmixia
7. Evolutionary factors are the source of formation of the genetic load of a population:
A) panmixia
B) mutation
C) migration
D) natural selection
E) isolation
8. The free, indiscriminate interbreeding of individuals in a population is called:
A) incest
B) positive assortative marriage
C) negative assortative marriage
D) inbreeding
E) panmixia
9. The conditions for fulfilment of Hardy-Weinberg equilibrium are:
A) absence of panmixia
B) presence of panmixia
C) absence of mutations, migration, natural selection
D) large population size
E) small population size
10. Population genetics studies:
A) populations, their species, genetic and demographic structure
B) genetic processes at the organismal level
C) conditions of origin and existence of human population
D) genetic processes at the population level
E) inheritance of traits in individuals in a population
11. Factors that limit panmixia in human populations:
A) gene drift
B) migration
C) isolation
D) national differences
E) religious differences
12. inbreeding is:
A) unrelated marriage
B) closely related marriage
C) marriage between blood relatives
D) positive assortative marriage
E) negative assortative marriage
13. Populations that have a population size of 1500 to 4000 individuals, a kinship rate of 80% to 90%, gene flow from other populations of 1- 2%, and a population growth rate of 20% per generation are:
A) isolates
B) demes
C) Mendelian populations
D) ideal populations
E) large populations

14. Causes of isolation in human populations:
A) growth barriers
B) religious barriers
C) racial barriers
D) social barriers
E) genetic barriers
15. Populations that have a population size of up to 1500 individuals, an intra-group mating rate of over 90%, gene flow from other populations of less than 1%, and a natural population growth rate of 25% per generation are:
A) isolates
B) demes
C) Mendelian populations
D) ideal populations
E) large populations
16. Outbred marriages include:
A) unrelated marriages
B) closely related marriages
C) marriages between blood relatives
D) accidental marriages
E) non-random marriages
17. Assortative crosses are crosses characterised by the mating of:
A) unrelated individuals
B) related individuals
C) individuals in isolated populations
D) individuals with similar genotype
E) individuals from different populations
18. The components of adaptability of individuals are:
A) growth
B) mass
C) viability
D) abundance
E) fecundity
19. The action of natural selection in recessive diseases results in:
A) elimination of the dominant
19. The action of natural selection in recessive diseases results in:
A) elimination of the dominant allele
B) elimination of heterozygotes
C) elimination of recessive allele
D) an increase in the frequency of the recessive gene
E) decrease in the frequency of the recessive gene
20. The action of natural selection in dominant diseases results in:
A) elimination of the dominant allele
B) elimination of recessive allele
C) increase in the frequency of the dominant gene
D) a decrease in the frequency of the dominant gene

E) a decrease in the frequency of the recessive gene
21. The action of natural selection in haemolytic disease of the newborn is an example of an action:
A) in favour of homozygotes
B) against homozygotes
C) against heterozygotes
D) in favour of a recessive allele
E) in favour of the dominant allele
22. Types of populations:
A) Darwinian
B) Mendelian
C) panmix
D) simple
E) isolated
23. Populations are divided into:
A) simple
B) large
C) complex
D) isolated
E) ideal
24. Populations are subdivided by size and mating type into:
A) morganovian
B) Mendelian
C) large
D) simple
E) small
25. Depending on the nature of crossbreeding, populations are divided into:
A) asymmetrical
B) outbred
C) allosteric
D) inbred
E) insertional
26. Classification of populations:
A) simple
B) isolated
C) constructive
D) Mendelian
E) outbred
27. The formation of the gene pool of a population is influenced by the processes of:
A) gene drift
B) protein drift
C) mutations
D) gene colour
E) natural selection
28. Factors that increase genetic polymorphism in populations:
A) gene drift

B) outbreeding
C) mutations
D) isolation
E) inbreeding
29. The fitness of a population is determined by:
A) age of individuals
B) survival rate of individuals
C) fecundity of individuals
D) growth of individuals
E) the mating success of individuals
30. The genetic load of populations results from the effects of:
A) climate
B) mutations
C) temperature
D) natural selection
E) atmospheric pressure
31. The accumulation of genetic pathology in populations is the result of:
A) increased adaptation of genotypes
B) increased fertility
C) a decrease in the adaptability of genotypes
D) increased survival of genotypes
E) elimination of unadapted genotypes
32. An increase in the frequency of genetic pathology in populations occurs under conditions of:
A) panmixia
B) inbreeding
C) outbreeding
D) isolation
D) gene drift
33. The genetic load of populations decreases under conditions of:
A) isolation
B) outbreeding
C) inbreeding
D) panmixia
E) gene drift
34. Population genetic cargo includes:
A) infectious diseases
B) nutritional diseases
C) hereditary diseases
D) spontaneous abortions
E) medical abortions
35. The genetic load of a population forms diseases:
A) influenza
B) haemophilia
C) pneumonia
D) phenylketonuria

D) ichthyosis

36. The action of natural selection in populations leads to:
A) retention of unfit genotypes in a population
B) preservation of genotypes with high growth in the population
C) preservation of the most adapted genotypes in the population
D) increase in viability of genotypes
E) retention of genotypes with high body weight in the population
37. Migration (exchange of genes between populations) contributes to:
A) decreasing the genetic diversity of populations
B) increase the genetic diversity of populations
C) an increase in the intensity of the mutation process
D) an increase in the frequency of homozygotes
E) an increase in the frequency of heterozygotes
38. Gene drift (random changes in gene frequency) in populations leads to:
A) an increase in the genetic polymorphism of populations
B) a decrease in the genetic polymorphism of populations
C) an increase in the genetic load of populations
D) a decrease in the genetic load of populations
E) a decrease in the adaptability of genotypes in populations
39. Population isolation (geographic, religious) contributes to:
A) an increase in the diversity of genes and genotypes
B) an increase in the adaptability of genotypes
C) decrease in the diversity of genes and genotypes
D) decrease in the genetic load of populations
E) increase the genetic load of populations
40. An increase in the frequency of mutations in a population results in:
A) an increase in the number of individuals in a population
B) an increase in the number of genetically different individuals in a population
C) an increase in the number of adapted genotypes
D) an increase in the number of unadapted genotypes
E) an increase in the pressure of natural selection
41. The action of elementary evolutionary processes in a population leads to:
A) maintenance of constancy of gene and genotype frequencies
B) changes in the frequencies of genes and genotypes
C) increase in the frequencies of homozygotes
D) formation of an optimal ratio of homo- and heterozygotes in a population
E) increase in the adaptability of genotypes

Solve the problem using knowledge of the Hardy-Weinberg law. When solving the problem it is necessary to write down the condition and the course of solving the problem.

Task 1: In a human population, the number of individuals with blue eye color is 49%. Determine the genetic structure of the population and the number of people with brown eyes.

Task 2: In Europe there is 1 albino per 10 000 people with normal melanin content. The albinism gene is inherited by autosomal recessive type. Calculate the frequency of carriers of the albinism gene. A carrier is an organism that is heterozygous for a gene that can cause a metabolic disorder in the homozygous state.

Task3. Predisposition to diabetes mellitus is inherited in an autosomal recessive pattern. The frequency of occurrence of the recessive predisposition gene in the USA is approximately 22, 5 %. What is the frequency of heterozygous carriers of the predisposition to diabetes mellitus gene in the USA.

Task4. In humans, the gene "rhesus positive" is dominant in relation to the gene "rhesus negative". In the population examined for this indicator, 1982 people were "Rh positive" and 368 were "Rh negative". What is the genetic structure of this population?

Task 5: One of the forms of glycosuria is inherited as an autosomal recessive trait and occurs with a frequency of 7:1000000. Determine the genetic structure of the population.

Task 6: Brachydactylic in humans is inherited as an autosomal dominant trait with a penetrance of 25%. The disease occurs with a frequency of 6:10 000. Determine the number of heterozygous carriers of the brachydactylic gene

Task7. Alkaptonuria occurs in 2% of people and is caused by autosomal dominant gene. In women, the alkaptonuria gene is not manifested; in men its penetrance is equal to 20% (V.P. Efroimson, 1968). Determine the genetic structure of the population for the analysed trait, based on these data.

Task 8: One of the forms of phenylketonuria is inherited as an autosomal recessive trait and occurs with a frequency of 7:1000000. Determine the frequency of heterozygotes in the population.

Task9. The population of Europeans on the system of blood groups Rhesus contains 85% of Rhesus positive individuals. Determine the saturation of the population with recessive allele.