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Modern problems of heredity

Educational materials for practical lessons

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Educational materials are developed according to the educational work program of special course "Modern problems of heredity". The publication contains methodological materials for practical works of this special course. Designed for full-time and part-time masters of specialty "Biology".

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THE CONTENT

EXPLANATORY NOTE

Human is a part of the biosphere and the product of its evolution, that’s why the patterns of biological processes, which have universal meaning, fully applies to humans. Thus, the laws of heredity and variation, which are explained by general genetics on more simple objects, can be transferred to humans and its population.

However, consider some features of the human as an object of study that makes us look for unconventional ways and methods of research. These features, which greatly complicate genetic analysis, are related not only to the sphere of biology, but also to the biosocial nature of human.

The research and teaching materials "Modern problems of heredity" include presentation of the material, which will help master students to learn more deeply and consolidate theoretical material on this special course and give an opportunity to show the value of the acquired knowledge for practical work.

Each topic of practical work contains what the student should know and do, tasks and brief theoretical knowledge on the topic, and test questions for the interview. At the end of the methodological publication is a list of recommended references.

Practical work №1

Theme: Dermatoglyphics. Twin method.

Dermatoglyphics is a study of fingers, palms and footsteps skin relief. It was be founded by F. Galton. Dermatoglyphics method can be used in the complex diagnosis of hereditary diseases for the determination of the twin zygosity and identification of person.

Twin method helps to study the influence of heredity in the development of trait. It is based on comparing of the similarity (concordance) and differences (discordance) on traits between monozygotic and dizygotic twins. Twins method helps to determine if the trait is hereditary, and to evaluate role of environmental factors in the manifestation of this trait.

To know:

1. Dermatoglyphics method, its value for medicine.
2. Peculiarities of skin pattern on palms, fingers and footstep that have diagnostic value.
3. Twins method, its value for medicine.
4. Formula of K. Holtsynher for calculating the heredity coefficient.

Be able to:

1. Explore of the fingers' papillary lines prints:

a) determine the types of fingers patterns;

b) write formula of fingers patterns;

c) count the deltoid index;

d) calculate a crests number of each finger and calculate their quantification of the total average for 10 fingers.

2. Explore prints of palm relief:

a) find finger threeradius, palmar's main lines, flexor furrows;

b) measure the *atd* angle of palm.

3. Determine the coefficient of heredity with the K.Holtzinger's formula.

The audience work.

Task 1. Consider the different variants of fingers skin patterns on the Figure 1. Draw in the protocol three basic types of patterns: arch, loop, whorl, and mark them accordingly letters as A, L, W.

A ***B*** ***C***

Fig. 1. Variants of fingers skin patterns: A – arch (A); B – loop (L); C – whorl (W).

Task 2. Get papillary lines prints of your own fingers. For this wash your hands with soap careful and wipe them dry. Dilute the ink with glycerine to consistency of thick cream. Put on a glass plate with size of 15x25 mm by glass rod paint in 2-3 places and allocate it evenly by a rubber roller. Swipe each finger by roller three times, applying it consistently to radial, medial and ulnarnal surfaces of last phalanx. Then make on a paper the each finger print on radial side, gently turning it to ulnarnal margin. Make fingerprints in a certain order - from left to right, at first for fingers of the left hand and then for fingers of the right hand.

Task 3. Consider Figure 2, which shows prints of fingers papillary lines of the right and left hand. Finger patterns of the right and left hand in the picture, write as in following example:

	I	**II**	**III**	**IV**	**V**
Right hand	W	L^u	L^r	L^r	L^r
Left hand	L^r	L^r	L^r	L^r	L^r

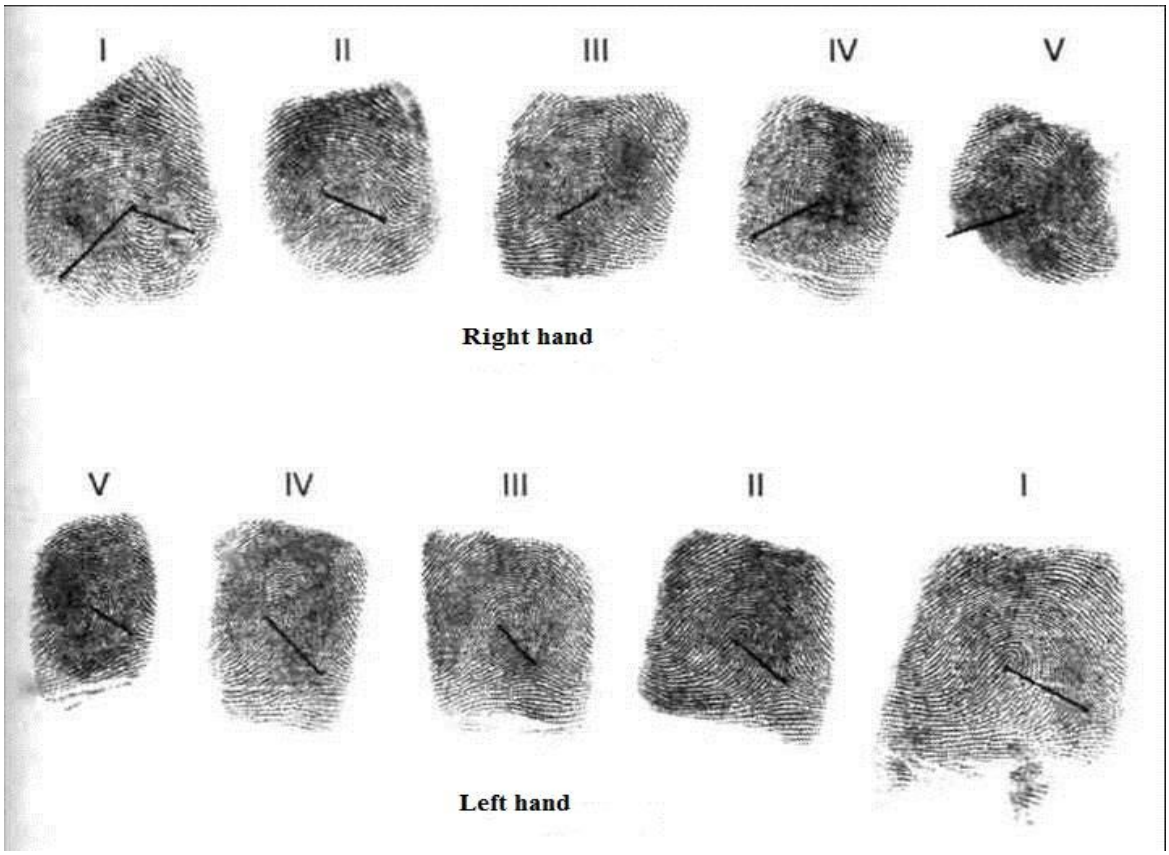

Fig 3. Papillary lines prints on the fingers of the left and right hands.

The first (I), the second (II), the third (III), the fourth (IV), the fifth (V) fingers of left and right hands are marked with Roman numerals. Fingers patterns are marked with letters: W (*eng*. whorl), A (*eng*. arch); L (*eng*. loop). If loop is deflected in direction of radius, it is called radial (L^r), if in the direction of the ulna – ulnarnal (L^u).

Delta index, which indicates the number of threeradiuses or deltas (their so called by the similarity of shape to the Greek letter ω), for some person is determined by the formula:

$Dl = L + 2W/A + L + W \times 10$,

where L – the total number of all loops on the right and left hand; W – the total number of all whorls on the right and left hand; A – the total number of all arches on the right and left hands. In our case deltoid index is:

$Dl = (9 + 2\ W + 1) / (0 + 9 + 1) \times 10$.

Remember that papillary lines of different directions never intersect, but can converge in certain areas and form threeradiuses (delta). Each whorl pattern has two

deltas, loop has one delta, and arch does not form any one.

Quantity index of dermatoglyphics is a counting of crests (a number of papillary lines between delta and center of pattern). Count a crests number between delta and center of each picture finger pattern and determine their summary average for ten fingers. Crest count do on the following way. Swipe a straight line from delta to center of pattern and count a crests number that cross this line. Crests number are written under the each finger print. It does count neither threeradius nor last crest that forms center of the pattern. The last crest is considered for loops. In our case, a crests number is the following:

(10 + 16 + 7 + 15 + 24) + (18 + 14 + 9 + 18 + 14) = 145.

After exploring of prints papillary lines, calculate your own deltoid index and count a crests number.

Task 4. Get relief imprint of your palm. Swipe by rubber roller several times on a glass plate with paint and then apply the paint evenly on palm and fingers for that. Place palm with paint by ulnarnogo edge over the paper sheet and slowly lower it to him. Press the other hand on a middle of the hand back to deep part of palm huddled tightly to paper. Push thumb at paper by nail phalanx, while turning it in direction to index. Then abruptly lift the brush from paper straight up. Fingerprint of the right hand do in the same way. Remove paint from palm with cotton wool soaked with turpentine. Wash hands with warm water and soap.

Task 5. Find angle *atd* on your palm print. At base of II, III, IV and V fingers are localized finger threeradiuses –places where three of different direction papillary lines are converged. They are marked with Latin letters a, b, c, d (Fig. 3). The main axial palm threeradius is situated around bracelet fold; it is marked with Latin letter t. Find and connect with lines the points a, t, d on your palm print. Use a protractor to measure angle *atd* (normally it should not exceed 57°).

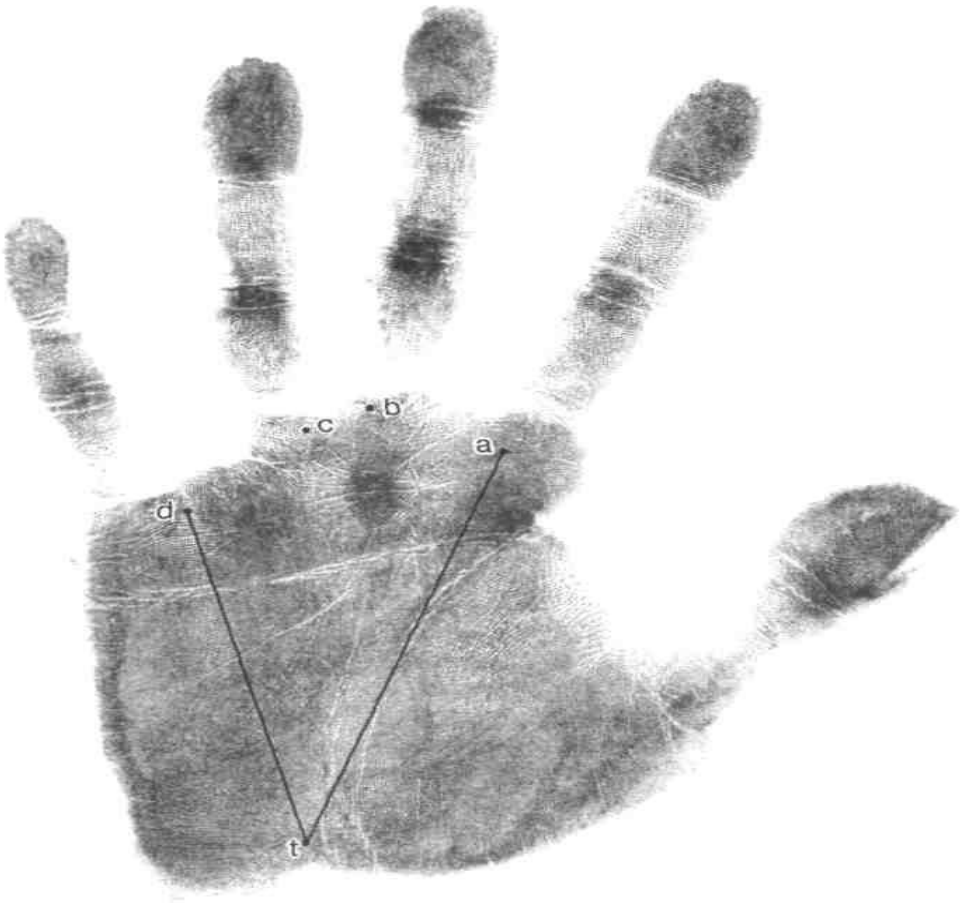

Fig. 3. Print of the left palm.

Task 6. Write to protocol the K. Holtsynher's formula for determining the coefficient of heredity H:

$H = (C_{MT} - CDT) / (100\ \% - CDT)$,

where H – a coefficient of heredity;

C_{MT} – a concordance of monozygotic twins, %;

CDT – a concordance of dizygotic twins, %.

Pay attention that when H is equal to 1, then the trait is due entirely by hereditary component, and if H is equal to 0, then decisive role in the formation of trait plays influence of environment. The coefficient closed to 0.5 indicates approximately the same effect of heredity and environment on the trait's formation. Determine from table 1 the heredity factor H for each of traits, which are shown in it; define the role of heredity and environment influence on the expression of these traits.

The degree of environment influence on traits' formation is determined by the formula:

$$C = 100\ \% - H.$$

Table 1.

Concordance of certain traits in monozygotic and dizygotic twins

Trait	Concordance, %	
	Monozygotic twins	Dizygotic twins
Epilepsy	**67**	**3**
Measles	**98**	**94**
Parotitis	**82**	**74**
Hypertension	**26,2**	**10**
Rheumatics	**20,3**	**6,1**
Splayfoot	**32**	**3**
Crack of lips	**33**	**5**
Congenital dislocation of the hip	**41**	**3**
Tuberculosis	**37**	**15**
Schizophrenia	**70**	**13**

Questions for the interview:

1. What is a dermatoglyphics?
2. Why a dermatoglyphics method is used?
3. Who and when proposed the classification of papillary patterns at first?
4. What are the papillary lines?
5. Are there people on the Earth with the same fingerprint patterns?
6. Over what period of fetal development do patterns on skin of fingers, palms and feet appear?
7. What patterns on the fingers are the most widespread?
8. What are threeradiuses, or delta?
9. By what letters are fingers threeradiuses marked?

10. Where is the main (axial) palmar threeradius?

11. How many deltas have whorls and loops?

12. What is a deltoid index and why is it determined?

13. What is the angle of palm atd? What is its value in normal?

14. What is dactyloscopy, palmeroscopy, plantaroscopy?

15. How much crests are on one finger in average?

16. What are caused the individual characteristics of patterns on skin fingers, palms and footsteps?

17. Is it appropriate to apply the method of dermatoglyphics for further diagnosis in people with changes in karyotype?

18. Is it appropriate to apply dermatoglyphics method for diagnosis of hereditary genetic diseases?

19. What typical dermatoglyphic indexes in patients with Down syndrome do you know?

20. What angle atd is in a patient with Klinefelter's syndrome?

21. Who did propose the twins research method at first?

22. What is the practical application of twin method?

23. What are monozygotic and dizygotic twins?

24. What is the concordance and discordance?

25. What formula is used for determining the coefficient of heredity?

26. Are the same genotypes in dizygotic twins?

27. How can be determined the influence of the environment on the traits’s formation?

28. Are monozygotic twins genetically identical?

Practical work № 2

Theme: Individual characteristics of human chromosomes. Determination of X-chromatin.

Karyotype is a chromosome complement with consider of their number, size, shape, and s characterized by a certain species. Cytogenetic method is used for the karyotype determination; it helps to learn the structure of chromosomes and number of chromosomes in the studied cells of organism. In medical practice to confirm the diagnosis of chromosomal diseases are needed to establish the patient's karyotype.

Sex chromatin is studied to determine the number of X-chromosomes in somatic cells very quickly. X-chromatin is a discoid corpuscles (Barr corpuscle), that is detected in interphase nuclei of human somatic cells: is situated directly on the inner nuclear membrane. X-chromatin is condensed X-chromosome, which condenses in women in early embryogenesis.

The determination of X-chromatin in somatic cells matters as rapid method for diagnosis of chromosomal disorders, associated with changes in the number of X chromosomes.

To know:

1. The essence and importance of cytogenetic method.
2. Karyotypes's peculiarities of people with chromosomal diseases (Klinefelter syndrome, Down syndrome, Turner, Trisomy-X).
3. Structural and functional organization of interphase nucleus chromatin.
4. The nature of X-chromatin and methods of its determination in somatic cells.

Be able to:

1. Determine the number of chromosomes in the karyotype.
2. Identify chromosomes in groups A, B, C, D, E, F, G.
3. Determine the chromosomal sex of the individual, exploring karyotype.
4. Identify X-chromatin in cells of buccal epithelial cells in the form of Barr corpuscles.

The audience work.

Task 1. Consider micrograph of cells metaphase plate of male and female, who have normal karyotype. It shows the chromosomes at metaphase. To study the characteristics of the karyotype it is necessary to place (classified) chromosomes according to a specific scheme (fig. 1-3). Chromosomes are divided into groups with decreasing their size and centromeric index. According to the Denver International classification human chromosomes are divided into seven groups: A, B, C, D, E, F, G. All chromosomes have serial numbers. The group A contains the biggest chromosomes (metacentric or close to it), and the group C - the smallest (acrocentric).

Arrange the chromosomes in groups, using the proposed micrograph of metaphase plate. Carefully cut out images of chromosomes for this. Identify them according to the size and position of centromere, pick for each chromosome the homologous pair and then organize them in groups A, B, C, D, E, F, G. First determine the chromosome of the first and second pairs. They are the largest in size, but the chromosomes of the first pair are metacentric, and of the second are submetacentric; 3rd pair of chromosomes is metacentric. They are shorter than chromosome in a first and second pairs. Chromosomes of the 1st - 3rd pairs form a group A; 4th and 5th pairs of chromosomes are submetacentric. They are almost identical in size and shape, so they are difficult to differentiate. They belong to group B. Chromosomes of the 6th-12th pairs - mostly submetacentric, have medium size, differ with lengths of shoulders, these chromosomes form a group C. The X sex chromosome also belongs to the group C. Chromosomes of the 13th-15th pairs (group D) are typically acrocentric and have average size; chromosomes of the 16th-18th pairs (group E) are submetacentric, they are small; chromosomes of the 19th-20th pairs (group F) are closer to metacentric by the form, they are short. Chromosomes of the 21st-22nd pairs (group G) - the smallest acrocentric chromosomes. Y-chromosome by the size and shape is similar to the 21st-and 22-nd pairs of chromosomes. The difference between them is that the long shoulders in Y-chromosome are parallel, but it is difficult to differentiate this chromosome on all metaphase plates.

Tucking homologous pairs, sort them according to the serial number and stick to protocol. Combine with the brace the chromosomes, which belong to the same group, and set their group. Make a conclusion about karyotype, noting: a) the number of chromosomes in it, and b) sex chromosome.

Task 2. Prepare the temporary micropreparations scraping buccal of epithelium cells. Take a sterile metal spatula for this and easily hitting it on the mucosa of the cheek, make scraping. Transfer these cells to a clean and pre-fat slide. Apply to the received scraping 2-3 drops of 1% solution acetoorseinum. After 3-4 min cover the preparation with covered lenses. Through the surplus paint, that owerflow covered lenses, with filter paper.

Look upon the ready micropreparations first with the small (eyepiece x 10, lens x 8) and medium (eyepiece x 10, lens x 40) magnification. You will see a layer of epithelial cells, which are visible in the cytoplasm of interphase nuclei in the view. Note the shape of nuclei, size, peculiarities of the heterochromatin placement, shape and thickness of the nuclear membrane. Then continue to explore the micropreparations at high magnification (eyepiece x 10, lens x 90).

To determine the percentage of X chromatin should examine 100 cells. Remember that the X-chromatin is considered only in cells that have correct oval nucleus, they are optically thick, with a smooth, intact and pit (Fig. 4). Number of X chromosomes in the cells equals the number of X-chromatin lumps plus one. Note that the X-chromatin is localized perimembrane in the nucleus and may be spherical, oval, triangular or as a thickening of the nuclear membrane. The number of cells with X-chromatin in normal is 20-50%. Determine the percentage of cells containing the X-chromatin in prepared temporary micropreparations. Write the results in the protocol. Draw in the protocol 1-2 epithelial cells, which have perimembrane X-chromatin in the nuclei. Mark on the figure: a) nucleus; b) cytoplasm; c) nub of X-chromatin.

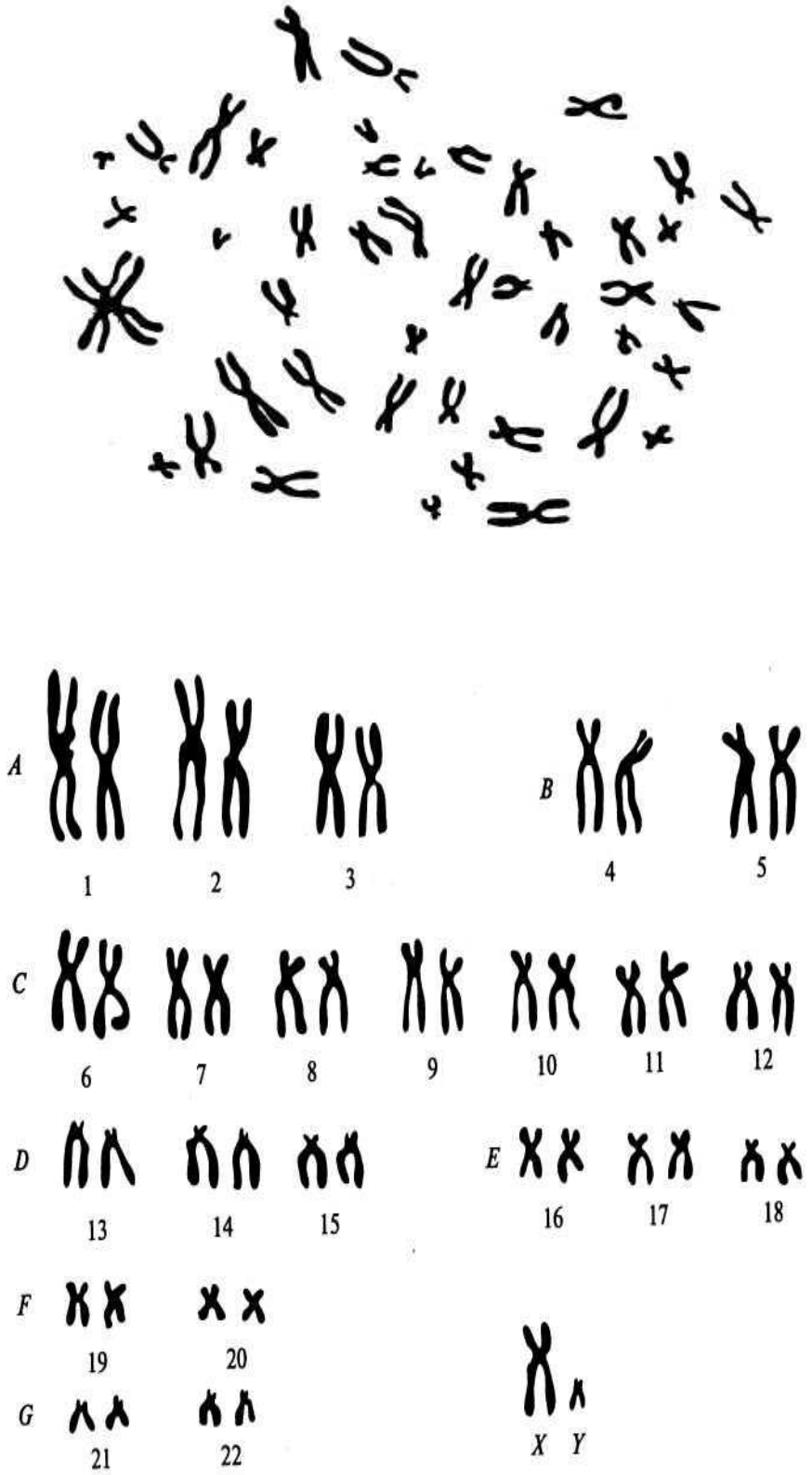

Fig.1. Man's normal karyotype (metaphase plate and karyogram).

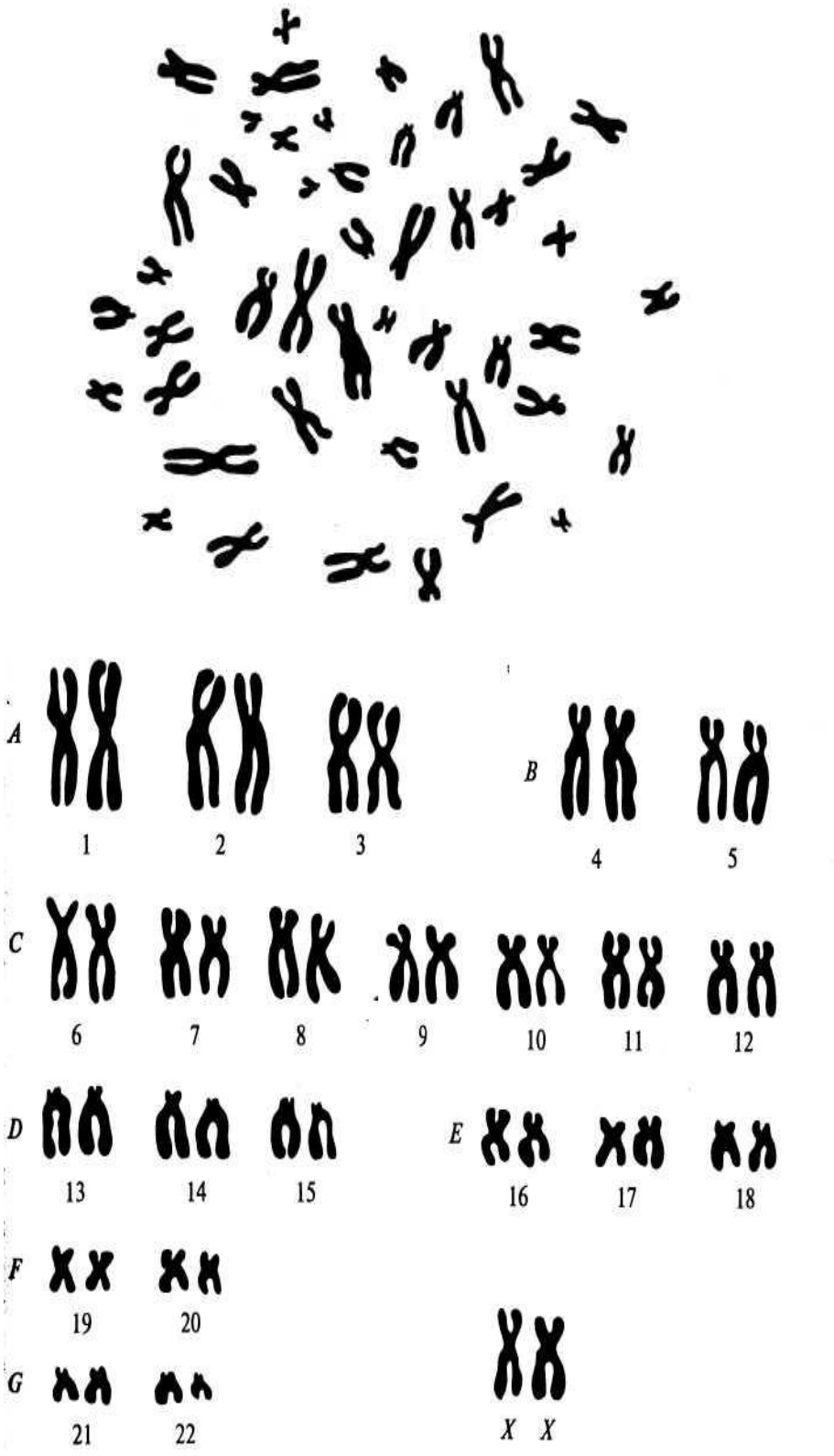

Fig. 2. Woman's normal karyotype (metaphase plate and karyogram).

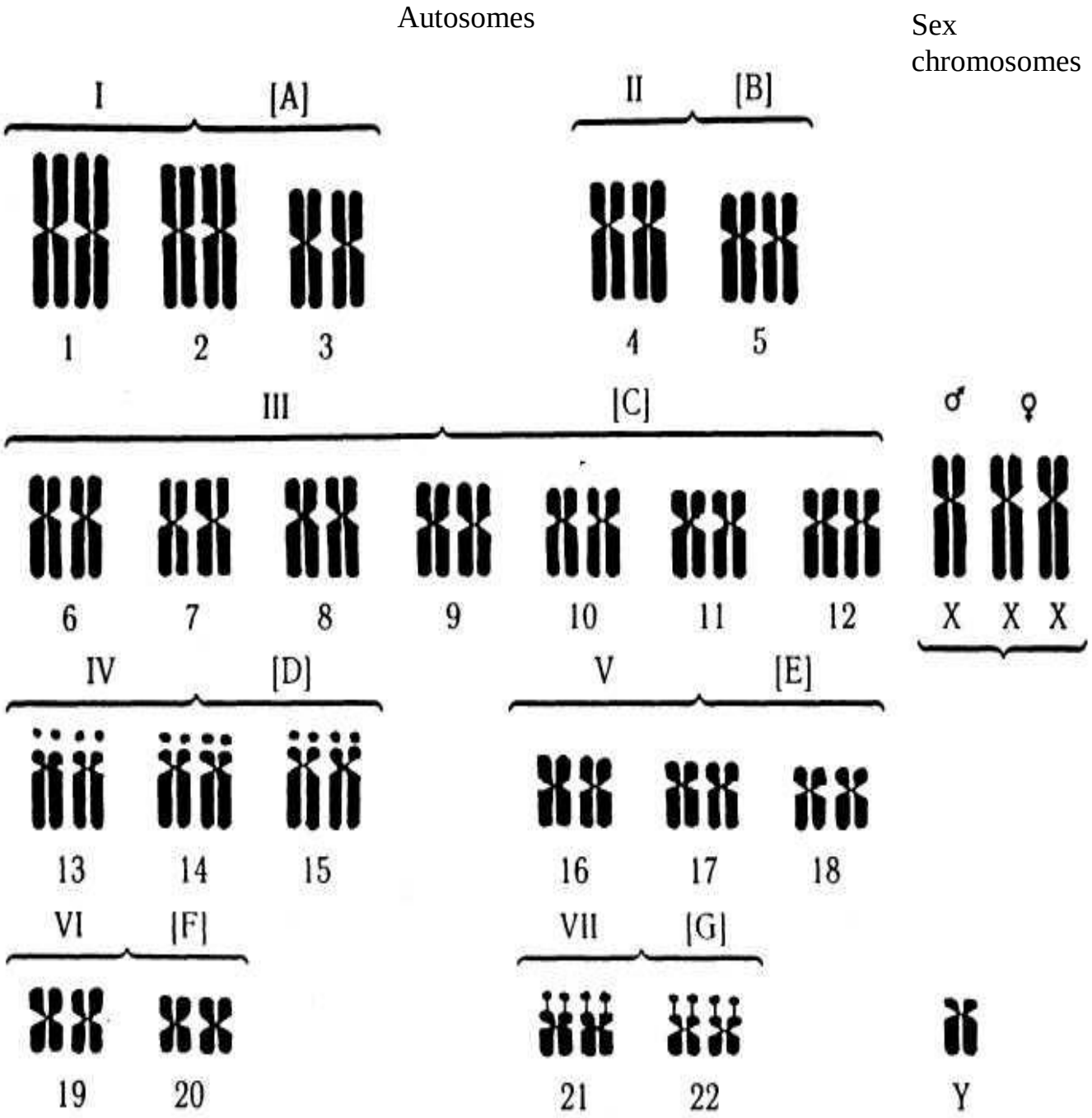

Fig.3. Idiogram of human's chromosomes according to the Denver nomenclature(1960).

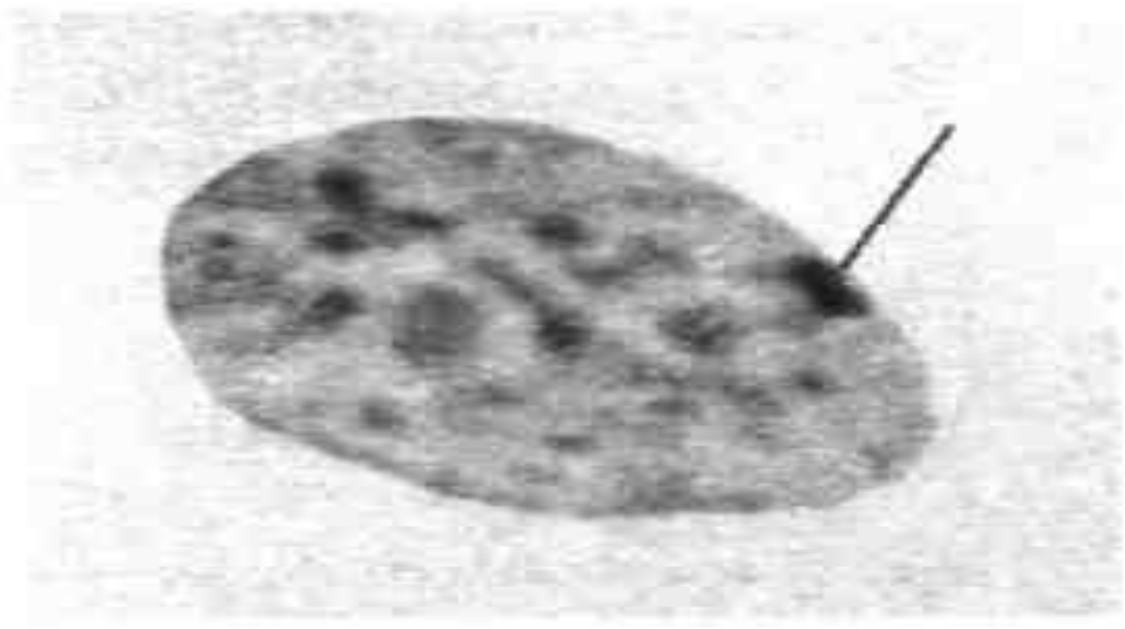

Fig. 4. X-chromatin (1) in cells of women's buccal epithelium (Barr corpuscle).

Questions for the interview:

1. What is the cytogenetic research method?
2. What is the significance of cytogenetic method in clinical practice?
3. What mutations can be detected by cytogenetic method?
4. Is it appropriate to use cytogenetic method for the diagnosis of Klinefelter's syndrome, Turner's, Down syndrome?
5. Is it enough for the diagnosis of hereditary diseases to study chromosomes by their visual observation in microscope at metaphase?
6. When was the first International Classification of human chromosomes designed?
7. How many chromosomes are in the karyotype: a) man b) Drosophila?
8. What karyotype's changes are observed in syndromes: a) Klinefelter b) Down c) Turner?
9. What karyotype does the male have, who is heterozygous for color blindness gene?
10. What karyotype does the girl have, who is hemizygous for color blindness gene?
11. What features show the chromosome's belonging to a certain pair during karyotyping?
12. What types of human chromosomes do you know, depending on the placement of

centromeres?

13. How are the sex chromosomes marked and to which group do they belong according to the Denver International Classification of human chromosomes?
14. What is the smallest metacentric chromosome numbers?
15. What is the metaphase plate?
16. Is it possible to apply the method of X-chromatin detection as a rapid method for diagnosis of hereditary diseases?
17. What indications for using the rapid method of determining the sex X-chromatin do you know?
18. Can you consider the X-chromatin as a facultative heterochromatin?
19. In what cells of tissue can the X-chromatin be determined?
20. What laboratory rapid method can be appropriately applied to diagnose Turner and Klinefelter syndrome?
21. Can you find the nubs of X-chromatin in a boy with Patau syndrome?
22. Is it possible to detect nubs of X-chromatin in somatic cells of a man who suffers from diabetes?
23. Is it possible to detect the nubs of Y-chromatin in somatic cells of healthy man?
24. What karyotype does a man have in the cells with X-chromatin nub?
25. Is it possible to detect the X-chromatin in man with karyotype 47, HYY?
26. How many nubs of X-chromatin are in the cells' nuclei of the cheeks' mucous membrane in woman with karyotype 47, XXX?

Practical work №3

Theme: Regularities of genes' inheritance that predetermine the manifestation of traits. Genotype as a system of interacting organism's genes.

Genetics - the science about the regularities of heredity and variation. Heredity is a property of organisms to repeat in several generations the similar traits and provide the specific character of individual development in certain environmental conditions. Due to the heredity parents and offspring have a similar composition of cells and tissues, the nature of metabolism, similar physiological functions, morphological features and other peculiarities. Due to the heredity each type of organisms reproduces itself from generation to generation.

The development of any organisms' traits is the result of the genes' interaction, rather is a consequence of the interaction between the products of their activity - proteins. There are allelic interaction of genes (complete and incomplete dominance, codomination, overdomination), and the interaction of nonallelic genes (complementarity, epistasis, polymerism). Consequence of the interaction of genes is the formation of phenotypic features of organisms.

To know:

1. Regularities of inheritance established by Mendel, and its cytological reasoning.
2. Forms of allelic genes' interaction (complete and incomplete dominance codomination, overdomination).
3. Forms of nonallelic genes' interaction (complementarity, epistasis, polymerism).
4. The concept of expressivity and genes' penetrance.
5. The phenomenon of multiple allelizm.
6. The phenomenon of genes' pleiotropism.
7. The genetics of blood groups (ABO system, MN) and Rh-system.

Be able to:

1. Determine the genotype and phenotype of offspring with the help of parent's and parents' genotypes with the help of children's genotype.

2. Apply the knowledge about the forms of genes' interaction to predict genotypes and phenotypes in next generations.

The audience work.

Solve the problems:

Task 1. The boy has a blonde mother and father is homozygote by the gene that causes dark hair. The gene that causes dark hair is dominant, and the gene that causes light hair is recessive. What is the possibility that this young man will have a blonde child if he marries a girl, who is heterozygote by the gene that determines hair color?

Task 2. The gene that causes the development of albinism is recessive and is localized in the autosome. It causes a lack of skin pigmentation in human. Dominant allele of this gene causes the normal skin pigmentation. A woman-albino delivered a child-albino from the man with normal skin pigmentation. What is the probability of albino's re-borning in this family?

Task 3. The mother has O (I), the father has AB (IV) blood group according to ABO system. What the genotypes and blood groups can their children have?

Task 4. How many types of gametes does the individual form:

a) homozygous for a dominant gene;

b) homozygous for a recessive gene;

c) heterozygous?

Task 5. How many types of gametes can individuals with following genotypes form: aabb, AaBb, AaBbCc if genes A, B and C are localized in different pairs of autosomes?

Task 6. The gene that causes webbing (fusion of fingers) is dominant and localized in the autosomes. A woman who has this defect got married with a man who has normal fingers. The couple has three children, two of whom had fusioned

fingers. What are the genotypes of parents and children?

Task 7. The gene that causes hondrodystrophy (violation of the skeleton) is dominant, localized in the autosomes and has full penetrance; homozygotes for this gene die before birth. Man and women have hondrodystrophy. Determine the probability of healthy child birth.

Task 8. The gene D, which causes brown eye color, is localized in the autosomes in human, it is dominant; the gene d, which causes blue eye color is recessive. Heterozygous brown eyed-woman got married with homozygous brown-eyed man. Can their child be blue-eyed?

Task 9. The gene A causes clubfoot in human, and gene a causes normal foot structure, gene D causes normal carbohydrate metabolism, gene d causes diabetes. Genes A and D are in different pairs of autosomes. A woman with normal foot structure and normal carbohydrate metabolism got married with toed men with diabetes. There were born three boys in this family, one of whom was the clubfoot, the second has diabetes, and the third has clubfoot and diabetes. Can be born a healthy baby in this family?

Task 10. The brown-eyed parents have three children. Two of them are brown-eyed with II and III blood groups; and one child is blue-eyed and has I blood group according to ABO system. What is the probability of child birth with IV blood group in this couple?

Task 11. The Rh-negative woman with I blood group according to ABO system got a child with III blood group. The child had hemolytic disease due to Rh disease. What are the possible genotypes in the husband of this woman?

Task 12. The reason of congenital blindness may be abnormalities of lens and cornea. Genes that cause these abnormalities are recessive and are localized in different pairs of autosomes. What is the probability of healthy child birth in a family where the father is blind as a result of abnormalities of lens, and is heterozygote for the gene that causes corneal anomaly and mother is blind through corneal anomaly and is heterozygote for the gene that causes the anomaly of the lens?

Questions for the interview:

1. What is the studding subject of the genetics?

2. What is a gene, a genotype and a phenotype?

3. What are the allelic genes?

4. What are homozygote, heterozygote and hemizyhota?

5. What are the Mendel's traits?

6. What is the analyzing crossing?

7. What is the difference between monohybrid, dihybrid and polihybrid crossing?

8. What genotypes can have the organism with a trait that is controlled by a dominant gene?

9. What genotypes can have the organism with a trait that is controlled by a recessive gene?

10. Can allelic genes be linked?

11. Which cells can not simultaneously have two any allelic genes normally?

12. What is the hypothesis of frequency gametes?

13. How do you understand the concept of "interaction genes"?

14. What are the features of allelic and nonallelic genes interaction in human?

15. What traits do inherit by the type of incomplete dominance in human?

16. What trait is the result of genes coddominance in human?

17. By what form of interaction of allelic genes by dominant allele expression of phenotypes will be stronger in the heterozygous state than in homozygous?

18. What form of interaction between nonallelic genes do have phenotypes effects opposite to those that arising in the case of complementarity?

19. What form of interaction of nonallelic gene can provide the affecting of different dominant genes on the same trait, intensify its manifestation?

20. What trait is caused by the interaction of genes' complementarity in human?

21. What is a pleyotropiya?

22. What are multiple alleles? When do the multiple alleles occur?

23. Which genes do as multiple alleles occur?

24. What is a penetrance?

25. What is a expressivity?

26. What are blood groups according to the MN system?

27. Can the blood of Rh-positive and Rh-negative people be compatible with? Does Rh disease appear in the mother?

28. What are the lethal genes?

Practical work №4

Theme: Full and partial clutch gene. Inheritance, linked with sex.

Localized in one chromosome genes that are inherited together are called coupled. The set of genes, located in the same chromosome, T.Morhan called a group of linked genes.

Clutch genes can be violated as a result of crossing-over. Thanks to crossing-over offspring appears with new combinations of traits. The frequency of crossing-over depends on the distance between genes, which is expressed by a percentage of crossing-over, or Morganides (in honour of T.Morhan, who created the chromosome theory of heredity).

Coupled with a sex are called traits that depend on genes, located in the sex chromosomes.

To know:

1. The position of the chromosome theory of heredity, its main point.
2. Peculiarities of inherit traits linked with sex.
3. Dependence on once of sex characteristics.
4. Limited sex signs.

Be able to:

1. Predict the inheritance of traits in live organisms with complete and incomplete clutch genes.
2. Determine the possibility of manifestation of traits in offspring by inheritance, coupled with sex.

The audience work.

Solve the problems:

Task 1. There are genes *A* and *B* in a distance of 6 Morganides in one of the chromosomes in Drosophila. In homologous chromosomes are genes *a* and *b*. Determine the percentage of gametes of Drosophila, which contains genes *A* and *b*?

Task 2. The distance between genes *A* and *B* is 6 Morganides between *A*

and *C* - 8 Morganides. What is the distance between genes *B* and *C*?

Task 3. Cataract (clouding of the lens) and polydactyly were caused dominant brand coupled genes. Recessive alleles of these genes determine the normal state of lens. One woman's autosome contains recessive alleles of these genes, and other - dominant. Her husband is healthy. Determine the possibility of birth of a healthy child in a married couple.

Task 4. The gene, which causes the presence of the Rh factor, and a gene, that determines the shape of red blood cells, are in one autosome in a distance of 3 Morganides. The woman received from her father a dominant gene *Rh* (causes the presence of Rh-factor) and dominant gene *E* (defines elliptical erythrocytes) and from mother - recessive alleles of these genes: *rh* (responsible for the absence of Rh-factor) and *e* (responsible for the normal form of erythrocytes)). Her husband is Rh-negative and has normal form red blood cells. Determine the possibility of birth of Rh-negative child with erythrocytes of normal form in a family.

Task 5. The gene that causes color blindness localizes in the X chromosome and is recessive. Determine the possibility of having children with color blindness, if a woman has normal vision, and her father is color-blind. In the family of a man there were not cases of color blindness.

Task 6. Determine what types of gametes and in what the percentage of individual forms genotype *ASas* if genes *A* and *C* are in the same group cohesion in a distance of 8 Morganides.

Task 7. Drosophila has short wings and a black body - recessive traits; long wings and grey body - dominant. During the crossing of dyheterozyhous females with a male, who is homozygote by two recessive genes, were received 42% of flies with a grey body and long wings, 41% - with a black body and short wings, 9% - with a grey body and short wings, 8% - with black body and long wings. Determine are genes linked that determine the length of the wings and the color of the body. Justify the answer making the necessary note.

Task 8. Cataract and polydactyly are determined by dominant and

localized in one autosome closely linked genes. The man has cataracts and polydactyly. His wife and sister are healthy. Determine the possibility of the birth of a healthy child in the family.

Task 9. Drosophila has genes that have the influence on the eye color and the color of the body; they are in the same group of cohesion and in a distance of 5 Morganides. Dominant genes cause a red color of the eyes and grey color of the body. Homozygous dominant genes for male crossed with black females, which had purple eyes. Their offspring were selected only males and crossed them with the homozygous recessive trait for females. Determine the genotypes and phenotypes of offspring, which appeared as a result of it, and the relationship between them (in percentage).

Task 10. When crossing normal females of Drosophila, which is the bearer of two located in one X chromosome recessive genes (one of them determines the development of truncated wings, and another - the striatum), with a male, which is available in the genotype dominant alleles of genes that determine the shape of the wings and body color was obtained offspring: 500 females with normal wings and normal body; 215 males with shortened wings and a striped body; 213 males with normal wings and normal body, 20 males with normal wings and striped body. Determine the distance between the genes, which determine the color of the body and the shape of the wings in Drosophila.

Task 11. Human has the gene that determines the dark color of the eyes is dominant and localized in the autosomes. Recessive gene that determines the development of color blindness is localized in the X chromosome. In a family where the father has dark eyes and suffers from color blindness, and his mother has dark eyes and normal vision, colorblind son was born with light eyes. What is the possibility of birth of a girl with phenotypic features of her brother in the family?

Task 12. Hemophilia and color blindness are caused by different recessive genes are localized on the X chromosome. The distance between them is 10 Morganides. Healthy woman inherited her father's chromosome with two recessive genes, married with a healthy husband. Determine the possibility of birth

of healthy children in this family.

Task 13. The gene, that causes hemophilia, is recessive and localized on the X chromosome. In a healthy woman brother suffers from hemophilia. Can the son of such a woman be born with hemophilia?

Questions for the interview:

1. What is linked inheritance?

2. What way are genes inherited, located in one chromosome?

3. What are the names of chromosomes, which have allelic genes? Can allelic genes be linked?

4. What depends on the phenomenon of linked inheritance of traits? What are the main positions of the chromosome theory of heredity?

5. Who is the founder of the chromosomal theory? What are the main positions of the chromosome theory of heredity?

6. What is the name of the unit of distance between genes?

7. What is crossing over? What is its biological meaning?

8. What does the frequency of crossing-over determine?

9. What does the number of groups of genes clutch determine?

10. In what quantity (in percentage) gametes the autosome genes, the distance between them is 15 Morganides, remain coupled?

11. What information does the genetic map of chromosomes contain?

12. What does in the construction of genetic maps of chromosomes underlie?

13. What is the mapping of chromosomes for?

14. What signs are called coupled with sex?

15. What are the common feature characteristics that depend on genes that are in the Y-chromosome?

16. What sex is called homohamete and what - heterohamete?

17. What is the difference between sex chromosomes?

18. How is your father's genotype that is located in the X- or the Y-

chromosome for a gene called?

19. What is the common feature of traits that depend on genes located on the X-or Y-chromosome?

20. What role does the Y-chromosome in human ontogenesis play?

21. What is the main role of abnormality of the girl's karyotype, which discovered hemizyhous on the gene, which is responsible for the ability to recognize the colors?

22. Which sex has a child with a normal karyotype, homozygous on the gene that causes normal blood clotting?

23. What are the signs called holandric?

24. What traits are called restricted by sex? Give examples of such traits.

25. What are the traits addicted from the sex? Give examples of such traits in humans.

Practical work №5

Theme: Population and statistical method. Medic and genetic counseling.

There is a wide range of methods that are applied in human genetics, including those which help to characterize the genofond of human populations. Thus, the distribution of certain genes in people's populations is studied by the population and statistical method; it also helps to determine the genetic structure of populations (correlation between the frequency of homozygotes and heterozygotes). Using the Hardy-Weinberg's law, we can analyze the prevalence of certain genes in different human populations; the frequency of occurring the heterozygous carriers of those recessive genes that cause certain hereditary diseases in homozygotes.

Genetic counseling is realized to provide specialized genetic help for patients.

To know:

1. Genetic characterization of populations.
2. The law of population's genetic equilibrium.
3. The use of population and statistical method in medicine.
4. Principle of medical and genetic counseling organization.

Be able to:

1. Analyze the genetic structure of populations.
2. Determine the concentration of allele and genotype frequencies in the population's genofond using the Hardy-Weinberg law.
3. Determine the optimal methods of genetics for various hereditary diseases' diagnosis.
4. Distinguish the field of application of population and statistical method and genetics method that are used in medic and genetic counseling.

The audience work.

Solve the problems:

Task 1. In particular human population, the number of which is 1000 people, there are no representatives with the first blood group according to the ABO system. 640 individuals have the second blood group. How many people in this population do III and IV blood group have?

Task 2. The blood group according to the MN system depends on the combination of codominant genes C^M and C^N; 16% people of certain population have N blood group. Determine the percentage of people with MN and N blood group.

Task 3. The gene that determines the color of dark eyes in mice localized in the autosomes and is dominant in relation to the gene that determines their light color. There are 51% of individuals with dark eyes in certain populations of mice. Define the percentage of heterozygous carriers of the gene that determines the color of light eyes.

Task 4. The gene that determines the presence of Rh positivity blood (Rh +) in human is dominant and localized in the autosomes. The concentration of the gene that determines the presence blood's Rh negativity in a particular population is 0.4. Identify in it: a) the percentage of heterozygous carriers of the gene that determines the Rh blood negativity b) the percentage of people with Rh-positive blood.

Task 5. Sickle-cell anemia is caused by a recessive gene, which is localized in the autosomes. People who are homozygotes for this gene suffer from hard form of sickle-cell anemia and die in early childhood. Explain why gene of sickle-cell anemia does not disappear from the genefond of the human population?

Task 6. The dark eye color is caused by dominant gene in human; it is localized in the autosomes. Determine the percentage of dark-eyed people in the population, where the concentration of the gene that determines the color of dark eyes is 0.6.

Task 7. The dark hair is caused by dominant gene in human; it is localized in

the autosomes. Determine the percentage of blond people in the population, where the concentration of the gene that determines the color of dark hair, is 0.5.

Task 8. The concentration of the recessive allele of the gene that determines the development of Owren disease (autosomal recessive disease) in a caertain population is 0.5. Determine the number of the heterozygous carriers of this gene in the population, which has 10,000 people.

Task 9. The gene that determines the development of albinism is recessive. It is localized in the autosomes. Its concentration in some African populations is 0.1. Determine: a) the percentage of heterozygous carriers of the gene that causes albinism, and b) the percentage of people who are healthy.

*Task 10.*The hereditary methemoglobinemia is determined by a recessive gene, which is localized in the autosomes. Such kind of the pathology in the population of Alaska occurs with the frequency of 0.09%. Determine the percentage of heterozygous carriers of the gene that causes methemoglobinemia.

Task 11. Alkaptonuria – is a genetic pathology, which is defined by the recessive gene that is localized in the autosomes. The children, who are suffer from alkaptonuria in certain populations are born with the frequency of 1:100 000. Define in this population: a) the percentage of heterozygous carriers of the gene that causes this pathology, b) the percentage of people who are healthy.

Questions for the interview:

1. What is the genetic characteristic of the population?
2. What is the ecological characteristic of the population?
3. What are the populations with number in 1500-4000 people called?
4. What are the populations with number less than 1500 people called?
5. What is the ideal population characterized?
6. What is the real population characterized?
7. What is the allelfond of population?
8. What kinds of insulation do you know?
9. What are the different types of biological isolation?

10. What is the geographic isolation?

11. Who and when discovered the law of maintaining of genetic equilibrium in a population?

12. What factors do lead to a genetic polymorphism in human populations?

13. Do all mutations change the genefond of the population?

14. Do somatic mutations change the genefond?

15. What is the elementary unit of evolution?

16. What the basic evolutionary factors do you know?

17. What are the population waves?

18. Who proposed the term "waves of life" at first?

19. Which equation does describe the law of genetic equilibrium?

20. Does migration of the genes have the matter in human populations?

21. Why the type can not be a unit of evolution?

22. What are the functions of the medical and genetic counseling?

23. What is the main point of medical and genetic counseling?

24. What kind of the successive stages of medical and genetic counseling do you know?

25. What is the prenatal diagnosis of hereditary diseases?

26. What kind of the modern methods of prenatal diagnosis do you know?

27. Why do we use the methods of prenatal diagnosis?

28. What is an amniocentesis?

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