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Chair of Medical Biology and Genetics

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**COLLECTION OF TASKS
ON MEDICAL GENETICS**

For overseas students in English medium



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Предложенный сборник включает задачи, решение которых предполагает закрепление теоретического материала по следующим темам: «Закономерности наследования признаков при моно- и полигенном наследовании. Формирование фенотипа как выражение единства генетических и средовых факторов», «Хромосомный и геномный уровни организации наследственного материала у про- и эукариот», «Наследственные болезни человека». Задачник способствует овладению приемами решения задач по курсу медицинской генетики. Полученные навыки могут быть использованы врачом общей практики и в медико-генетической консультации.

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ПРЕДИСЛОВИЕ

В последние годы генетика становится одним из главных разделов биологии. Повышение интереса к генетике обуславливает необходимость издания учебных пособий, которые способствовали доведению до студентов в доступной форме специальных знаний.

Основной целью данного пособия является оказание помощи в решении задач по генетике, что будет способствовать усвоению и закреплению знаний. Полагаем, это поможет освоить, закрепить и усовершенствовать навыки генетического анализа при изучении генетики.

В данном сборнике приводятся примеры решения типовых задач, предлагаются задачи, направленные на закрепление теоретических знаний.

Материал сборника систематизирован в соответствии с программой изучения основ общей генетики.

Сборник задач включает 103 задачи по следующим темам: моногибридное скрещивание, промежуточное наследование, ди- и полигибридное скрещивание, взаимодействие генов, сцепленное наследование, пенетрантность, популяционная генетика, анализ родословных. При подготовке пособия использованы учебники и учебные пособия, приведенные в списке литературы, а также накопленный авторами практический опыт проведения практических занятий.

Необходимость издания сборника задач на английском языке обусловлена тем, что при организации учебного процесса по медицинской биологии и генетике со студентами факультета по подготовке специалистов для зарубежных стран, обучающихся на английском языке, одной из трудностей является отсутствие учебно-методической литературы.

Introduction

During last years the genetics has become one of the main sections of biology. Increase of interest in genetics causes the necessity of editing manuals which easily promote informing of students.

The main purpose of this manual is rendering assistance in the solution of tasks on genetics. We believe, it will help in mastering, fixing and improving skills of the genetic analysis in process of studying genetics.

In the given collection examples of the solution of typical tasks are resulted, tasks for independent decision, directed on fastening of theoretical knowledge are offered. The material of the collection is systematized according to the program of general genetics.

The collection of tasks includes 103 tasks on the following topics: monohybrid crossing, intermediate inheritance, di- and polyhybrid crossing, interaction of genes, associated inheritance, penetration, population genetics, the analysis of family trees. During preparation of the manual textbooks, mentioned in the list of the literature, were used as well as the practical experience of the authors.

The necessity of creating a manual on English is acute, because one of the difficulties is the absence of teaching and methodical literature for overseas students.

PRINCIPLES OF ACCOMPLISHING TYPICAL TASKS

1. Rules of inheritance in case of mono- and polyhybrid crossing

Task 1.1.

How many and what types of gametes do the individuals with following genotypes form?

a) P: AA P: AABB P: AABBCC
 G: A G: AB G: ABC

(In a gamete there is one gene from a pair, and individuals give only one type of gametes, because they are homozygous).

б) P: Aa P: AaBB P: AaBBCC
 G: A a G: AB aB G: ABC aBC

в) P: AaBb P: AaBBCc P: AaBbCc P: AaBbCcDd
 G: AB Ab G: ABC ABc number of gametes number of gametes
 aB ab aBC aBc 2^3 2^4

Task 1.2.

In tomatoes the gene causing red color of fruits dominates over a gene of yellow color. What will be the color of fruits in plants received by crossing heterozygous plants with red fruits and plants with yellow fruits.

(Terms of the task are tabulated).

Attribute	Gene	Genotype
Red color of fruits	A	A – (AA, Aa)
Yellow color of fruits	a	aa

Genetic record of fulfillment:
 P: Aa x aa
 G: A a a
 F: Aa aa
 red fruits yellow fruits

The answer: 50% of plants appear erythrocarpous (red color), 50% — xanthocarpous (yellow color).

Task 1.3. a) In a person a gene causing one of the forms of hereditary deafness is recessive in relation to a gene of normal hearing. What probability is possible to expect from a marriage of heterozygous parents?

Attribute	Gene	Genotype
Congenital deafness	b	bb
Normal hearing	B	B-

P: ♀Bb x ♂Bb
 G: B b B b
 F₁: BB, Bb, Bb, bb

The answer: probability of a birth of a healthy child is 75%, of an ill one — 25%.

6) *Deaf-and-dumb* child is a result of a marriage between *deaf-and-dumb* woman and a *normal* man. What are the genotypes of the parents?

P: bb x Bb
G: b B b
F ₁ : BB, bb

Mother is homozygous, as *congenital deafness* is a recessive attribute. The genotype of the father can be either BB or Bb: in both cases he will be healthy. But the child was born ill, so, one recessive gene he had received from the mother, and another one he should receive from the father. The genotype of the father — **Bb**.

The answer: the genotype of the mother is **bb**, the genotype of the father is **Bb**.

2. Interaction of genes

1. Incomplete domination

Task 2.1.

One of the spouse suffers from *acatalasia*, another one has *lowered activity of catalase*. What are possible phenotypes of their children?

Attribute	Gene	Genotype
Acatalasia (absence of catalase in blood)	a	aa
Presence of catalase	A	AA
The reduced maintenance of catalase	A, a	Aa

P: ♀aa x ♂Aa
 G: a A a
 F₁: Aa, aa

The answer: 50% of children will suffer from *acatalasia*, 50 % will have the reduced maintenance of catalase.

2. Plural allelism, codomination

Task 2.2.

The heterozygous woman with II blood group has married the heterozygous man with III blood group.

What blood groups can children have?

Attribute	Gene	Genotype
I blood group	I ⁰	I ⁰ I ⁰
II blood group	I ^A	I ^A I ^A , I ^A I ⁰
III blood group	I ^B	I ^B I ^B , I ^B I ⁰
IV blood group	I ^A , I ^B	I ^A I ^B

P: ♀I^A I⁰ x ♂I^B I⁰
 G: I^A I⁰ I^B I⁰
 F₁: I^AI^B, I^AI⁰, I^BI⁰, I⁰I⁰

The answer: 25% of children can have I blood group, 25% — II, 25% — III, 25% — IV blood group.

3. Complementarity

Task 2.3.

Deafness can be caused by recessive by genes **d** and **e**, which lay in different pairs of chromosomes. For normal hearing presence of both dominant genes is necessary.

A deaf woman (DDee) marries a deaf man (EEdd).

Determine, whether there will be deaf children.

Attribute	Gene	Genotype
Deafness	d, e	ddee, D-ee, ddE-
Normal hearing	D, E	D-E-

P: ♀ DDee x ♂ ddEE

G: De dE

F₁: DdEe

The answer: all children will have normal hearing.

4. Epistasis

Task 2.4.

The majority breeds of hens have coloured plumage, determined by gene **C**, and white plumage, determined by gene **c** (recessive allele). The breed *leghorns* has epistatic gene **I**, which overwhelms the development of a pigment (even in the presence of the gene **C**). Gene **i** (its allele) doesn't make an impact on gene **C**.

Two white deheterozygous individuals are crossed.

Determine the segregation of a phenotype of descendants.

Attribute	Gene	Genotype
Coloured plumage	C	C-ii
White plumage	c	C-I-, ccI- ccii

P: ♀ CcIi x ♂ CcIi

G: CI Ci CI Ci

cI ci cI ci

F₁: 9 C-I-, 3 C-ii, 3 ccI-, 1 ccii

The answer: 81,25% of white hens, 18,75% of coloured ones.

5. Polymeria

Task 2.5.

Growth of a person is supervised by three pairs of unlinked genes, which cooperate on type of polymeria. The most undersized people have all recessive genes and growth of about 150 cm, the highest — all dominant genes and growth — 180 cm.

Undersized homozygous woman married high homozygous man.

What growth will their children have?

Attribute	Gene	Genotype
High growth (180 cm)	A ₁ , A ₂ , A ₃	A ₁ A ₁ A ₂ A ₂ A ₃ A ₃
Low growth (150 cm)	a ₁ , a ₂ , a ₃	a ₁ a ₁ a ₂ a ₂ a ₃ a ₃

P: ♀ a₁a₁a₂a₂a₃a₃ x ♂ A₁A₁A₂A₂A₃A₃

G: a₁a₂a₃ A₁A₂A₃

F₁: A₁a₁A₂a₂A₃a₃

The answer: All children will be 165cm long.

6. A lethal gene

Task 2.6.

In the genome of mice the gene of prepotent yellow pigmentation of a hair have a recessive lethal effect (genotype **AA** causes the death of an embryo). Its allele **a** determines black recessive pigmentation and provides viability of mice.

Two yellow individuals are crossed.

What segregation of coloring is expected in F₁?

Attribute	Gene	Genotype
Yellow hair	A	Aa
Black hair	a	aa
Individual lacking vitality		AA

P: ♀ Aa x ♂ Aa

G: A a A a

F₁: AA, Aa, Aa, aa

The answer: 25% of individuals will die, 50% of individuals will be with yellow hair, 25% — with black hair.

3. Adhesion of genes

Task 3.1.

How many and what types of gametes are formed at flies drosophils with following genotypes:

a) P: A B

a B

G: A B a b

A b a B

б) P: AB

aB

G: AB ab

Task 3.2.

The classical hemophilia and daltonism are inherited as recessive, linked with a sex. The distance between genes is 9,8 Morgan units.

The woman, whose mother suffered from daltonism and father - from hemophilia, marries the man who suffers from both of diseases.

What is the probability of a birth of a child with both diseases?

Attribute	Gene	Genotype
Hemophilia	X ^h	X ^h X ^h , X ^h Y
Normal coagulability	X ^H	X ^H X ^H , X ^H X ^h , X ^H Y

P: ♀ $\frac{X^H X^d}{X^h X^D}$

♂ $\frac{X^h X^d}{Y}$

Daltonism	X^d	X^dX^d, X^dY
Normal sight	X^D	X^DX^D, X^DX^d, X^DY

G:	$\frac{X^HX^d}{X^hX^D}$	$\frac{XhXd}{Y}$		
F1:	$\frac{X^HX^d}{X^hX}$	$\frac{X^hX^D}{X^hX^d}$	$\frac{X^HX^D}{X^hX^d}$	$\frac{X^hX^d}{X^hXd}$
	$\frac{X^HX^d}{Y}$	$\frac{X^hX^D}{Y}$	$\frac{X^HX^D}{Y}$	$\frac{X^hX^d}{Y}$

In the organism of a woman 4 types of gametes are formed, 2 of them are crossing-overed. As the distance between genes is 9.8 Morgan unit, the amount of these gametes will be 9,8% or 4,9% on each type. In the F1 there will be 4,9% of cross-overed female individuals and as much as male ones, half of them (2,45%) will have both anomalies.

The answer: probability of a birth of a girl and a boy with both anomalies is in 2,45%.

TASKS

Monohybrid crossing

1. *Nyctalopia* (*night blindness*) in some cases is hereditary conditioned and is determined by dominant gene N. The woman suffering *nyctalopia* has married a healthy man. All children (6) have inherited this disease. The native sister of the woman (also suffering *nyctalopia*) has married a healthy man and from this marriage three healthy children and one with *night blindness* were born.

What are the genotypes of sisters and their parents (they suffered *nyctalopia*, too)?

2. In a marriage between healthy cousins 5 children were born. Three of them suffered *pentoseuria* and died in the age of 14.

What gene (dominant or recessive) is this disease determined by?

Is there a danger of transfer of this disease to further generation from a marriage between survived healthy children and healthy persons who are:

- not consisting in relationships;
- consisting in relationships?

3. In a family two acranial children (they died at once after birth), and one normal were born. Further his child was born acranial.

Give a genetic explanation to this phenomenon.

4. *Myoplegia* is transferred as dominant attribute.

Determine the probability of a birth of children with anomalies in a family where father is heterozygous and mother does not suffer *Myoplegia*.

5. *Phenylketonuria* is inherited as recessive attribute.
What children can be born in a family, where parents are heterozygous?
6. The gene of *polydactyly* of a person dominates over a normal structure of a hand.
a) Determine the probability of a birth of six-fingered children in a family, where both parents are heterozygous.
b) In a family where one of the parents has a normal structure of a hand and another one is six-fingered, a child with a normal structure of a hand was born. Determine the probability of a birth of the next child without anomalies.
7. Gene causing one of the forms of hereditary surdomutism of a person is recessive in relation to a gene of normal hearing.
a) What posterity is it possible to expect from a marriage between heterozygous parents?
b) The deaf-and-dumb child was born from a marriage between deaf-and-dumb woman and normal man. What are the genotypes of the parents?
8. One of the forms of hemeralopia is inherited as dominant attribute.
a) What is the probability of a birth of a children suffering hemeralopia from heterozygous sick parents?
b) What is the probability of a birth of children with analyzed anomaly in family where one of the parents suffers from night blindness, and another doesn't suffer (both spouses are homozygous)?
9. Galactosemia is inherited as autosomal recessive attribute. Successes of modern medicine allow to warn the development of illness and to avoid heavy consequences of infringement of exchange. Determine the probability of a birth of sick children in family where one of the spouses is homozygous on gene of galactosemia (but the development of illness was prevented by a diet) and the second is heterozygous on galactosemia.
10. *Wilson's illness* is inherited as recessive autosomal attribute. Calculate the probability of a birth of sick children in family where one of the spouses suffers from analyzed disease, and another is healthy (his parents, brothers and sisters were healthy, too).
11. Albinism is inherited as autosomal recessive attribute. Determine the probability of a birth of healthy children in family where one of parents suffers from albinism and another is normal (only the father of sick parent has this anomaly)?
12. Alkaptonuria is inherited as autosomal recessive attribute. Determine the probability of a birth of sick children in the family, where one of the parents is heterozygous, and another is homozygous.
13. The infant form of amaurotic familial idiocy is inherited as autosomal recessive attribute and comes to an end usually fatally by 4-5 years. The first child in family has died of analyzed illness when another one should be born. What is the probability that the second child will suffer the same illness?

14. A shoulder-omohyoid form of *myodistrophy* is inherited as dominant autosomal attribute. Determine the probability of disease of children in a family where both parents suffer from this anomaly, but one of them is homozygous and another is heterozygous.

15. Albinism is inherited as autosomal recessive attribute. In a family where one of spouses is an albino and another is normal, hetero-ovular twins were born. One of them is normal concerning analyzed illness, and another is an albino. What is the chance of a birth of the next child with albinism.

16. Parahemophilia is inherited as autosomal recessive attribute. Settle the probability of a birth of children with anomaly in family where both spouses suffer from parahemophilia.

17. One of the forms of agammaglobulinemia is inherited as autosomal recessive attribute, combined with almost full absence of lymphoid tissue. In a family the child with agammaglobulinemia was born. His parents are healthy. What is the probability of a birth of the next healthy child?

18. The late degeneration of a cornea (develops after 50) is inherited as dominant autosomal attribute.

Determine the probability of occurrence of disease in the following family. Grandmother and grandfather (on a mother's line) and all their relatives (who have lived till 70 years) suffered from indicated. On a father's line all ancestors were healthy.

19. Achondroplasia is transferred as dominant autosomal attribute. In a family, where both spouses suffered achondroplasia, the normal child was born. Settle the probability that the following child will be normal.

20. Absence of small molars is inherited as dominant autosomal attribute. Determine the probability of a birth of children with anomaly in family, where both parents are heterozygous to analyzed characteristic.

21. Afibrinogenemia is inherited as autosomal recessive attribute. In a family where parents are healthy, the child with attributes of afibrinogenemia was born. Find out the probability of a birth of the second child with the same illness.

22. Merge of the bottom dairy cutters is inherited as autosomal dominant attribute. In one family baby with bottom dairy cutters grown together was born. Parents do not remember, whether they had such anomaly. Find out possible genotypes of parents. For each variant calculate probability of a birth of the following child without anomaly.

23. Syndactyly is inherited as dominant autosomal attribute. What is the probability of a birth of children with accrete fingers in a family where one of parents is heterozygous to an analyzed attribute, and another has a normal structure of fingers.

24. The dominant gene D causes anomaly in the development of a skeleton — cranial-clavicular dysostosis.

a) The woman with a normal structure of a skeleton married the man with cranial-clavicular dysostosis. The child from this marriage had a normal structure of a skeleton. Specify, whether it is possible to determine a genotype of his father judging by the phenotype of the child.

б) The woman suffering cranial-clavicular dysostosis married the man with a normal structure of a skeleton. The child from this marriage inherited defect of a skeleton from mother. Is it possible to determine a genotype of the mother?

в) Both parents suffer from cranial-clavicular dysostosis. The child from this marriage has a normal structure of a skeleton. Find out genotypes of both parents and the child.

25. Recessive gene s determines congenital surdomutism.

a) Congenitally deaf-and-dumb man married the woman with normal hearing. Their child has normal hearing. Is it possible to determine a genotype of the mother?

б) Congenitally deaf-and-dumb woman married the man with normal hearing. The deaf-and-dumb child was born. Determine genotypes of parents.

26. Family myoplegia is caused by dominant gene M .

a) The sick man (heterozygotic on a gene of family myoplegia) married the healthy woman. What is the probability of a birth of the sick child?

б) The husband and the wife are relatives. Both of them are heterozygotic on gene M . Determine the probability of a birth of a sick child.

27. Phenylketonuria is inherited as recessive attribute (p).

a) Parents are heterozygous on a gene of phenylketonuria.

б) The wife is heterozygotic on a gene of phenylketonuria, and the husband is homozygous on a normal allele of this gene.

Specify the probability of a birth of a sick child.

Intermediate inheritance

28. The rare gene a causes in the person hereditary anophthalmia. Its allele A causes normal development of eyes, heterozygotes have reduced eyeballs. Spouses are heterozygous on gene A . Determine gene splitting of phenotype and genotype in posterity. The man, heterozygotic on a gene A , married the woman with normal eyes. What splitting of phenotype is expected in posterity?

29. Dominant gene D determines the development of the stiffened and bent little finger on a hand. This gene in a heterozygotic condition causes development of such little finger only on one hand. Determine, whether the child with normal or two abnormal hands at parents faulty on a little finger only on one hand can be born.

30. Sickle-cell disease at natives of Africa is caused by prepotent gene S which in homozygotic condition causes death from an anemia. People with a genotype ss in local conditions perish from malaria, but they do not suffer from anemia. Heterozygotes (Ss) survive, as they do not suffer from anemia and malaria.

What is the share of viable posterity at heterozygous parents; at heterozygous mother and healthy father?

31. One of the forms of cystinuria is inherited as autosomal recessive attribute. But at heterozygotes there is only increased contents of cystine in urine, at homozygotes — formation of stones in kidneys.

a) Determine possible forms of occurrence of cystinuria at children in family where one of the spouses suffered from this disease, and another had only increased contents of cystine in urine.

б) Determine possible forms of display of cystinuria at children in family where one of the spouses suffered from illness, and another was normal.

32. Pelger's anomaly of segmentation of nucleus of leukocytes is inherited as autosomal dominant attribute. Segmentation of nucleus is absent at homozygotes, heterozygotes have normal segmentation of it.

a) Determine the character of the nucleus in segmental leukocytes at children in family, where one of the spouses has leukocytes with unusual segmentation of nucleus, and another is normal to this attribute.

б) Determine character of a nucleus in segmental leukocytes at children in family where one of the spouses has non-segmented nucleus of leukocytes and another has normal nucleus.

33. Thalassemia is inherited as not completely dominant autosomal attribute. In homozygote disease ends fatally in 90–95% of cases, and heterozygotes pass it in rather light form.

a) What is the probability of a birth of healthy children in family where one of spouses suffers from the easy form of thalassemia, and another is normal concerning analyzed attribute?

б) What is the probability of a birth of healthy children in family where both parents suffer from the light form thalassemia?

34. Acatasia is caused by rare autosomal recessive gene. In heterozygote activity of catalase is a little bit lowered.

a) In both parents and their singular son activity of catalase is below norm. Determine the probability of a birth of the following child without anomaly.

б) Determine probable phenotypes of children in family where one of the spouses suffers acatalasia, and another has only lowered activity of catalase.

35. Sickle-cell disease is inherited as not completely dominant autosomal attribute. Homozygous individuals die usually before puberty, heterozygous are viable (the anemia appears subclinically more often). Plasmodium vivax cannot

use S-hemoglobin for nourishment. Therefore people having this form of hemoglobin don't suffer from malaria.

a) Specify the probability of a birth of children, steady against a malaria, in family where one of the parents is heterozygous according to sickle-cell anemia and another is normal concerning this attribute.

б) Specify the probability of a birth of unstable to malaria children in family where both parents are steady against this parasite.

36. Family hypercholesterinemia is inherited dominant through autosome. In heterozygote this disease is shown in high contents of cholesterol in blood; in homozygote, besides that, xanthoma of skin and sinews, atherosclerosis develops.

a) Determine a possible degree of development of hypercholesterinemia at children in family where both parents have only high contents of cholesterol in blood.

б) Specify the probability of a birth of children with anomaly and a degree of its development in a family where one of the parents besides high contents of cholesterol in blood has advanced xanthoma and atherosclerosis, and another is normal concerning analyzed attribute.

37. Sickle-cell disease and thalassemia are inherited as two attribute with incomplete domination; genes are not linked among themselves and are localized in autosoma. In heterozygote both diseases don't carry the expressed clinical picture. In all cases carriers of a gene of thalassemia or sickle-cell anemia are steady against malaria. Anemia develops in double heterozygote. Homozygote on sickle-cell anemia and thalassemia in overwhelming majority of cases die in the childhood.

Determine the probability of a birth of healthy children in family where one of parents is heterozygous on sickle-cell anemia, but is normal on thalassemia, and another is heterozygous on thalassemia, but is normal concerning anemia.

Dihybrid and polyhybrid crossing

38. Brown color of eyes dominates over blue, and the ability to own the right hand better than left hand dominates above left-handedness. Genes of both attributes are in different pairs of chromosomes.

a) The brown-eyed right-handed person married the blue-eyed lefthander. What attributes is it possible to expect at children if the man is homozygous (heterozygous)?

b) The blue-eyed right-handed person married the brown-eyed lefthander. Their two children were the brown-eyed lefthander and the blue-eyed right-handed person. From the second marriage between this man and another brown-eyed right-handed person 9 brown-eyed children were born. All of them were right-handed persons. What are the genotypes of each of three parents?

c) The blue-eyed right-handed person (his father was lefthander) married the brown-eyed lefthander from the family where all members during several

generations had had brown eyes. What posterity concerning these two attributes is possible to expect from such marriage?

d) The brown-eyed right-handed person married the blue-eyed right-handed person. Their first child, the lefthander, also had blue eyes. What attributes will be at further descendants?

39. Blond and short-sighted men and the dark-haired woman with normal sight have 4 children: dark-haired with normal sight, blond short-sighted, dark-haired short-sighted and blond with normal sight.

Enter genetic designations and determine genotypes of parents and children.

40. Red-haired (dominant attribute) man without freckles and light-haired woman with freckles (dominant attribute) have 5 children.

Specify the probability of a birth of children with red hair and with freckles, and non-red without freckles.

41. In a person syndactyly and glaucoma (infringement of outflow of the intraocular liquid) are determined by autosomal dominant genes located in different pairs of chromosomes. The woman suffering glaucoma has married the man with syndactyly. Mother of this woman, as well as many of her relatives, suffered from glaucoma, and father was free from this disease. Mother of the man suffered from syndactyly, and all relatives of the father had no anomalies.

Determine the probability of a birth of the child with two diseases at once and the child with one of these hereditary infringements.

42. Surdomutism is inherited as autosomal recessive attribute, gout is inherited as dominant one. Both genes lay in different pairs of chromosomes.

Determine the probability of a birth of a deaf-and-dumb child with predisposition to a gout at a deaf-and-dumb mother, not suffering from gout, and a man with normal hearing and speech, suffering from gout.

43. Phenylketonuria and Swiss-type agammaglobulinemia (usually leads to death up to 6-month's age) are inherited as autosomal recessive attributes. Successes of modern medicine allow to avoid heavy consequences of infringement of the phenylalanine exchange.

a) What is the probability of a birth of healthy children in family, where both parents are heterozygous on both pairs of pathological genes.

b) Determine the probability of a birth of persons suffering phenylketonuria and hopes for rescue.

44. Fructoseuria has two forms. One proceeds without clinically expressed symptoms, the second leads to braking of physical and intellectual development. Both are inherited as recessive not linked together attributes. One of spouses has the increased contents of fructose in the urine, therefore, he is homozygous under fructoseuria, not shown clinically, but is heterozygous under the second form of disease. The second spouse in due time has received medical treatment

successfully under the second form of fructoseuria, but is heterozygous under its asymptomatic form. What is the probability of a birth of children suffering clinically expressed form of fructoseuria in this family.

45. People can have two kinds of blindness and both are determined by autosomal recessive gene. Genes of both attributes lie in different pairs of chromosomes.

a) Determine the probability that the child will be born blind if her father and mother suffer the same kind of hereditary blindness, and on other pair of genes of blindness are normal.

b) What is the probability of a birth of a blind child in family where father and mother suffer from different kinds of hereditary blindness, meaning, that on both pairs of genes they are homozygous?

c) Determine the probability of a birth of a child blind if the following is known:
— his parents are sighted;
— both grandmothers suffer an identical kind of hereditary blindness, and under other pair of analyzed genes they are normal and homozygous;
— in a family tree on the part of grandfathers hereditary blindness is not marked.

46. There are two forms of surdomutism in a person which are determined by autosomal recessive by not linked genes.

a) Specify the chance of a birth of deaf-and-dumb children in a family where mother and father suffer the same form of surdomutism.

b) Determine the probability of a birth of deaf-and-dumb children in a family where parents suffer different forms of surdomutism, and on the second pair of genes they are heterozygous.

47. The glaucoma of adults is inherited by several ways. One of forms is formed by dominant autosomal gene, another — by recessive autosomal one, not linked with the previous gene.

a) Specify the chance of a birth of the child with anomaly if both parents are heterozygous under both pairs of pathological genes.

b) Determine the probability of a birth of children with anomaly in family where one of parents is heterozygous under both pairs of pathological genes, and another is normal concerning sight and homozygous under both pairs of genes.

48. Parents heard well and one of them had smooth hair, and another — twisted. Their first child was deaf with smooth hair. Their second child heard well and had curly hair.

Define probability of further occurrence of deaf children with curly hair in this family. It is known, that the gene of curly hair dominates above the gene of smooth hair, deafness is a recessive attribute, and both pairs of genes lie in different chromosomes.

49. Polydactyly, myopia (near sightedness) and absence of small molars are transferred as dominant autosomal attributes. Genes of all three attributes are situated in different pairs of chromosomes.

a) Define the probability of a birth of children without anomalies in family where both parents suffer from all three lacks and are heterozygous under all three pairs of genes.

б) Determine the probability of a birth of children without anomalies in family about which the following is known: grandmother under wife's line was six-fingered, grandfather — short-sighted. Concerning other attributes they are normal. The daughter has inherited both anomalies from the parents, too. The grandmother by the husband's line had no molars, had normal sight and a five-fingered hand. The grandfather was normal concerning all three attributes. The son has inherited anomaly from mother.

50. Some forms of cataract and surdomutism are transferred as autosomal recessive not linked attributes. Absence of cutters and canines of the top jaw also can be transferred as recessive attribute which isn't linked with cataract and surdomutism.

a) Find out the probability of a birth of children with three anomalies in family, where both parents are heterozygous under all three pairs of genes.

b) Determine the probability of a birth of children with all three anomalies in family where one of parents suffers from cataract and surdomutism, but is heterozygous under third attribute, and the second spouse is heterozygous.

51. The cataract has different hereditary forms. The majority of them are inherited as dominant autosomal attributes, others — as autosomal recessive non-linked attributes.

Specify the probability of a birth of children with anomaly if both parents suffer from it it is dominant the inherited form, but heterozygous on it and still heterozygous on two recessive to forms of a cataract.

52. At the person anti-genes of **ABO** system are determined by plural alleles I^O , I^A , I^B ; rhesus anti-gene (Rh^+ and Rh^-) by alleles D and d ; MN-groups of blood — by co-dominant alleles $L^M L^N$ (alleles L^M and L^N produce three phenotypes: $L^M L^M$ — there is anti-gene M in erythrocytes, $L^N L^N$ — antigen N, at $L^M L^N$ — both antigens).

a) How many various phenotypes on three systems of blood groups are there in a person?

b) Genotype of mother is $I^A I^O L^M L^M Dd$, father's genotype is $I^B I^B L^M L^N Dd$.

How many and what combinations of antigens is possible to expect at their children?

c) Genotype of mother is $I^O I^O L^N L^N Dd$, of father is $I^A I^B L^M L^M dd$.

How many various phenotypes and what genotypes is it possible to expect at their children?

d) The woman having a phenotype A (Rh^-) MN (her father had I group of blood) has married the man, whose blood contains antigens AB (Rh^+) NN. His mother was Rhesus factor negative.

Establish the probability that the child will have the same combination of antigens as the father has.

e) Phenotype of the mother is ABMRh⁻, phenotype of the father - ONRh⁺. One of the father's parents was Rhesus factor negative.

Find out combinations of blood antigens at their children.

Interaction of genes

53. At the person ABO system of blood groups is caused by alleles of gene I. Recessive allele I^O determines I group of blood. Alleles $I^A I^B$, causing II and III groups of blood, dominate above allele I^O , and over the relation to each other are codominant, genotype $I^A I^B$ causes IV group of blood.

a) The woman having I group of blood has married heterozygotic man with II group. One child was born. What are his blood group and genotype?

b) The woman with I group has married heterozygotic man with III group. What groups of blood can their children have?

c) Mother is heterozygous on gene I^A and father - on gene I^B . What groups of blood and genotype does their child have?

d) The woman with I group has married the man with IV group. Will the children inherit mother's or father's group of blood?

e) Heterozygous woman with II group has married the man with I group. Establish what groups of blood can (can't) their children have.

f) Heterozygotic woman with II group has married for heterozygotic man with III group of blood. What group of blood and genotype can their children have?

54. At a person I group of blood is caused by recessive gene I^O , II group — by gene I^A , III group - by gene I^B . Genotype $I^A I^B$ causes IV group of blood.

a) Mother has I group of blood, father has II group, their child is with I group. Determine the genotype of the father.

b) The child has I group of blood, his mother has II group, father — III group. What are the genotypes of the parents?

c) Mother has II group of blood, father has III group. Is it possible to establish their genotype if their child has IV group of blood.

55. At the person system ABO is caused by alleles of gene I. Recessive allele I^O determines I group of blood. Alleles I^A and I^B , causing II and III groups of blood, dominate above allele I^O . Genotype $I^A I^B$ causes IV group of blood.

a) The husband and the wife are heterozygous and have II group. Settle the probability of a birth of the child with I, II, III and IV groups of blood.

b) The husband has I group of blood, the wife — IV group of blood. Determine the probability of a birth of the child with I, II, III and IV groups of blood.

c) Parents are heterozygous on III group of blood. Find out probability of a birth of the child with III group.

56. At the person antigens of system ABO are not only in erythrocytes, but also in other cells of a body. For some people (secretors) water-soluble forms of these antigens are excreted with saliva and other liquids. Other part of people (non-secretors), haven't got these antigens in saliva. ABO — groups of blood, are determined by plural alleles I^O , I^A , I^B ; and presence of antigens A and B in the saliva — by prepotent gene Se (Secretor).

a) Parents do not excrete antigens A and B in a saliva, their genotypes are $I^A I^B sese$ and $I^O I^O Sese$. What is the probability of a birth of the child excreting an antigen A in a saliva?

b) Mother has antigen B in erythrocytes, but does not contain it in saliva; father has antigen A in erythrocytes and saliva; their first child contains antigen A and B in erythrocytes, but does not contain it in saliva; their second child has I group of blood.

Determine genotypes of parents.

57. The so-called Bombay phenomenon is expressed that in family where father had I group of blood, and mother had the third, the girl with I group was born. She has married the man with II group, at them two girls were born: the first with IV, the second with I group. Occurrence of the girl with IV group of blood in the third generation from the mother with III and the father with I group of blood has caused bewilderment. However, in the literature some more similar cases have been described. Some geneticists explain this phenomenon: there is rare recessive epistatic gene capable to suppress action of genes determining group of blood (A and B).

a) Establish probable genotypes of all three generations described in the Bombay phenomenon.

b) Determine probability of a birth of children with I group in a family of the first daughter from third generation if she marries the same man on a genotype.

c) Determine probable groups of blood at children in family of the second daughter from the third generation if she marries the man with IV group of blood, but heterozygous on rare epistatic gene.

58. At the person distinction in colour of skin is caused basically by two pairs of independently splitted genes: $BBCC$ — black skin, $bbcc$ — white skin. Any three alleles of black skin give dark skin, any two — swarthy skin, one — light skin.

a) Determine genotypes of parents if two swarthy parents have black and white child.

b) Establish whether children at parents-negros can be born light-skinned.

c) Is it possible to expect a birth of more dark children from white parents; from light parents; from swarty, similar and dissimilar on a genotype?

59. According to some information, color of skin at the person is determined by 5 genes located in different loci not linked among themselves. At missed marriages of people with dark and light skin their children (mulattoes) have intermediate color of a skin.

Calculate the probability of a birth at two such mulattoes a child with white skin.

What is the probability of a birth of the child with black skin?

60. At the person distinction in colour of skin is caused basically by two pairs of independently splitted genes: BBCC — black skin, bbcc — white skin. Any three alleles of black skin give dark skin, any two — swarthy skin, one — light skin.

What are the genotypes of parents, if:

a) Both of them are swarthy and have one black and one white child.

b) Both are black and have the albino child.

c) Both are swarthy, all children are swarthy, too.

d) One of the parents is swarthy, another has light skin. Their children: 3 swarthy, 3 with light skin, 1 black and 1 white.

61. The person has some forms of hereditary short-sightedness. The moderate form (from — 2,0 up to — 4,0) and high form (higher than — 5,0) are transferred as autosomal prepotent attributes not linked among themselves. In family where mother was short-sighted, and father had normal sight a daughter and a son were born. The daughter had a moderate form of short-sightedness, the son had a high form of it.

Determine the chance of a birth of the next child without anomaly. Only one parent of the mother suffered from short-sightedness. It is necessary to know, that people having genes of both forms of short-sightedness, suffer from its high form only.

62. Growth of the person is supervised by several pairs of not linked genes which cooperate on polymeric type. It is possible to admit (if we neglect factors of environment and conditionally limit only to three pairs of genes), that in any population the most undersized people have all recessive genes and growth about 150 cm, the highest — all prepotent genes and growth of 180 cm.

a) What is the growth of people heterozygous on three pairs of genes?

b) The undersized woman has married the man of average growth. They had four children who had growth of 165 cm, 160 cm, 155 cm and 150 cm. Find out genotypes of parents and their growth.

The linked inheritance

63. At a person daltonism is caused by recessive gene linked with sex, the ability to distinguish taste of phenylthiourea (FTM) — by autosomal dominant gene T. People not distinguishing bitter taste of FTM have genotype *tt*.

The woman with normal sight, not distinguishing taste of FTM, has married the colour-blind person, capable to distinguish this taste. As a result: two daughters with normal sight who distinguished taste of FTM and four sons also with normal sight, but two of them didn't distinguish this taste.

Specify probable genotypes of parents and children.

64. Absence of sexual glands is handed down as recessive attribute linked with X-chromosome. The young man not suffering from this lack married the girl whose father had no sexual glands and mother (and her ancestors) were healthy.

Sons and daughters from this marriage will suffer absence glands, won't they? What is the probability?

65. Albinism is caused by autosomal recessive gene. Absence of sweat glands is shown as recessive attribute linked with sex. At one married couple, normal to these attributes, the son with both specified anomalies was born.

Genotypes of parents are...

66. At the person pseudo-hypertrophic muscular dystrophy resulting with death in the age of 10–20 years in some cases is caused by recessive gene linked with sex. Illness is registered only at boys.

Explain, why illness is registered only at boys. Why do patients die, not leaving posterity, and illness doesn't disappear from a population?

67. Graham with employees studied in Northern Carolina occurrence of persons with the lack of phosphorus in blood that is connected with the specific form of the rachitis which can't be treated by vitamin D. This disease is determined by prepotent gene. In posterity from a marriage of 14 ill men with healthy women 21 daughter and 16 sons were born. Daughters suffered from the lack of phosphorus in blood, sons were healthy.

What is genetic conditionality of this disease? How does it differs from hemophilia?

68. At the person harmless anomaly — presence of membrane between toes — is described. From a marriage between the woman with normal toes and the man, having a membrane, three children were born: the daughter was normal, and sons possessed anomaly.

One of sons, in a marriage with normal woman, had 6 daughters with normal toes and 4 sons with anomaly. Three from them had children and have transferred this defect to sons, but not to daughters. Wives of these men had no anomalies of toes.

Three from six healthy daughters have married normal men and had children with normal structure of toes (first daughter had 3 daughters and 2 sons, second and third had 1 daughter). Draw a family tree and try to determine character of inheritance of this anomaly. Explain, why anomaly is peculiar only

to men and why healthy men are born only in marriages where husband and wife are normal.

69. Aniridia (kind of blindness) depends on dominant autosomal gene, and optical atrophy (another kind of blindness) — on recessive gene, connected with sex and located in the X-chromosome. The man with optical atrophy married the woman with aniridia.

Determine possible phenotype of the posterity. Can a child (a son or a daughter) with both kinds of blindness be born?

70. Hypoplasia of dental enamel is inherited as dominant attribute connected with the X-chromosome. In a family where both parents suffered from hypoplasia the son with normal teeth was born.

What will be their second son?

71. Human dysplasia is transferred as recessive attribute linked with X-chromosome.

a) The young man without this lack married the girl whose father had no sweat glands but mother and her ancestors were healthy.

What is the chance that children from this marriage will have absence of sweat glands?

b) The healthy woman marries the man ill with dysplasia. The sick girl and the healthy son are born.

Can their next child be born without anomaly?

72. Dimness of teeth can be determined by two dominant genes, one of which is located in autosomes, another — in the X-chromosome. In a family with parents having dark teeth, daughter and son with normal color of teeth were born.

It was possible to establish, that dark teeth of mother were caused only by gene linked with the X-chromosome, and a dark teeth of father — by autosomal gene, on which he was heterozygous. Can their next child be born without anomaly?

73. One of forms of agammaglobulinemia is inherited as autosomal recessive attribute, another — as recessive attribute linked with X-chromosome.

Mother is heterozygous on both pairs of genes and father is healthy and has only dominant genes of analyzed alleles. What is the chance that their children will be sick.

74. Daltonism is caused by recessive gene linked with the X-chromosome. Thalassemia is inherited as autosomal dominant attribute and has two forms: heavy (at homozygote), and less heavy (at heterozygote). The woman with normal sight, but with light form of thalassemia in a marriage with healthy but colour-blind man, has the colour-blind son with the easy form of thalassemia.

What is the probability of a birth of the following son without anomalies?

75. At the person albinism is caused by autosomal recessive gene. Dysplasia is transferred as recessive attribute linked with the X-chromosome. At

a married couple, normal on both attributes, the son with both anomalies was born.

What is the chance that their second child will be the girl (son), normal on both attributes?

76. Hypertrichosis is transferred by Y-chromosome, and polydactyly — as dominant autosomal attribute. In a family where father had hypertrichosis, and mother — polydactyly, normal daughter was born.

What is the chance that the following child in this family will also have both anomalies?

77. Pigmentary retinite can be inherited in three ways: as autosomal dominant, recessive and recessive attribute linked with X-chromosome.

Determine the probability of a birth of sick children in a family where mother is sick with pigmentary retinite and is heterozygous on all three pairs of genes, and father is healthy and normal to all three attributes.

78. Brown color of eyes and skill to own mainly right hand are autosomal dominant attributes not linked among themselves, and daltonism is recessive attribute linked with X-chromosome. The woman — right-handed person with brown eyes and normal sight — marries the man — right-handed, colour-blind, blue-eyed person. At them the blue-eyed, left-handed, colour-blind daughter was born.

What is the chance that their next child will be the lefthander suffering from daltonism?

What color of eyes can sick children have?

79. At the person daltonism (inability to distinguish red and green colors) is caused by recessive gene linked with sex. At the population of mediterranean countries one kind of anemia is common - thalassemia. At other peoples this disease is rare. Thalassemia is inherited as autosomal attribute and is observed in two forms; *thalassemia major* (heavier form, usually fatal for children), and *thalassemia minor*. Let's designate people with thalassemia major with symbols TT , people with thalassemia minor — symbols Tt and healthy people — with symbols tt . The woman — colour-blind person with thalassemia minor — has married the person with normal sight and thalassemia minor.

a) Determine genotypes of these two people.

b) Specify possible phenotypes and genotypes of all children from such marriage, and correlation of these phenotypes and genotypes.

c) To determine, what part of children is threatened with death from thalassemia.

80. Human daltonism is caused by recessive gene linked with sex. Ability to distinguish taste of phenylthiocarbamide is caused by autosomal dominant gene T ; people who doesn't distinguishing taste of this substance have genotype tt .

The woman with normal sight distinguishing taste of phenylthiocarbamide has married the colour-blind person unable to distinguish taste of phenylthiocarbamide. Six children were born (two daughters and four sons). All of them don't suffer daltonism, daughters and two sons distinguish taste of phenylthiocarbamide, but two sons don't distinguish this taste.

Specify probable genotypes of parents and children.

81. Absence of sweat glands is shown as recessive attribute linked with sex. Albinism, i. e. absence of pigmentation, is caused by autosomal recessive gene.

From one married couple, normal to these two attributes, the son with both specified anomalies was born.

- a) Specify probable genotypes of parents.
- b) Their second child was son, too. What is the chance that he will have both anomalies?
- c) Can their third child be a normal girl?

82. A gene of color blindness and a gene of night blindness, inherited through the X-chromosome, are at a distance of 50 centimorgans from each other. Both attributes are recessive.

a) Determine the probability of a birth of children with both anomalies simultaneously in a family, where the wife has normal sight (her mother suffered from night blindness and father from color blindness) and the husband is normal concerning both attributes.

b) Determine the probability of a birth of children with both anomalies simultaneously in a family, where the wife is heterozygous under both attributes and has inherited both anomalies from the father, and the husband has both forms of blindness.

83. Genes of rhesus factor and elliptocytosis are in one chromosome at a distance of 3 centimorgan. Rhesus factor - positive and elliptocytosis are determined by dominant genes. The gene of color blindness and a gene of night blindness are in the X-chromosome at a distance 50 centimorgan. Both attribute are transferred on recessive type.

The rhesus factor-positive woman with a normal form of erythrocytes and with normal sight marries the man negative on rhesus factor with elliptocytosis and suffering from night blindness. It is known, that the father of the wife was negative on rhesus factor and did not distinguish colors, and her mother distinguished colors normally, but suffered from night blindness. Man's father suffered from elliptocytosis only, and mother suffered from night blindness. Determine the probability of a birth of children negative on Rhesus factor and without other anomalies.

Penetrance

84. Angiomatosis of reticulated membrane is inherited as dominant autosomal attribute with penetrance of 50%.

What is the chance that in a family where both parents are heterozygotic on angiomatosis sick children will be born?

85. Craniofacial dysostosis is inherited as autosomal dominant attribute with 50% penetrance.

Find out the percentage of sick children in a family where one of the parents is heterozygous and another is normal concerning analyzed disease.

86. Arachnodactyly is inherited as autosomal dominant attribute with penetrance of 30%. The attribute of left-handers is recessive autosomal and with full penetrance.

Determine the probability of display of both anomalies simultaneously at children in family where both parents are heterozygous on both attributes.

87. Otosclerosis is inherited as dominant autosomal attribute with penetrance of 30%. Hypertrichosis is inherited as an attribute linked with Y-chromosome.

Specify probability of simultaneous display of both anomalies at children in family where wife is normal and homozygous and the husband has both anomalies (but his mother was normal homozygote).

88. Brown color of eyes dominates above blue and is determined by another dominant autosomal gene. Retinoblastoma is determined by another dominant autosomal gene. Penetrance of retinoblastoma is 60%.

a) Can brown eyed children from a marriage between heterozygotic parents be sick?

b) Can brown eyed children from a marriage between heterozygotic parents be healthy?

Genetics of population

89. Common albinism is inherited as recessive autosomal attribute. Its frequency is 1:20000.

Calculate amount of heterozygotes in population.

90. Surdomutism is connected with congenital deafness which interferes with normal mastering of speech. Inheritance is autosomal recessive. For European countries its frequency is approximately 2:10000.

Determine possible number of deaf and dumb people (heterozygous on surdomutism) in the area with 8 000 000 people.

91. Hereditary methemoglobinemia is caused by autosomal recessive gene and is found among eskimoes of Alaska with a frequency of 0,09.

Determine genetic structure of analyzed population on methemoglobinemia.

92. In area with the population of 500 000 thousand people four persons suffering alkaptonuria are registered (autosomal recessive inheritance).

What is the amount of heterozygotes in this population?

The analysis of family trees

93. Newlyweds master the right hand normally. In woman's family there were two sisters owning the right hand, and three brothers — lefthanders. Mother of the woman is the right-handed person, father is the lefthander. The father has sister and brother (lefthanders), sister and two brothers (right-handed person). Father's grandfather is a right-handed person, the grandmother is the lefthander. Woman's mother has two brothers and a sister (all right-handed persons). Mother of the husband is the right-handed person, father is the lefthander. Grandmothers and grandfathers on the part of mother and the father of the husband normally own the right hand.

Determine the probability of a birth in this family children owning left hand.

94. Proband — a healthy woman — has two healthy brothers and two brothers ill with alkaptonuria. Mother of a proband is healthy and has two healthy brothers. Father of a proband is ill with alkaptonuria and is uncle of the wife. He has healthy brother and healthy sister. The grandmother (on a father's line) was ill and was married with the once removed healthy brother. The grandmother and the grandfather of a proband (on a mother's line) are healthy, father and mother of the grandfather are healthy, too, thus mother of the grandfather is the native sister of the grandfather of the proband on father's line. Make a family tree.

95. Proband is a healthy woman. Her sister is healthy, too. But two brothers suffer from daltonism. Mother and father of a proband are healthy. Four sisters of mother a proband are healthy, husbands of them are healthy, too. Facts about cousins: in one family one sick brother, two sisters and the brother are healthy; in two other families there are one sick brother and one healthy sister; in the fourth family — one healthy sister. The grandmother of a proband on the mother's line is healthy, the grandfather suffered from daltonism. On the father's line there were no daltonics.

Determine probability of a birth of children with daltonism, if the proband weds with healthy man.

96. Proband suffers from the light form of sickle-cell anemia. His spouse is healthy. She has the daughter suffering the light form of anemia. Mother and grandmother of a proband suffered from the same form of sickle-cell anemia, the others mother's cousins and her father are healthy. The proband's wife has a sister sick of the easy form of an anemia, the second sister has died of an anemia. Mother and father of the proband's wife suffered from anemia. Except that, the father had two brothers and the sister with the light form of the anemia; in a family of the father's sister two children have died from sickle-cell anemia.

Find out the chance of a birth of children with the heavy form of anemia in a family of the proband's daughter if she will marry the man like her father.

97. Proband and his five brothers are healthy. Mother and father of a proband are deaf mutes. Two uncles and aunt (on father's line) are deaf mutes, too. From the side of the mother four aunts and the uncle are healthy, though one aunt and one uncle are deaf and dumb. Grandmother and grandfather on mother's side are healthy. Grandmother and grandfather on father's side are deaf mutes. The grandmother on father's side has deaf-and-dumb brother and two deaf-and-dumb sisters. The grandfather on father's side has two brothers, one of whom is healthy, and another is a deaf mute. Mother and father of the grandfather on the father's side are healthy, mother and father of the grandmother are deaf mutes. Determine the probability of a birth of deaf-and-dumb children in family of a proband if he marries with healthy woman.

98. Proband (the woman) suffers from ataxy. Her spouse is healthy. Six sons and three daughters were born at them. One son and one daughter are sick with ataxy, other children are healthy. Proband has a healthy sister and three sick brothers. The healthy sister has married for healthy man and has the healthy daughter. Three sick brothers of a proband have married healthy women. In a family of one brother there are 2 healthy sons and one healthy daughter, in a family of the second brother there are healthy son and sick daughter. Father a proband is sick, and mother is healthy.

What is the probability of occurrence of sick children at the sick daughter of a proband if she marries healthy man?

99. Proband suffers from congenital cataract. He is married with the healthy woman and has a sick daughter and healthy son. Father of a proband is ill with cataract, and mother is healthy. Mother of the proband has a healthy sister and healthy parents. Grandfather (under father's side) is sick, and grandmother is healthy. Proband has healthy native aunt and uncle (from the father's side). The uncle is married with the healthy woman. Their three sons (proband's cousins from the side of the father) are healthy.

Can the proband's daughter have sick grandsons, if she marries the man heterozygous on a cataract of this type?

100. Proband suffers defect of nails and patella and his brother is normal. Proband's father had this syndrome and mother was healthy. Proband's grandfather (under father's side) is sick and grandmother is healthy. Father of a proband has three brothers and four sisters, from them two brothers and two sisters have a syndrome. Sick uncle (on a father's side) is married with the healthy woman and has two daughters and a son. All of them are healthy.

Determine the probability of occurrence of children with disease in proband's family if his spouse doesn't suffer from the defect of nails and patella.

101. Proband of normal growth has a sister suffering achondroplasia. Proband's mother is normal and father suffers achondroplasia. Aunt having achondroplasia is married for healthy man. They have son-dwarf. Healthy aunt (from the side of healthy husband) has two boys and two girls (all of them are healthy). The uncle-dwarf is married with healthy woman. Two normal girls and a son-dwarf were born. Grandfather is a dwarf, and grandmother is normal.

a) Find out the probability of occurrence of dwarfs in proband's family if his wife has the same genotype as he has.

б) Specify the probability of occurrence of dwarfs in family of the sister of the proband if she marries a healthy man.

102. One of the forms of rickets does not recover with usual doses of vitamin D. Proband is a young man suffering this form of rickets. His sister is healthy. Mother of a proband is sick, father is healthy. Proband's mother has three brothers who are healthy. The grandfather of the proband (on mother's side) is sick, grandmother is healthy. Grandfather has two healthy brothers and one sick. Sick grandfather's brother has healthy wife. At them three sick daughters and two healthy sons were born. Two sick daughters have one healthy daughter each.

Determine the probability of a birth of children suffering rickets in proband's family if he marries the sick three times removed sister.

103. Epiloiiy is determined by a gene with lethal action. The majority of persons with congenital epiloiiy (pathological growth of skin, intellectual backwardness, convulsive attacks, presence of tumours of heart, kidneys and other bodies) perishes, not having reached a sexual maturity. In case of light expressiveness of a syndrome some persons survive and produce posterity. Proband - the woman suffering epiloiiy — in a marriage with healthy man had three children: a healthy son and a daughter and a sick daughter who later had five children: two healthy sons, two normal daughters and one daughter with epiloiiy. It is established, that this sick woman (proband's daughter had two deadly-born children. What gene(dominant or recessive one) is this disease determined by? How can you explain various effects of this gene?

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